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Hearing and hearing loss management in Children with Down syndrome in Qatar

by

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A Research Dissertation

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Master of Arts

in

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in the Faculty of Humanities, School of Human and Community Development, University of the Witwatersrand, Johannesburg

Supervisors: Dhanashree Pillay, Amisha Kanji

DECLARATION

I, Jade Evrille Govender, hereby declare that this dissertation submitted for the degree Master of Arts in Audiology has been composed entirely by myself with the guidance of my supervisors. It has not previously, neither in part nor in full, been submitted for a degree at any other institution. The work herein is my own, unless stated otherwise and acknowledged and/or referenced accordingly.



Jade E. Govender 23/05/2025

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To Elijah, thanks to you my vision is clearer, and my sense of purpose renewed. May you leave the world better than you found it my precious boy.

ABSTRACT

Background

Untreated hearing impairment affects the speech and language development, academic progression and overall quality of life of children with Down syndrome. Published data regarding otolaryngological conditions and hearing in children with Down syndrome in Qatar is difficult to find, despite the prevalence of the condition here, which may negatively impact planning for their audiological care.

Methods

Using a retrospective record review for patients with DS aged 1 day to 18 years seen between 01 January 2018 to 31 December 2022, this study explored methods of assessment and management utilised by the ENT and Audiology teams at Sidra Medicine, in Doha, Qatar. Non-probability, purposive sampling was employed, with descriptive and correlation statistics used in the analysis of otolaryngological manifestations, audiological profiles, hearing surveillance as well as to determine the relationship between hearing loss management and changes in hearing over time.

Results

A high incidence of otolaryngological conditions was observed in children aged between 0-5.11 years. In the 92 records reviewed, hearing impairment was present in 53 (57.6%) children, with 42 (79.2%) cases of conductive hearing impairment. Surgical interventions for otolaryngological conditions corresponded with better hearing outcomes than medical treatments. Hearing aids were not routinely offered to patients despite persistent hearing impairment.

Conclusion

Children with DS in the region remain at high risk for persistent otolaryngological conditions and hearing impairment. National and institutional levels guidelines to organize and prioritize their ENT and audiological care should be available to clinicians to facilitate timeous care and appropriate solutions for hearing loss management.

Keywords: Down Syndrome, Qatar, hearing loss, guidelines, management.

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LIST OF ABBREVIATIONS

ABBREVIATION	DEFINITION
ABR	Auditory Brainstem Response
CAGS	Centre for Arab Genomic Studies
CDC	Centers for Disease Control and Prevention
ConHI	Conductive hearing impairment
CPG	Clinical practice guideline
CTGA	Catalogue for Transmission Genetics in Arabs
DS	Down syndrome
EMR	Electronic Medical Record
ETD	Eustachian tube dysfunction
HI	Hearing impairment
HL	Hearing loss
IRB	Institutional Review Board
JCIH	Joint Committee on Infant Hearing
LMIC	Low- or middle- income country
MDT	Multidisciplinary team
MHI	Mixed hearing impairment
ME	Middle East
MENA	Middle East and North Africa
OME	Otitis media with effusion
RRR	Retrospective record review
SNHI	Sensorineural hearing impairment

WHO	World Health Organization

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CHAPTER 1: INTRODUCTION

1.1 Background

Hearing impairment (HI) poses profound consequences for speech and language development, communication competence, psychological wellbeing, educational and vocational opportunities, as well as quality of life (McDaid, Park & Chadha, 2021). The annual financial implication of unaddressed HI for the education and healthcare sectors, resultant loss in productivity and increased societal costs, amounts to over US$980 billion globally (WHO, 2021). Over 5% of the global population, including approximately 34 million children are currently affected by HI (WHO, 2023). Of these, 80% reside in low- and middle- income countries (LMIC) with underdeveloped healthcare systems (WHO, 2021). The gap in knowledge of prevalence and causes of HI in LMIC countries, as well as those in which HI is under-studied, impedes the design and implementation of suitable prevention and treatment programs (Ganek et al, 2023).

Hearing impairment (HI), despite its notable prevalence, is understudied in Arab countries (Sidenna, Fadl & Zayed, 2020). Qatar, a country with a predominately Arab population, forms part of the larger Middle East and North Africa region (MENA) from which there is evidence to conclude that genetic and congenital conditions contribute to higher rates of morbidity, mortality, and disability in Arab communities (Al Gazali, Hamamy & Al Arayad, 2006). A scarcity of epidemiological studies in the country impedes adequate healthcare planning and initiation of public health awareness programs in Qatar (Al-Kuwari, 2021). The World Health Organisation (WHO) Regional Office for the Eastern Mediterranean (2013)

identified a notable lack of regional statistics on ear disease and hearing disorders and recommended that scholars undertake population-based epidemiological studies in this area focused on congenital conditions placing children at risk for HI.

One such condition with high prevalence in Qatar is Down syndrome (DS). The incidence of DS in Qatar is amongst the highest worldwide and is postulated to relate to, as in other Arab communities, high rates of advanced maternal age, continued conception till menopause and parental consanguinity (Sijeeni et al, 2016). Notwithstanding the high incidence of DS, there appears to be a paucity of DS epidemiological studies conducted in Qatar. This was confirmed by Wahab et al (2006), who conducted the only epidemiological study of DS in the country, and confirmed a high incidence of the condition, consistent findings from other countries in the region. The lack of available data about DS raises concern as it is the most common genetic anomaly and a leading cause of disability worldwide.

Down syndrome (DS) shows an extensive phenotype with affected individuals frequently requiring lifelong medical as well as rehabilitative interventions (Asim et al, 2015). DS is a recognized risk factor for HI (Porter et al, 2022). The audiological profile of patients with DS, studied by numerous scholars showed high rates of middle ear dysfunction and hearing impairment (HI) of varying severities, of which conductive hearing impairment (ConHI) was the most observed (Earley, Sher and Hill, 2022, Kriecher et al, 2018, Nightengale et al, 2017, Porter et al, 2022, Tedeschi et al, 2015). The high risk for HI and otolaryngological conditions demands effective audiological and Ear Nose and Throat (ENT) interventions in this population. The suspected under-prioritization of hearing in children with DS may have a compounding effect on

the communication difficulties they already face. Population-based research into their audiological profiles and HI management options may provide useful information to facilitate improved audiological and ENT services for the population, as well as serve to substantiate their ongoing audiological care.

The absence of scientifically sound data generated from DS population-based research in Qatar limits knowledge of the clinical needs of children with DS and hinders the improvement of current healthcare practices. Clinical policies and practice guidelines supported by empirical research, including those that inform hearing care provision, would serve to optimize healthcare service delivery in the country. Numerous scholars have recommended the development of region-specific guidelines for effective healthcare surveillance programs including screening, monitoring, diagnosis and treatment of conditions prevalent in at-risk groups, including DS (AAP Health Supervision Guidelines, 2001, Basonbul et al 2020, European Down Syndrome Association, Fidler, Needham & Schworer, 2019, UK Down Syndrome Medical Interest Group).

1.2 Outline and Summary

The need for more research into hearing and HI in Qatar and the greater Middle East (ME) region is substantiated by the high prevalence of HI in Arab communities. The notably high incidence of DS, and its associated risk of HI, in Qatar warrants investigation. The proposed study aims to address the current knowledge gap by providing HL epidemiological information from the vulnerable group of children with DS who have an established risk for hearing loss. Audiological assessment and management practices will be evaluated to determine its efficacy. It is hoped that barriers to the effective and optimized management of children with DS who

present with hearing impairment will be identified, such that recommendations for improvements can be suggested.

CHAPTER TWO: LITERATURE REVIEW

2.1 Introduction

This chapter provides insight into the current literature available globally as well as within the local region regarding HI and pertinent information on DS. It explores the risks for and consequences of otolaryngological conditions and hearing loss in the paediatric DS population, describing the implications for quality of life in affected populations. This chapter also outlines factors related to the management of these conditions aimed at improving hearing outcomes for children with DS. It highlights the current gaps related to the study topic within the local contexts and concluded with a rationale supporting the study objectives.

2.1.1 Focus on Hearing loss

The prevalence of HI has been on the rise since the World Health Organization (WHO) first reported global statistics of sensory loss of hearing in 1985 (WHO, 2023). Recent reports show that by 2050, 900 million of 2.5 billion individuals with HI will present with disabling hearing loss (HL) requiring rehabilitation (WHO, 2023). Approximately 34 million, an estimated 0.5 to 5 children in every 1000 children, are affected by disabling HL globally (WHO, 2021). The WHO (2022) reported that high-income countries report lower prevalence rates than other regions, with approximately 80% of hearing-impaired individuals residing in Low- and Middle-income countries (LMIC). Higher incidence rates in LMIC may be attributed, in part, to systemic issues associated with underdeveloped and resourced constrained health systems resulting in poor antenatal care and inaccessibility to essential testing and treatments.

Weak maternal vaccination and childhood immunization programs, absence of hearing screening programs, use of ototoxic medications to treat neonatal illness as well as higher incidence of risk factors for HI in populations in LMIC correlated with high prevalence of HI in infants (Graydon et al, 2019, Park et al 2012). Graydon et al (2019) reported that 60% of childhood HI in LMIC was preventable and associated with prematurity, low birth weight, congenital or early acquired neonatal infections and birth asphyxia, with 40% having genetic etiologies. Other risk factors for HI outlined in the Joint Committee on Infant Hearing (JCIH) 2007 position statement include family history of HI, congenital infections, craniofacial anomalies, ototoxic medications, low Apgar scores, mechanical ventilation for at least 5 days, and physical findings or the presence of a syndrome associated with congenital hearing loss. Syndromic HI accounts for 30% of genetic hearing losses and is associated with approximately 400 syndromes, one of which is Down syndrome (Sheffield & Smith, 2019, Allen & Goldman, 2023).

2.1.2 Focus on Down syndrome

Down syndrome (DS) or Trisomy 21 is a genetic anomaly resulting from the presence of three instead of two chromosome 21 and is marked by cognitive impairment ranging from mild to severe (Perkins, 2017). First described in 1866, DS remains one of the most frequently occurring chromosomal anomalies in medical history (Ramia et al, 2014, Lehman et al, 2021). Three types of DS, based on the respective chromosome error, namely nondysjunction, translocation and mosaicism have been described. Nondysjunction is the most frequently occurring, with a reported 2-4% incidence of translocation and 1% incidence of mosaicism (Perkins, 2017). The global incidence and prevalence of DS shows notable variability between

populations with an estimated 1 in 400-1500 babies born with DS across the globe (Kazemi et al, 2016).

Table 1

Incidence of Down Syndrome, Europe, USA, selected Arab countries.

COUNTRY/REGION	INCIDENCE	AUTHOR	DATE
Global	1:1000 -1:1100	WHO	2022
Western Europe	0.9:1000	De Graaf et al	2021
Northern Europe	1.1:1000	De Graaf et al	2021
Southern Europe	0.8:1000	De Graaf et al	2021
Eastern Europe	1.1:1000	De Graaf et al	2021
Total Europe	1:1000	De Graaf et al	2021
USA	1.4:1000	CDC	2022
Egypt	1.4:1000	CAGS	1998
Israel	1.8:1000	CAGS	2013
Libya	1.9:1000	CAGS	2013
Kuwait	1.7:1000	CAGS	2013
UAE nationals	2.7:1000	CAGS	2013
Saudi Arabia	1.8:1000	CAGS	2013

Oman	2:1000	CAGS	2013
Qatar	1.95:1000	CAGS	2013

Table 1 displays the most recently published DS incidence data from Europe, USA and selected Arab countries. In the USA, the Centre for Disease Control (CDC) reported that 6000 infants a year were diagnosed with DS. De Graaf et al (2021) published DS live birth prevalence figures for Europe from 2011-2015, with Russia, Germany, and the United Kingdom showing high prevalence. An atypically high incidence of 1.7:1000 was recorded in Ireland, displaying an obvious discordance with other European countries (Caples et al, 2018). The ME region showed a marked heterogeneity of DS incidence compared with Europe and the USA. Specifically in Qatar, the reportedly high live birth incidence of 1.95:1000 was consistent most other Arab countries (Wahab et al, 2006).

It is postulated that higher incidences of genetic disorders, including DS, in Arab populations relate to social, religious, and cultural factors. These factors are said to inform the practice of consanguinity in marriage, advanced maternal age, continued conception until menopause and large family sizes (Wahab, et al, 2006, Al Gazali et al, 2006, Sijeeni, 2016). Conflicting reports refuting the claim that parental consanguinity is associated with DS have been made, and remains a topic in contention (Ray et al, 2018, Rezayat et al, 2013). The availability and utilization of prenatal screening techniques to identify the risk for DS and termination versus continuation of pregnancy following positive screening are also said to influence incidence patterns (Riehle-Colarusso & Oster, 2016). Notably, in addition to

differences in incidence patterns, research suggests that the phenotype of DS also display some variability amongst different ethnic groups (Roper & Reeves, 2006).

2.1.3 Down syndrome phenotype

Extensive phenotypes of varied severity affecting various organ systems is characteristic of DS. Down syndrome is the leading genetic cause of cognitive impairment associated with developmental delay (Silverman, 2007, Wiseman et al, 2009) Clinical manifestations associated with DS include cardiac anomalies, pulmonary disorders, autoimmune disorders, musculoskeletal disorders, otolaryngological conditions as well as hearing loss, neurodevelopmental disorders as well as hematologic and oncologic disorders (Ali, et al 2006; Bull 2020; Roizen et al, 2014; Wiseman et al, 2009). Psychiatric conditions such as anxiety, obsessive compulsive disorder and depression as well as risk for Autism Spectrum disorder is present in the DS population (Vicaro, Pontillo, Armando, 2016) Linguistic impairment in children with DS is typically characterized by poor expressive language development with syntax and phonology most significantly affected (Martin et al, 2009, Buckley, 2002).

2.1.4 Otolaryngological conditions associated with DS

Otolaryngological manifestations in children with DS, resulting from characteristic upper airway and orofacial abnormalities, place them at significant risk of middle ear pathologies (Chin 2014, Nightengale, 2018, Schrijver et al, 2019, Shott et al, 2001). Small pinnae and stenotic external ear canals with impacted wax are common (Chin et al, 2014). Shott (2006) and Maris et al (2014) reported increased risk of Eustachian tube dysfunction (ETD) and otitis media with effusion (OME) in children with DS due to hypoplasia of the mid-face, underdeveloped

Eustachian tube and the crowed nasopharynx. Clark et al (2017) identified that 52% of 38 subjects presented with inner ear anomalies including enlarged vestibular aqueducts, internal auditory meatus hypoplasia and cochlear nerve deficiency. These structural anomalies which increase the risk for HI warrant early identification, surveillance and ongoing management, commencing with neonatal hearing screening.

2.1.5 Hearing impairment in children with DS

Neonatal hearing screening measures, which is the first step in early identification of hearing impairment in infants, frequently yield "refer" results in infants with DS. Yüksel and Uğur (2021) reported that 57.1% of 84 newborns with DS screened for HI in Turkey did not pass the screening test, compared with 20.2% in the well-baby control group. In Singapore, a refer rate of 32.4% and confirmed presence of HI in the first year of life was reported in 34.1% patients with DS (Raut et al, 2011). In the USA, Park et al (2012) reported that 26% of infants with DS failed their neonatal hearing screen, with subsequent diagnostic testing revealing 33 infants had conductive hearing impairment (ConHI), 5 had sensorineural hearing impairment (SNHI) and 3 had mixed hearing impairment (MHI). Tedeschi et al (2015) studied 109 infants with DS in the USA and found 25.7% to have congenital HI, and concluded that HI was more common in children with DS than in those without DS or congenital anomalies. A recent study in the USA showed only 9% of 36% of infants with DS who did not pass their hearing screener presented with normal hearing on follow up testing and emphasized the need for targeted follow up and hearing surveillance to determine if hearing impairment persisted into early years of life (Basonbul et al, 2020).

The persistence of HI in toddlers and children with DS was studied by Shott et al (2001), who reported the presence of HI in 38-78% in a sample of 1088 children with DS, with similar findings also reported by Davies (1996) and Roizen (1997). Raut et al (2011) reported a high incidence of mild to moderate HI in children with DS in the first year of life. A third of those patients continued to exhibit HI after the first year, while a third were lost to follow up. A remarkable prevalence of HI of 36% was also reported in the UK by Nightengale (2018). This finding was supported by a study by Roizen et al (2014) that revealed a HI prevalence of 39% in a sample of 440 children with DS. Transient and fixed ConHI as well as SNHI and MHI were reported (Basonbul, 2020, Kreicher et al, 2018; Roizen et al, 2014; Schrijver et al, 2019).

Table 2

Hearing loss statistics from studies investigating hearing in children with DS

Author/ Date	Sample size	Country	Mean Age/Age Range	Normal Hearing	CHI	SNHI	MHI	Undefined
Austeng et al (2013)	*N=53*	Norway	8 years		16%	18%	5.67%	-
Kreicher et al, (2018)	*N=1088*	USA	7.1years	167	11.4%	2.7%	7.8%	73.4%
Park et al, (2012)	*N=332*	USA	9 months	245	37.9%	5.8%	3.5%	-
Roizen et al (1993)	*N=47*	USA	2 months – 3.5 years	16	19 (ears)	16 (ears)	14 (ears)	-
Nigthengale et al, (2017)	*N=308*	USA	5.9 years			24.9%		

Schrivjer et al, 2019	*N=291*	Belgium	7.4 years	156	90.6%	9.4%	-	-
Lau, Ko & Cheng (2015)	N=50	Hong Kong	11.7	32	72.2%	27.8%	-	-

Table 2 displays data from several studies investigating hearing in children with DS. Whilst typically uncommon, Austeng et al (2013) study of 8-year-old children with DS in Norway was the only study reviewed which reported a higher incidence of SNHI than ConHI. SNHI in this population is postulated to occur due to hypoplasia of the inner ear and cochlear nerve deficiency and is thought to often be progressive (Schrivjer et al, 2019, Kreicher et al, 2018). The overwhelming finding, however, was the high incidence of CHI in this vulnerable group of patients. Persistent otitis media with effusion was the most reported underlying cause of ConHI (Park et al, 2012, Schrivjer et al, 2019).

Conductive hearing impairment (ConHI) associated with frequent, persistent otitis media was reported in 80% of children with DS in the USA (Shott et al, 2001). Kreicher et al (2018) identified 921 subjects with HI in at least one ear resulting from acute/chronic otitis media with effusion (OME), cholesteatoma, and Eustachian tube dysfunction (ETD). Patel, et al (2018), Basonbul, et al (2020) and Shott, et al (2001) all reported similar otologic and HI findings. Barr et al (2011) reported 91% of babies with DS had OME in their first year of life, with a 68% HI prevalence at age 5. Nightengale (2018) found 53% of 1- to 6-year-old children with DS experienced at least one episode of OME in the first year, increasing to 61% in the second year

(Nightengale, 2018). Sensorineural hearing impairment (SNHI) was reportedly less common, ranging between 4% and 20% (Shott, et al, 2006, Nightengale, 2018, Basonbul, 2020).
The impact of hearing impairment on the lives of children with DS, particularly as it relates to speech and language development and communication, must be carefully considered when planning for their ongoing audiological care.

2.1.6 Consequences of untreated HI

Numerous scholars have highlighted the compounding effect of untreated HI on already disordered speech and language development in children with DS (Laws & Hall, 2014, Roberts, Price, Malkin, 2007, Schworer et al, 2022). The language phenotype of children with DS is reportedly variable, affecting phonology, syntax, semantics, as well as pragmatics and results in problems with comprehension and expressive language skills (Stoel-Gammon, 1990, Roberts, Price and Malkin, 2007, Abbeduto et al, 2007, Ching et al, 2010, Kennedy et al, 2006, Moeller, 2000). The speech and language delays have a consequential negative impact in the areas of education, social-emotional development, and social integration (Price and Malkin, 2007, Moeller, 2000). Additionally, the auditory skills requisite for the development of receptive and expressive language are delayed or impaired in children with DS who are hearing impaired, exacerbating existing challenges with language development and impacting their acquisition of an auditory-verbal communication system (Matkin and Wilcox, 1999, Yoshinaga-Itano et al, 2017). The need for effective otolaryngological care and optimal management of HI therefore remains fundamental to ensuring speech and language difficulties are not exacerbated, academic progress is not hindered and the attainment of acceptable quality of life for children with DS is better assured.

2.1.7 Hearing impairment management options

Children with DS are over-represented in surgical procedures including Tympanoplasty, Adenotonsillectomy, Myringoplasty and Myringtomy and Grommet tube insertion as well as Mastoidectomy, frequently with sub-optimal surgical outcomes (Tweedie et al, 2018). Ghadersohi et al (2018) reported that disease recurrence, tympanic membrane (TM) retraction and residual disease following mastoidectomy for cholesteatoma was common. Adenotonsillectomy outcomes were less successful with a higher rate of post-operative complications in patients with DS (Shott and Donelly, 2004, Tweedie et al 2018). Mitchell, Call & Kelly (2003) found that common otological procedures typically for same-day discharge frequently required overnight admission due to postoperative complications in children with DS.

Pressure equalizing tubes (PET) or grommet tubes were less effective in limiting episodes of OME and improving hearing with DS, with higher need for repeat placement (Omar et al, 2021, Khalid-Raja & Tzifa, 2016). Conflicting views on the indication for, viability, efficacy and timing of PET placement were also reported (Sait et al, 2022, Kreicher et al, 2018). Fortnum et al (2014) recommended more research into alternative management options for ETD, OME and temporary HI in children with DS, specifically to address individual requirements and facilitate better hearing outcomes and mitigates its' detrimental effects.

Hearing amplification as an alternate management option for persistent ConHI, as well MHI and SNHI, was recommended (Khalid-Raja & Tzifa, 2016, Hickey, Hickey & Summar,

2012). Concerns regarding external ear stenosis, wax impaction and recurrent OME contraindicating the use of conventional hearing aids in younger children with DS have been raised (Hickey, Hickey & Summar, 2012). Two studies in the UK reported successful implantation and high levels of patient satisfaction with bone anchored hearing aids when the use of conventional hearing aids was contraindicated (Mc Dermott et al, 2008, Sheehan & Hans, 2006). Recent reports established that few children with DS who met the criteria for amplification wore hearing aids, raising questions of availability of devices, compliance with device usage and prioritization of hearing (Porter et al, 2022, Hickey, Hickey & Summar, 2012).

2.1.8 Team based management approach

A collaborative, multidisciplinary team (MDT) approach is recommended for the otolaryngological and audiological care of children with DS (Khalid-Raja & Tzifa, 2016, Bull, 2011). Optimized healthcare quality indicators and maximised potential of the child with DS requires ongoing MDT care as well as audiological interventions and speech therapy to be initiated as early as possible (van den Driessen Maareuw, 2020). Combined Audiology and Speech therapy clinics were suggested for optimized speech therapy outcomes (Laws & Hall, 2014). Adequate hearing surveillance is routinely supported as a foundational component of effective hearing health care in this population. A fundamental component of an effective hearing surveillance program is newborn hearing screening, with targeted follow up for diagnostic assessments as indicated, to ensure early detection and management of HI (Basonbul et al, 2020). Thereafter, surveillance for HI from infancy to adolescence in patients with DS, achieved through regular assessments and timeous follow up care, was recommended by Malt et al (2013), and Lau, Ko & Cheng, (2015). Shott, Joseph and Heitaus (2001) showed that majority

of the HI observed in children with DS was consequential to inadequate hearing surveillance programs. Despite the recommendations, hearing surveillance, or medical monitoring practices in patients with DS remain insufficient, even in places where guidelines exist (Määttä et al, 2011). With the advancement of medical services, the life expectancy for people with DS has increased considerably. To mitigate the impact of HI and ensure enhanced quality of life outcomes for people with DS, continued research and creation of evidence-based clinical care guidelines are required (Bull, 2020, Nigthengale et al, 2020).

2.2 Rationale

2.2.1 Hearing Impairment

Hearing impairment (HI) remains a public health concern affecting millions across the globe. The negative impact of HI on the wellbeing and quality of life of adults is well understood. In children, the effects of late identified and/or untreated HI, impedes access to spoken language, thereby delaying development which consequently affects communication, cognition, education, social integration as well vocational opportunities. The global concern regarding the impact of HI demands an immediate call to action. Chadha, Cieza and Krug (2018) reported however, that despite the concerning predictions, HI remains underprioritized by the public as well as national policy makers. This is particularly relevant to children with special needs who constitute a significant portion of children affected by HI. One such group of children are those with Down syndrome, in whom Whicker et al (2020) reported that hearing loss is particularly underprioritized.

2.2.2 Prevalence of Down syndrome

Down syndrome (DS) is the most frequently occurring genetic anomaly worldwide with a higher prevalence of the condition observed in Arab countries. WHO (2021) reported a global incidence of DS of 1:1000-1100. The Centre for Arab Genomic Studies (CAGS) Catalogue for Transmission Genetics in Arabs (CTGA) database which provides statistics on genetic conditions reported an incidence of DS ranging from 1:319 to 1:546 in Middle East and North Africa (MENA) countries (CAGS, 2013). The database, which includes research findings from the region, indicates a notable lack of genetic studies in the region. An extensive search for published literature investigating the incidence and prevalence of DS in Qatar revealed only one

study conducted more than a decade ago. Al Wahab et al (2006) reported the incidence of DS in non-Qatari residents and Qatari Nationals to be 1:485 and 1:546 live births respectively. This report, although arguably outdated, is consistent with the notion that DS occurs with greater frequency in Arab communities than in Western, non-Arab, populations (Ray et al, 2018, Corder et al, 2017).

2.2.3 Down syndrome a risk factor for hearing impairment

Down syndrome is a recognized risk factor for HI, and while extensively researched in Western countries, little is known of its occurrence and presentation in the ME region (Al Aama, Alem & Al Harouni, 2014, Al Haimor 2013, Corder et al, 2017). Literature suggests that conductive HI (ConHI) is the most frequently occurring type of HI identified in the paediatric DS population (Basonbul et al, 2020, Shott et al, 2001). ConHI is associated with otolaryngological and otologic conditions to which children with DS are predisposed, due to their ear and mid face anatomy (Basonbul et al, 2020, Tedeschi et al, 2015, Shott et al, 2001, Shott, 2006, Chin et al, 2014). SNHI observed in children with DS is linked to structural anomalies of the cochlea (Tedeschi et al, 2015, Nightengale et al, 2017, Kreicher et al, 2018, Shott et al, 2001, Earley, Sher & Hill, 2022). Mixed hearing impairment (MHI) is also common in the DS population, occurring due to a combination of anatomical factors (Tedeschi et al, 2015, Kreicher et al, 2018,). The severity of HI is varied and may be transient and/or progressive in some cases (Nightengale et al, 2017, Kreicher et al, 2018). HL threatens to compound the existing speech and language disorders prevalent in the paediatric Down syndrome population (Laws and Hall, 2014). Furthermore, HI is linked to poor academic performance, behavioural challenges and social isolation which are associated with reduced quality of life. Increased life

expectancy in persons with Down syndrome now necessitates focus on HI to facilitate improve quality of life. It is notable however, that, notwithstanding the high prevalence of HI, hearing care needs of children with DS are often under-prioritized as other, potentially more critical, medical concerns take precedence over hearing. The prioritization of hearing is crucial, and the Audiologist should be responsible for advocating for hearing care for this population. A first step would be to provide services based on sound Audiology guidelines aligned with best practice standards. Thereafter, ongoing caregiver and family support, adequate hearing surveillance and routine HI monitoring and management would aid in encouraging and sustaining hearing care prioritization. Whicker et al (2014) reported a higher hearing and hearing aid prioritization rate in parents who had highly engaged ongoing hearing care professional support. They recommended routine audiological follow up and support for parents and caregivers of children with DS.

2.2.4 Investigating hearing impairment and Down syndrome in Qatar

A search of popular medical databases for published literature about hearing and/or HI in Qatar, returned minimal findings. Girotto et al (2014) investigated hereditary HI associated with consanguinity in the country. Similar studies exploring the relation between consanguinity and HI were conducted by Al Kowari et al, (2010) and Vozzi et al, (2014). Abdul Hadi et al (2012) investigated newborn hearing assessments in the public sector and evaluated the newborn hearing screening program implemented by the state in 2003. More recently, hearing screening practices were evaluated at a private hospital in Qatar, showing that the proposed hearing screening protocol of 2003 was still adhered to (Elsanadiky & Afifi, 2017). Extensive searches of the same databases did not reveal any studies related to otolaryngology, otology or HI in children

with DS in Qatar. The limited availability of clinical data is understandable as Qatar's healthcare system is still developing and researchers may face potential challenges in accessing suitable sample populations as well as in obtaining clearances for research due to institutional, cultural and religious factors. In addition, the significant majority of Qatar's medical staff are expatriate whose transient lifestyles impact their ability to remain in the country committed to long term research studies. Notwithstanding these factors, the potential value of locally sourced healthcare data cannot be denied. Scientifically sound data acquired through population-based research in the local DS community, would translate to actionable knowledge as well as facilitate the improvement of current healthcare practices, particularly for hearing care in this population.

2.2.5 Guidelines for hearing care provision

Extensive empirical research about HL in the DS community has provided information which has been useful for planning for the provision of Audiology services for this group of patients outside the ME region (Bull, 2011, Santoro, 2018, Tsou et al, 2020). Further to this, ongoing research in the area has served to evaluate these services and make recommendations for improvements. Such research is limited in the ME region (Alanazi et al, 2024). Many researchers, as well as international DS Associations suggest that the formulation and implementation of country or region-specific practice guidelines, have ensured sustained and successful provision of evidence-based healthcare services (Basonbul et al 2020, Fidler, Needham & Schworer, 2019, European Down Syndrome Association, UK Down Syndrome Medical Interest Group, AAP Health Supervision Guidelines, 2001). In the USA the AAP Health Supervisory guidelines have been used to inform the clinical care offered by primary physicians caring for children with DS as well as to ensure that all healthcare service providers instrumental

in the management of the child with DS are timeously engaged. These guidelines as detailed by Bull (2011, 2022) described the different assessments and interventions which should be provided for children diagnosed with DS based on all identified risk factors including hearing loss from hearing screening at birth, referrals for diagnostic hearing testing as required in infancy, periodic hearing testing in childhood (early and late) as well as continued monitoring in adolescence until early adulthood. In the United Kingdom, assessment guidelines for several conditions, including hearing loss, have been proposed by the Down Syndrome Medical Interest Group. The groups hearing care guideline recommending newborn hearing screening, DS showed high compliance rates (Mengoni & Redman, 2020). Guidelines for audiological surveillance in the DS population as previously mentioned are depicted below (see Figure 1, Figure 2 & Figure 3).

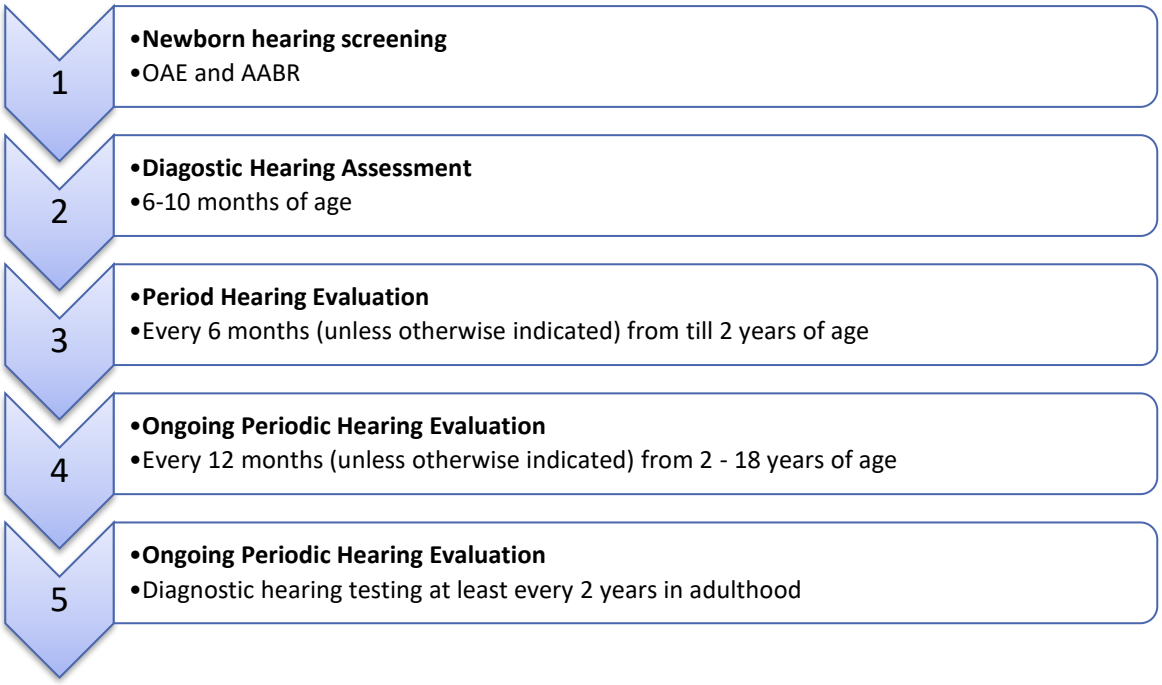
2.2.6 Global Audiological Surveillance Guidelines for people with Down syndrome

2.2.6.1 UNITED KINGDOM

The Down Syndrome Medical Interest Group (DSMIG) proposed healthcare surveillance guidelines including those for audiological care for use throughout the United Kingdom. The group advocated for audiological surveillance citing the risk for the hearing loss and the impact of HI on speech and language outcomes in the Down syndrome population. They recommended a multidisciplinary team approach which included audiologists, speech and language therapist, ENT and Paediatricians and highlighted the importance of considering developmental capabilities and possible barriers to testing (eg. External ear stenosis) when dealing with this special population

Figure *1*

Breakdown of DSMIG Audiological Surveillance Guidelines

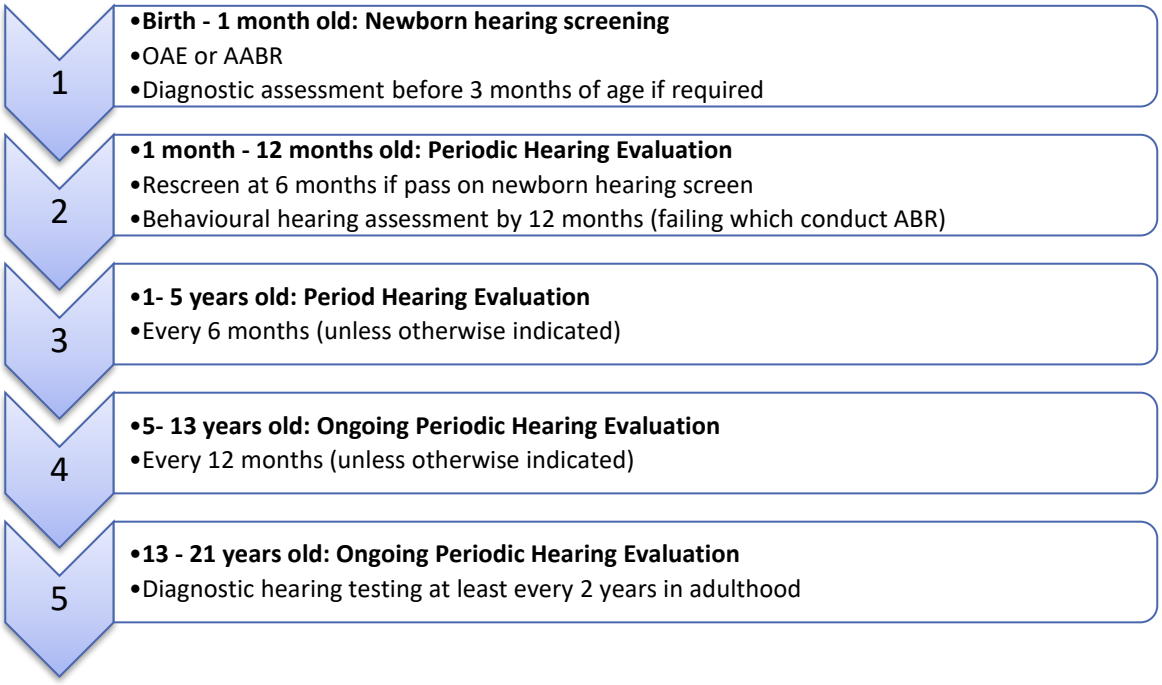


2.2.6.2 UNITES STATES OF AMERICA

The American Academy of Pediatrics (AAP) provided healthcare guidelines for paediatricians and families of people with DS. Additionally, they provide anticipatory guidelines to support families in addressing the complex conditions affecting a child with Down syndrome.

Figure 2

Breakdown of AAP Audiological Surveillance Guidelines

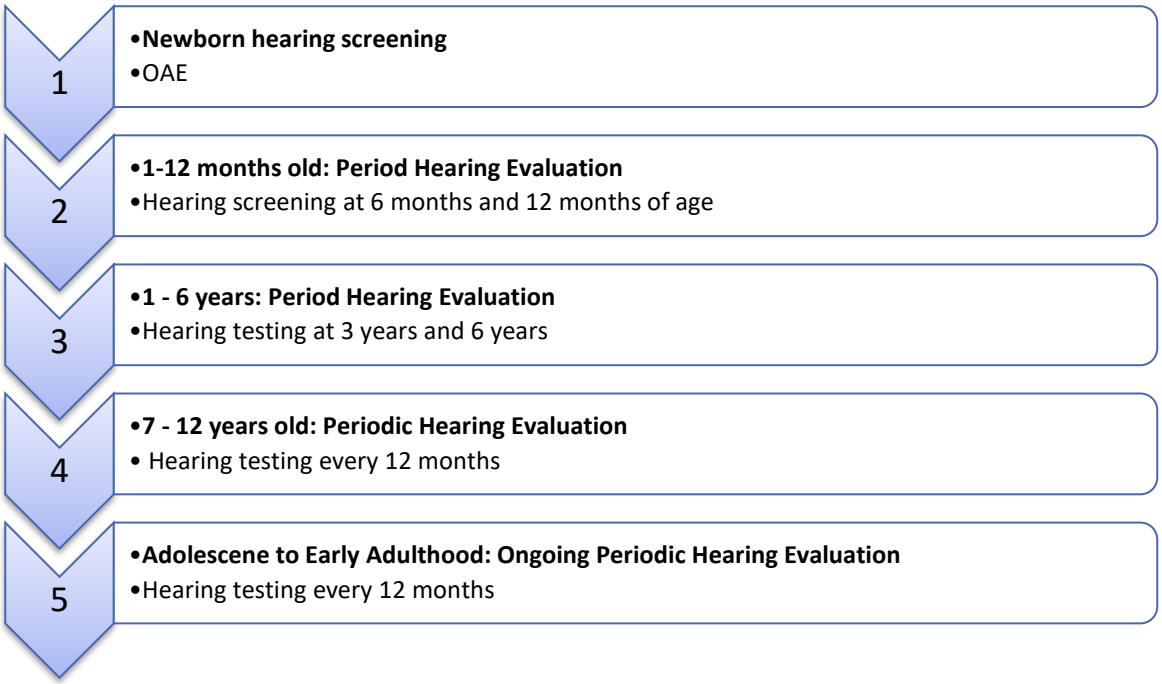


2.2.6.3 EUROPE

The European Down Syndrome Association (EDSA) provided recommendations for the care of children with Down syndrome aimed at paediatricians and general practitioners. The guiding document is aimed at improving access to required services and optimizing rehabilitation efforts.

Figure 3

Breakdown of EDSA Audiological Surveillance Guidelines



The development of clinical practice guidelines, however, presents inherent challenges including its requirement to be diverse and widely applicable in various healthcare contexts. Importantly, guidelines are not intended to dictate the course of medical treatment, nor are they meant to be viewed as gold standards for care. Instead, they should be used to support clinicians in provision of services and in creation of effective healthcare surveillance programs including screening, monitoring, diagnosis and treatment of conditions prevalent in at risk groups. Country or region-specific guidelines may better ensure that practical factors including culture and tradition, the specific demands and requirements of the local population, as well as resource availability/constraints are duly considered.

2.2.7 Availability of Clinical Guidelines in Qatar

An online search for published medical practice guidelines yielded limited results in the ME. Almazrou et al (2021) also searched clinical practice guidelines (CPG) in the MENA region, to review and assess the CPG quality. They identified 61 published documents, most of which were from Saudi Arabia. None of these were from Qatar. A search of the Qatari Ministry of Public Health website revealed 51 Clinical Guidelines for the assessment and management of various medical conditions. A refined search of the Ministry of Public Health online resources did not yield any results for the guideline documents for the assessment, management and surveillance of hearing and hearing loss in children with DS in Qatar. In 2013, the WHO Regional Office for the Eastern Mediterranean after identifying a notable lack of regional statistics on ear disease and hearing disorders, recommended scholars undertake population-based epidemiological studies in this area. They further recommended clinicians "Develop minimum service delivery standards for primary ear and hearing health, secondary level care and tertiary services, which are comprehensive and inclusive to the extent possible" (WHO Regional Office for the Eastern Mediterranean, 2013). Almost a decade later, there is no published literature on hearing loss in the Down syndrome community in Qatar. In the absence of practice guidelines, Audiological services for children with DS across Qatar remain unstandardized and disjointed, rooted in rudimentary clinic-based protocols which differ between institutions and clinicians. Sidra Medicine currently lacks definitive guidelines, as well as valuable inter-disciplinary collaboration, for the assessment and management of HI in children with DS. This raises concerns about the whether the Audiology service is in fact meeting best practice recommendations. Unfortunately, the apparent absence of locally acquired data will continue to limit policy maker opportunities to create policies and guidelines for the standardization of audiological care as well as recommendations for audiological surveillance in children with DS.

Qatar has a healthcare system that has potential to develop rapidly due to the country's considerable wealth. The system currently offers expert healthcare services and modern medical equipment, which has led to increased accessibility and specialization of healthcare services, especially for children with complex medical conditions. The shortage of population-based healthcare data is apparent. Specifically, the lack of population healthcare data and limited clinical research on HI in children with DS in the country is a concern, as the risk for HI in children with DS is high. There is a need, however, to establish clinical guidelines and protocols to standardize care and optimize the services provided. This study is the first of its kind in Qatar and the larger Gulf area, focusing on hearing and HI management in children with DS. This study sought to provide valuable information regarding the hearing in children with DS in Qatar.

CHAPTER 3: METHODOLOGY

3.1. Research Question

What are the audiological assessment practices, management strategies and surveillance practices employed for children with Down syndrome, who are at risk for otolaryngological manifestations and hearing impairment at Sidra Medicine in Doha, Qatar?

3.2. Aim

To explore audiological assessment and management practices for children with Down syndrome at Sidra Medicine, Doha.

3.3. Objectives

1) To determine the prevalence of otolaryngological manifestations in children with Down syndrome at Sidra Medicine, Doha.

2) To describe the audiological profiles of children with Down syndrome at Sidra Medicine, Doha.

3) To describe hearing surveillance practices undertaken for children with Down Syndrome at Sidra Medicine, Doha.

4) To describe the audiological management practices for children with Down Syndrome at Sidra Medicine, Doha.

5) To determine the relationship between management strategies and audiological findings over time in children with Down syndrome at Sidra Medicine, Doha.

3.4. Study Type

3.4.1 Study Design

This study utilized a retrospective cohort design. The cohort comprised of patients with a confirmed diagnosis of DS aged between 1 day and 18 years. A comprehensive retrospective review of their medical records over a four-year period commencing 01 January 2018 to 31 December 2022 was undertaken. Audiological and otolaryngological data for the cohort was analysed to determine their audiological profiles and determine their risk for hearing loss and otolaryngological conditions over the specified time frame. The study explored strategies used by the ENT and Audiologists to manage hearing loss. Setia (2016) supported the use of retrospective cohort designs in research, highlighting that it was quicker to complete than prospective studies and was of relatively low cost. This specific study design was selected based on its advantages of feasibility, cost effectiveness and time efficiency and proved a suitable choice to meet the study objectives.

3.4.2. Sampling technique

The proper selection of data is fundamental to a sound theoretical framework and cannot be compensated for, even by the most detailed analysis techniques. Andrade (2020) stated that non-probability purposive sampling did not require prescribed participant numbers nor underlying theories and involved the deliberate selection of participants, based shared quality or characteristic, from whom to retrieve data. Non-probability purposive sampling was therefore employed for the study as records for all patients with a diagnosis of Down syndrome meeting the stipulated inclusion criteria were included in the study.

3.4.3. Study Population

Medical records obtained from 92 children with a confirmed diagnosis of Down syndrome aged between 1 day to 18 years, who have accessed Ear Nose and Throat (ENT) and Audiological services at Sidra Medicine were included in the study, provided they met the specified inclusion criteria. The children must have received ENT and Audiology services in the period 01 January 2018 to 31 December 2022.

3.4.3.1 Inclusion Criteria

Patient medical records displaying at least one consultation with the ENT and Audiology Departments, with detailed documentation for investigations performed by ENT and results for all essential Audiological test procedures i.e., Otoscopy, Tympanometry, Audiometry and/or Auditory Brainstem Response (ABR) between the period 01 January 2018 to 31 December 2022 were included in the study. The specified period was selected as the Audiology department was fully operational with an adequate staff complement from January 2018. The protocols used in 2018 remained in use until the end of the study period assuring consistent and standardized Audiological testing which consequently enhanced the reliability of data obtained for the study. Additionally, the selected medical records included a plan or recommendations for Audiology and ENT follow-up care as determined by the attending clinician or physician. Frequency of follow-up to audiology appointments for patients presenting with normal hearing was included to determine current time frames for hearing loss surveillance.

3.4.3.2 Exclusion Criteria

Participant records with entries dated prior to January 2018 were excluded from the study. Participant records without test findings for Otoscopy, Tympanometry, Audiometry/ABR (except in cases where this was not medically possible e.g. microtia and atresia of the outer ear), had the potential to significantly degrade the value adding capabilities of the data and were therefore excluded.

3.4.4. Study Setting

The study was undertaken at the Audiology Department of Sidra Medicine, a private hospital for public benefit in Doha, Qatar. The Department is part of the larger outpatient clinic at Sidra Medicine and offers Audiological services to patients from 1 day - 18 years of age referred from medical facilities around Qatar as well as from other clinical departments within Sidra Medicine, most frequently collaborating with the Ear Nose and Throat Department for patient care. Four Audiologists and one Audiology technician provide objective and subjective Audiology assessment services as well as Audiology management services including hearing aid fitting to patients. Prior to the study, no Departmental or organizational guidelines for the hearing care for children with Down syndrome existed at the organisation, which served in part as the motivation to conduct the study. At the time of the study, patients were followed up based on their presenting audiological symptoms as well as on the recommendations of ENT. Hearing surveillance was rudimentary, decided upon by the needs of the patient and the requests of caregivers in comparison to evidence based surveillance programs. Decisions regarding initial assessments, targeted follow up and hearing loss management strategies were supported by clinical judgement and/or case discussion and peer review in situations where a patient's

presentation was more complex resulting in a lack of uniformity in the management of children with Down syndrome.

3.4.5 Data Management

3.4.5.1 Data Extraction and Collection Tool

A data abstraction sheet was designed by the primary investigator to capture the requisite information for the study (see Appendix A 1-3). A brief pre-testing of the data collection tool was conducted during which sample data was collected and the suitability of the tool evaluated. This process highlighted the need for slight refinement of the data collection tool to reduce ambiguity and optimize data collection. Thereafter, the refined the tool was used for the collation of requisite data essential to answer the research question. The tool captured salient information obtained during Audiology and ENT clinic visits including demographic information, medical information and audiological information. Demographic data comprised of the gender, age, nationality, and ethnicity. It also included the maternal age at birth of the study subject, as well as whether there was parental consanguinity in the family. Medical data captured by the tool included otologic and otolaryngological findings i.e. otitis media, Eustachian tube dysfunction, cholesteatoma, chronic rhinitis, adenoid hypertrophy, and obstructive sleep apnea. Audiological data including otoscopy, immittance measures and audiometric findings were also captured. Time-frames between appointments as well as missed follow up and no shows were also recorded. Frequency of attendance of audiology appointments will be recorded. In cases where multiple audiology sessions were completed, results from all visits were included in the study to determine if any changes in Audiological findings were evident following repeated clinic visits and various treatment methodologies. Audiometric data was collected and synthesized to

determine the type of [1]hearing impairment as well as its' severity. Auditory brainstem response findings were available were included. Management strategies employed by ENT as well as the Audiologist were included.

3.4.5.2. Data Collection Process

Permission to access and extract data from Cerner PowerChart was granted by Sidra Medicine Institutional Review Board (IRB) on 04 April 2023. Cerner PowerChart is an Electronic Medical Record (EMR) system used for the electronic documentation and review of medical information entered by authorised medical personnel responsible for clinical service provision at Sidra Medicine (see Appendix B 1). The use of the EMR was guided by the organisations Medical Records Documentation policy which outlined the process for documentation, content requirements and timeframes as well as the management of medical records throughout the organisation. The use of EMR systems is widely supported for its transformational benefits in healthcare and is said to correlate with enhancing quality of care, improved efficiency in patient interactions and reduction medical errors (Belletti, Zacker & Mullins, 2010, Jawhari et al, 2016)

During the period 1 January 2024 to 4 February 2024, data was extracted through a comprehensive retrospective record review (RRR). RRR is a popular, economical research methodology, with the benefit of time efficiency through its utilisation of secondary data and the potential of providing preliminary data to inform future research (Gearing et al, 2006). Limitations of RRR relate to the quality of the records documented, which when inaccurate or missing important information, can result in misleading findings (Hart & Yoost, 2025). The

[1] Undefined HI refers to HL which cannot be classified as SNHL/MHL/ConHL owing to absence of BC data.

current study utilized strict inclusion and exclusion criteria which ensured the records selected were as accurate as possible thereby assuring the suitability of records and addressing the inherent limitations of RRR.

During the RRR, medical records with mandatory diagnosis coding on the system corresponding with Down syndrome (Q90.0 Trisomy 21 non-mosaicism (meiotic nondisjunction), Q90.1 Trisomy 21 mosaicism (miotic nondisjunction), Q90.2 Trisomy 21, translocation, Q90.9 Down syndrome, unspecified) were selected and preliminarily reviewed. At this stage, records not meeting the stipulated inclusion criteria were excluded from the study. The qualifying data which met the inclusion criteria was entered into the data collection tool and organized accordingly.

3.4.5.3. Data Analysis

IBM SPSS Statistics (29.0.2.0) was used for data analysis. Descriptive statistical analysis was used to determine the frequency, proportions, means and medians of otologic conditions, hearing and hearing loss as well as hearing loss management strategies employed by both the Audiologist and the ENT physician for children with Down syndrome.

Descriptive statistical analysis was used to determine hearing surveillance practices including frequency of follow up appointments, missed follow up appointments, mean wait times between Audiology and ENT follow up appointments as well as the average time frame during which patients presented with hearing loss.

Chi squared test was used to test for a relationship between otolaryngological conditions and hearing loss.

Spearman's rho correlation analysis was utilized to explore the relationship between hearing loss management strategies and changes in hearing thresholds over time. The variables included watchful wait, medical management, surgical management, and hearing aid fitting.

3.4.5.4. Data Security

A data management plan (DMP) was created to provide a clear guideline for the safe and ethical management of the data being handled. Data was consistently maintained in a manner intended to preserve its integrity and accuracy. The data collection tool was stored only in the personal drive of the primary researcher which is inaccessible to third parties without knowledge of the password. To further increase security, the data collection tool itself was password protected, and the collection, organization, storage, and analysis of the data was be managed by the primary researcher only. Access restrictions and controlled data retrievability ensured that data was handled by the primary investigator only and safeguarded the confidentially of records used in the study. The data was used for the purpose of a Master study and as such de-identified, data pre-and post-analysis was, on occasion, made accessible to research supervisors for the purpose of receiving guidance during the drafting of the results section and completion of the final research dissertation.

3.4.5. Reliability and Validity

Retrospective record review is designed to utilize previously collected medical records to extract relevant data. One inherent limitation of this type of research design threatening the reliability and validity of findings is the possibility of missing information, incoherent or absent session notes, lack of systematic recording of documentation and missing information on variables

relevant to the proposed study. To account for this, a standardized data extraction tool was created specifically for the study and was the sole instrument used in the data extraction and collection process. The tool accounted for all variables relevant to the study ensuring content validity. The record review process was carefully planned and the data extraction and collection tool tested and refined prior to its utilization in the final data extraction and collection process. Data extraction was completed by the primary investigation eliminating the need for testing for inter-rater reliability. To further confirm reliability and the content validity of the data collection tool, 9 participant records were selected at random for data collection and repeat review. No differences in the data extracted identified on the secondary collection. The test result data collected from the participant records was cross referenced against test procedure graphs, tracings and audiograms to ensure their accuracy. The aforementioned served to improve the validity and reliability in the study.

3.4.6. Ethical Considerations

3.4.6.1 Anonymity of Patient Data

All data utilized in the study was stripped of any possible identifier at the initial stages of the retrospective record review, thereby rendering data fully anonymized. Junod and Elger (2010) described two types of anonymizations; reversible and irreversible anonymization and concluded that irreversible anonymization ensured extreme difficulty in uncovering patient identity post data collection. As such, irreversible anonymization was utilized in the study to ensure that confidentiality of the subject data. To anonymize the data obtained, direct and indirect patient identifiers routinely available in the medical record software including names, hospital

identification numbers and telephone contact details were excluded from the database and replaced by alphanumeric codes.

3.4.6.2 Ethical Clearance

A research proposal was submitted to the Institutional Review Board (IRB) at Sidra Medicine for approval to collect data for the study from existing hospital records as is the institutions' requirement when accessing patient related information. In accordance with U.S. Department of Health and Human Services (HHS) 45 CFR 46.117 (Protection of Human Subjects 2018) which guided research at the study site, the study posed no more than minimal risk to participants therefore a waiver of written informed consent was requested. The Council for International Organizations of Medical Sciences (CIOMS) also identified studies involving no more than minimal risk to subjects as being eligible for a waiver of written informed consent subject to the approval of an Ethics committee or Institutional Review Board.

Furthermore, retrospective chart review involved the utilization of secondary data originally obtained during routine clinical procedures to which parents or legal guardians provided written general consent on initial hospital visits. The general consent document informed parents or caregivers of the possible disclosure/use of protected, anonymous de-identified healthcare information (see Appendix C1- C3 for General Consent form).

Approval to conduct the study with a waiver of written informed consent was granted by the board on 04 April 2023 (see Appendix D for IBR approval letter).

Ethical approval was sought from the Medical Ethics Review committee at the University of Witwatersrand, Johannesburg as is the requirement when conducting research involving human subjects. Unconditional approval was obtained on 12 January 2024 (see Appendix E). Despite obtaining approval to conduct the study without obtaining informed consent, the principles of research as set out in the Declaration of Helsinki were adhered to, ensuring ethical optimization when handling sensitive health related data

CHAPTER 4: RESULTS

4.1. Introduction

The aim of the study was *to describe the audiological assessment practices and management strategies employed for children with Down syndrome (DS), who are at risk for otolaryngological manifestations and hearing impairment in Doha, Qatar.* The results of the study are presented as per the specific objectives outlined in Chapter 3.

4.2. Selection of participant records for inclusion in the study

Figure 4

Medical record selection process for patients with Down syndrome seen in the ENT and Audiology departments from first January 2018 to 31 December 2022

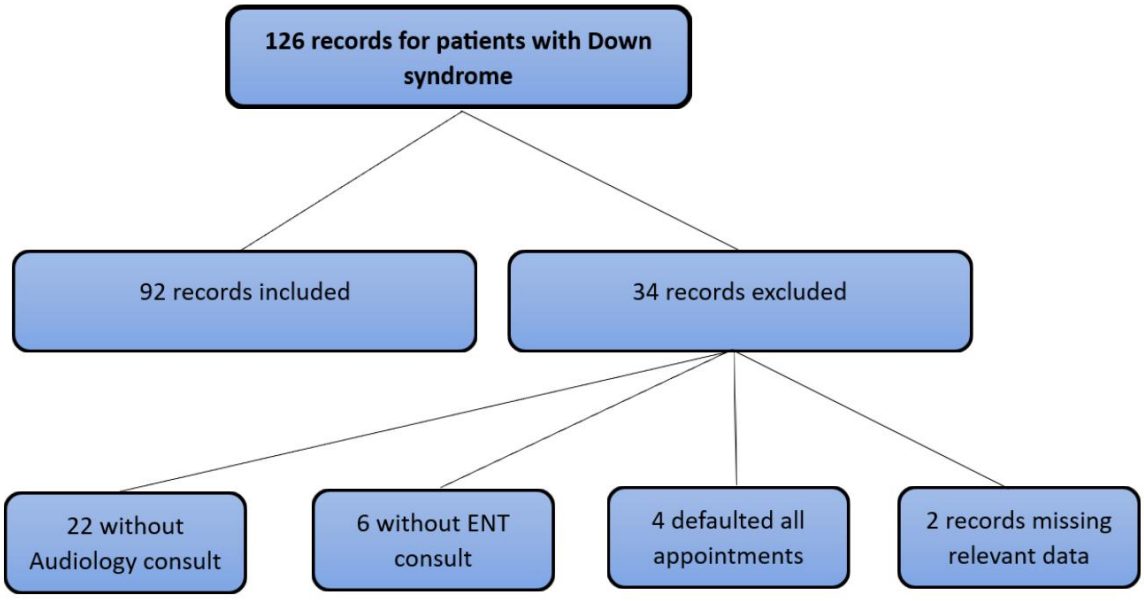


Figure 4 shows that a total of 126 participant records with the relevant ICD 10 codes corresponding to DS (Q90.0 Trisomy 21 non-mosaicism (meiotic nondisjunction), Q90.1

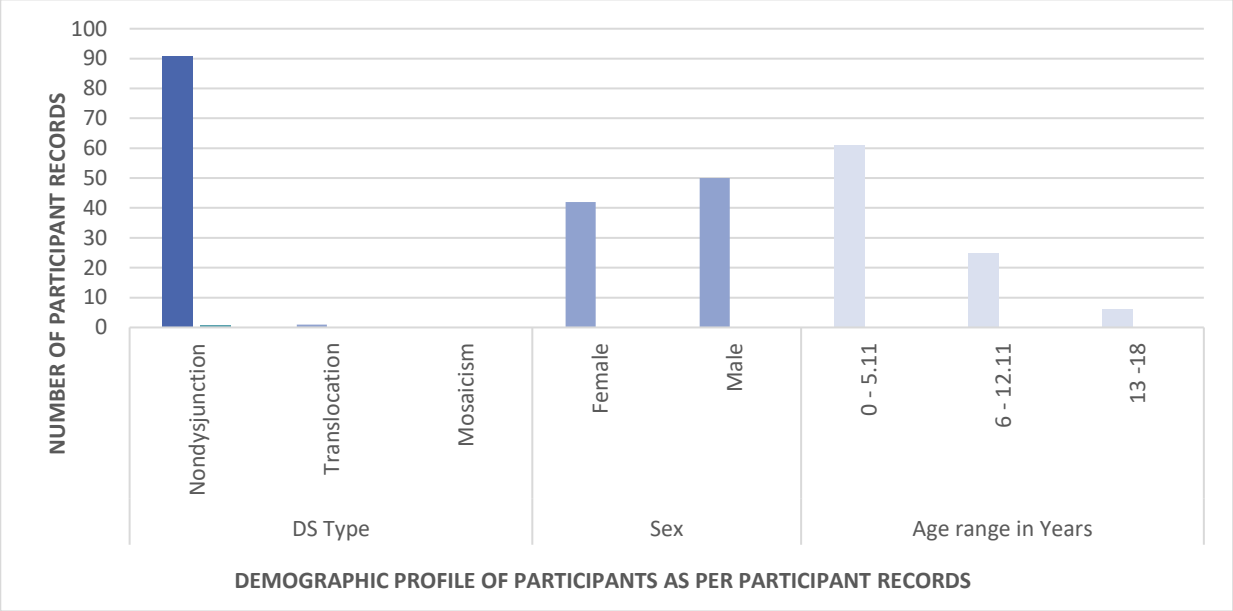
Trisomy 21 mosaicism (mitotic nondisjunction), Q90.2 Trisomy 21, translocation, Q90.9 Down syndrome, unspecified) were documented at the Audiology and/or ENT departments. Of these 126 participant records, 34 (26%.) records not meeting the criteria for inclusion were excluded. From the 34 excluded participant records, 22 (64.71%) included a consultation with ENT but no Audiology consultation, six (17.65%) included a consultation with Audiology but no ENT consultation, four (11.76%) reflected that the patient did not attend any scheduled appointments at both departments and two (5.88%) records contained incomplete clinical documentation. The remaining 92 records were included in the study.

4.3. Demographic profile as per participant records

The demographic information detailed below includes the type of DS, the gender profile and age range of participant records included in the study.

Figure 5

Demographic profile of participant records with Down syndrome (N = 92) seen in the ENT and Audiology departments from first January 2018 to 31 December 2022



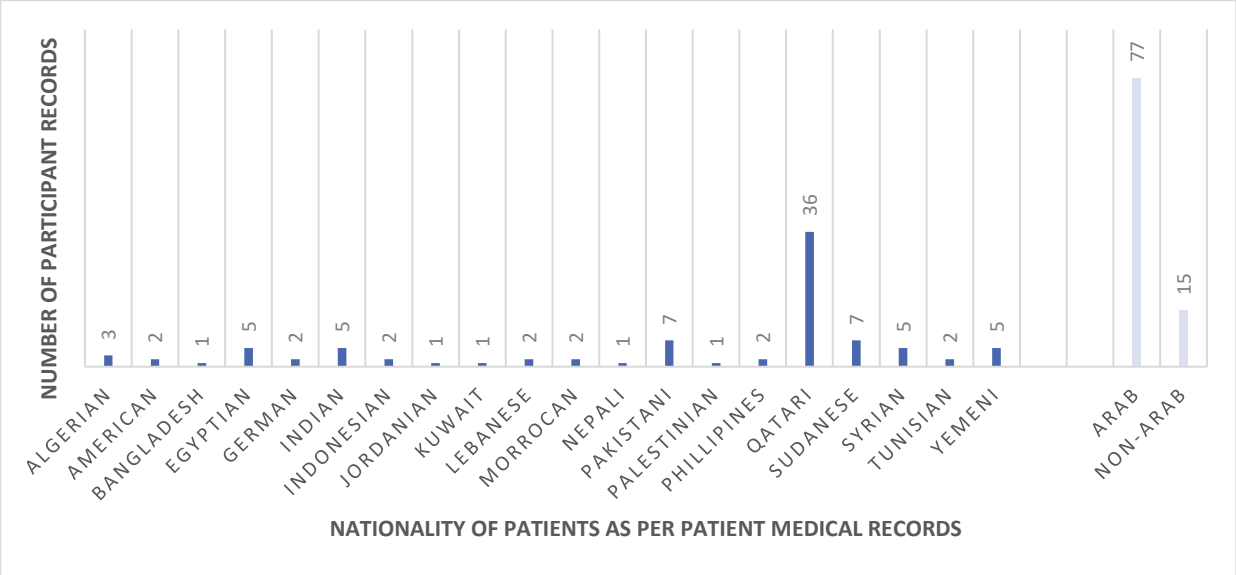
As displayed in Figure 5, 91 (98.9%) participant records reflected a diagnosis Nondisjunction DS and one (1.1%) of Translocation DS. The study included records belonging to 42 (45.7%) female and 50 (54.3%) male patients, aged between eight months and 15.9 years, with a mean age of 5.3 years at the initial audiology visit. A majority of patients (n=59, 64.1%) were between 0 – 5,11 years, 27 (29.3%) between 6 – 12,11 years and six (6,6%) were older than 13 years (*m = 0.4, sd = 0.61*).

Additional demographic information related to the participant records was analysed, to enhance the understanding of the participant group. The mean age of patients' mothers in the study was 35.9 years (sd = 6.6), with the youngest recorded age of 22 years and the oldest

recorded age of 52 years. Parental consanguinity, as reported by primary caregivers during initial case history interviews, was present in 48 (52.2%), absent in 14 (15.2%) and undetermined in 30 (32.6%) participant records.

Figure 6

Nationalities and ethnicity of patients with Down syndrome (N = 92) seen in the ENT and Audiology departments from first January 2018 to 31 December 2022



As shown in Figure 6, participant records reflected that 36 (39,1%) patients were Qatari nationals with the remaining 56 (60.9%) having 19 different nationalities. Those patients of different nationalities were permanent residents of the country during the study period and were therefore able to access healthcare services at the institution by supplying their national identification cards which confirmed their resident status. The study comprised of 11 (11,9%) patients from Asia, 77 (83.7%) from Middle East and North Africa (MENA), two (2.2%) from

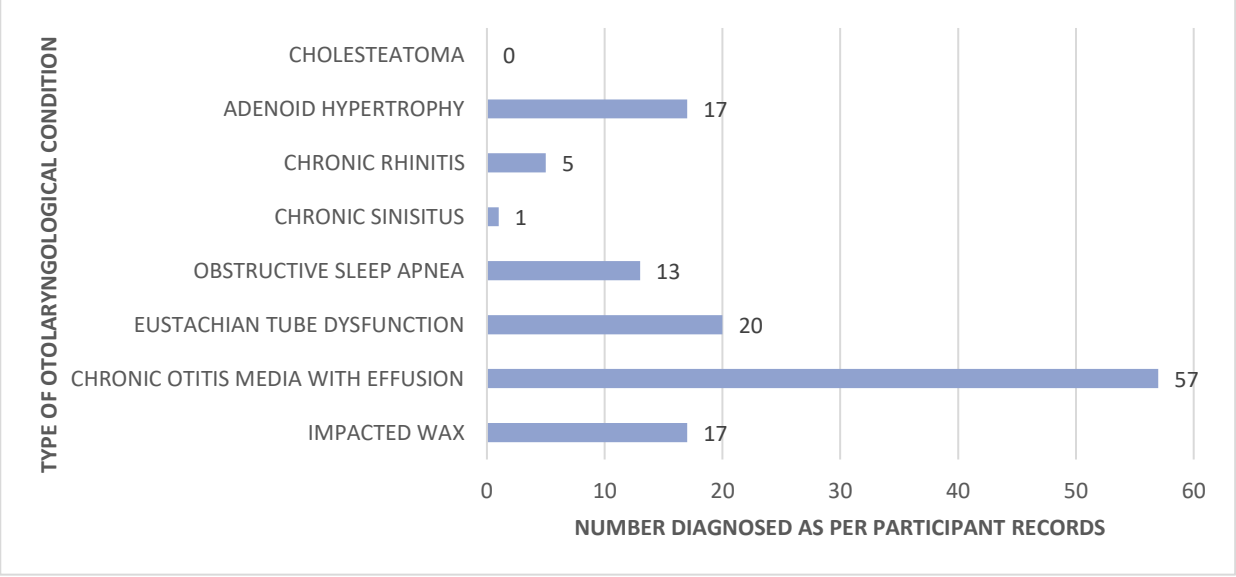
Europe and two (2.2%) from North America. Records indicated that 77 (83.7%) patients were of Arab descent and 15 (16,3%) were non-Arab.

4.4 Frequency of otolaryngological conditions

The first objective of the study was *to determine the frequency of otolaryngological conditions observed in children with Down syndrome at Sidra Medicine, Doha* during the period 01 January 2018 to 31 December 2022. An initial diagnosis of one or more of the frequently occurring otolaryngological conditions in children with DS was documented in 77 (83.7%) participant records ($m = 0.16$, $sd = 0.371$). The remaining 15 (16.3%) did not present with any otolaryngological condition.

Figure 7

Otolaryngological conditions in records of patients with Down syndrome (n = 77)



The most frequently occurring condition was chronic otitis media with effusion (OME) reported in at least one ear in 57 (62.0%) of the participant records. Other conditions commonly diagnosed were Eustachian tube dysfunction (ETD) in 20 (21.7%), adenoidal hypertrophy in 17 (18.5%) and impacted wax in 17 (18.5%). Other less frequently occurring conditions, were also observed in the sample (see Figure 7).

Of the 77 patients presenting with one or more otolaryngological condition on initial assessment, 42 (54.5%) presented with one otolaryngological condition (m=1.41, sd=1.007). Two or more co-occurring otolaryngological conditions affecting at least one ear (where applicable) was documented in 35 (45.5%) participant records. Further analysis revealed that while 15 participant records did not include an otolaryngological diagnosis at the initial assessment, seven (46.7%) of these reflected a diagnosis of one or more otolaryngological conditions on subsequent visits while six (40%) records reflected no new otolaryngological findings. Two patients (13.3%) did not have subsequent visits. Hearing within normal limits was reported in 24 (31.2%) patients with otolaryngological conditions. Hearing impairment (HI) of varying degrees of severity was reported in five (71.4%) patients who presented with otolaryngological conditions at follow- up visits (and who did not have any of these conditions on the initial visit).

Figure 8

Occurrence of otolaryngology conditions in three different age categories (n=77)

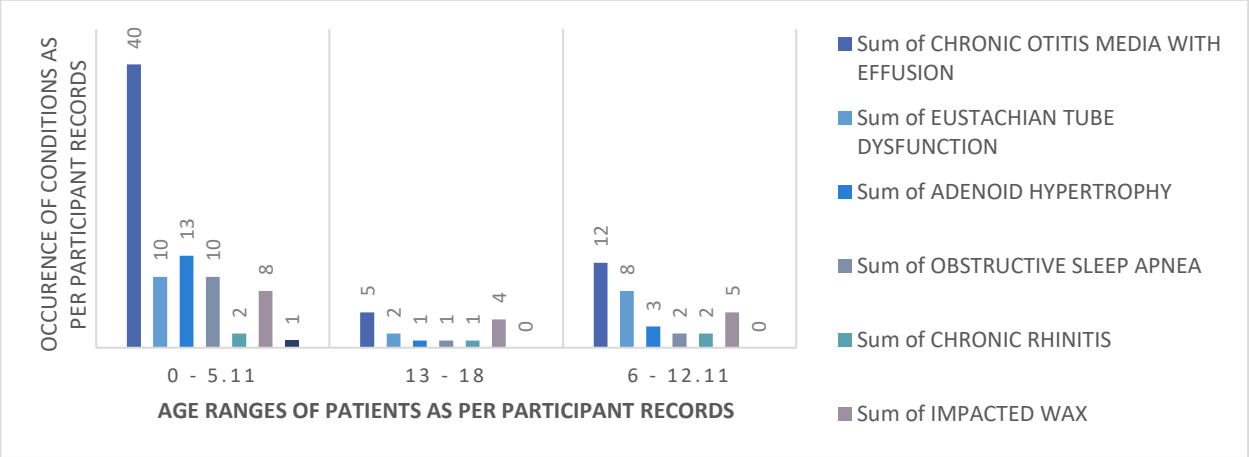


Figure 8 shows the distribution of otolaryngological conditions in the three different age categories. All conditions except chronic rhinitis were most prevalent in the 0 - 5.11 age group. Participant records of patients under the age of six years showed a higher incidence of otitis media with effusion, Eustachian tube dysfunction, adenoidal hypertrophy and obstructive sleep apnea. Otolaryngological conditions occurred in 14 patients older than 13 years of age, notably less than in the 0 - 5.11-year age group. Overall, the study showed that 77 patients had otolaryngological conditions, the most common being chronic OME, which was present in 57 patients, 40 of whom were aged between 0 - 5.11 years. Otitis media with effusion persisted to a lesser extent as children aged, with 5 patients aged 13 years or older presenting with OME.

4.5 Audiological profiles and their relationship with otolaryngological conditions

The second objective of the study was to *describe the audiological profiles of children with DS at Sidra Medicine, Doha.* The audiological profile of 92 patients with DS was determined by

the final diagnosis as concluded based on the outcome of the audiological assessment procedures used to evaluate each patient.

Small pinnae and narrow ear canals were observed in 85 participant records. No cases of microtia or complete stenosis of ear canals were observed. Participant records reflected tympanometric findings of a type B tympanogram in at least one ear over two consecutive clinic appointments in 57 (62.0%), indicative of chronic otitis media. A type C tympanogram was recorded in at least one ear in 20 (21.7%) participant records indicative of Eustachian tube dysfunction. Flat tympanograms with a small ear canal volume (documented as Type B tympanograms with ear canal volume $\leq$ 0.2ml) were recorded in at least one ear in 17 (18.5%) participant records, and when cross-referenced with findings from otoscopic examination, confirmed impacted wax.

Table 3

Descriptions of hearing impairment presentations in patients with DS (n = 53)

Variable	Number (n)	Mean (m)	Standard Deviation (sd)	Variance (v)
HI		1.42	0.500	0.250
Yes	53			
No	39			
HI Type		0.45	0.970	0.940
ConHI	42			
MHI	3			
SNHI	3			
UNDEF	5			
ConHI		1.52	0.51	0.26
Mild	20			
Moderate	22			
Severe	0			
MHI		1.33	0.58	0.33
Mild	0			
Moderate	2			
Severe	1			
SNHI		0.67	1.16	1.33
Mild	2			
Moderate	0			
Severe	1			
Undefined		0.2	0.45	0.2
Mild	4			
Moderate	1			
Severe	0			

Table 3 shows the hearing impairment data as documented in participant records. The data revealed that 53 (57.6%) patients presented with hearing impairment (HI), whilst 39 (42.4%) records revealed hearing within normal limits on initial assessment. The type of hearing

impairment was recorded as undefined in 5 (9.4%) charts owing to the absence of bone conduction results. Of those records indicating the presence of HI, 42 (79.2%) had conductive HI (ConHI). Regarding laterality of ConHI, three (7.1%) were unilateral ConHI and 11 (26.2%) bilateral ConHI. Laterality could not be recorded for 28 records (66.7%) as testing was conducted through free field. Bilateral MHI was recorded in three (5.7%) participant records. Sensorineural HI (SNHI) was recorded in three (5.7%) participant records, and all cases of SNHI were unilateral. Regarding HI severity, of the 42 cases of ConHI, moderate ConHI was the most frequently occurring degree of loss. Of the three cases of MHI, moderate MHI was the most frequently occurring with no cases of mild MHI noted in the sample. Mild SNHI was the most common degree of SNHI with one recorded case of severe SNHI also documented. Of the five cases of undefined HI, four cases were of mild degree and one case was of moderate degree.

Table 4

Relationship between presence of otolaryngological conditions and hearing impairment

		Otolaryngological conditions		Total
		YES	NO	
Hearing impairment	NO	27	12	39
	YES	50	3	53
Total		77	15	92

Of those participant records that included a diagnosis of hearing impairment (n=53), the majority, 50 (94.34%) presented with at least one otolaryngological diagnosis (see **Table 4**). Chi-square analysis ($p = 0.001$) indicated a statistically significant association between otolaryngological conditions and the presence of HI. Further analysis using the Chi-Square test

showed a statistically significant relationship between the number of otolaryngological symptoms and the degree of hearing impairment ($\chi^2 = 20.908$, df = 12, $p = 0.052$). A statistically significant relationship between these variables was further confirmed by the Likelihood Ratio test ($p = 0.011$) and the Linear-by-Linear associated test ($p = 0.004$)

Figure 9

Otolaryngological manifestations in cases of hearing within normal limits, conductive hearing impairment, mixed hearing impairment and undefined hearing impairment.

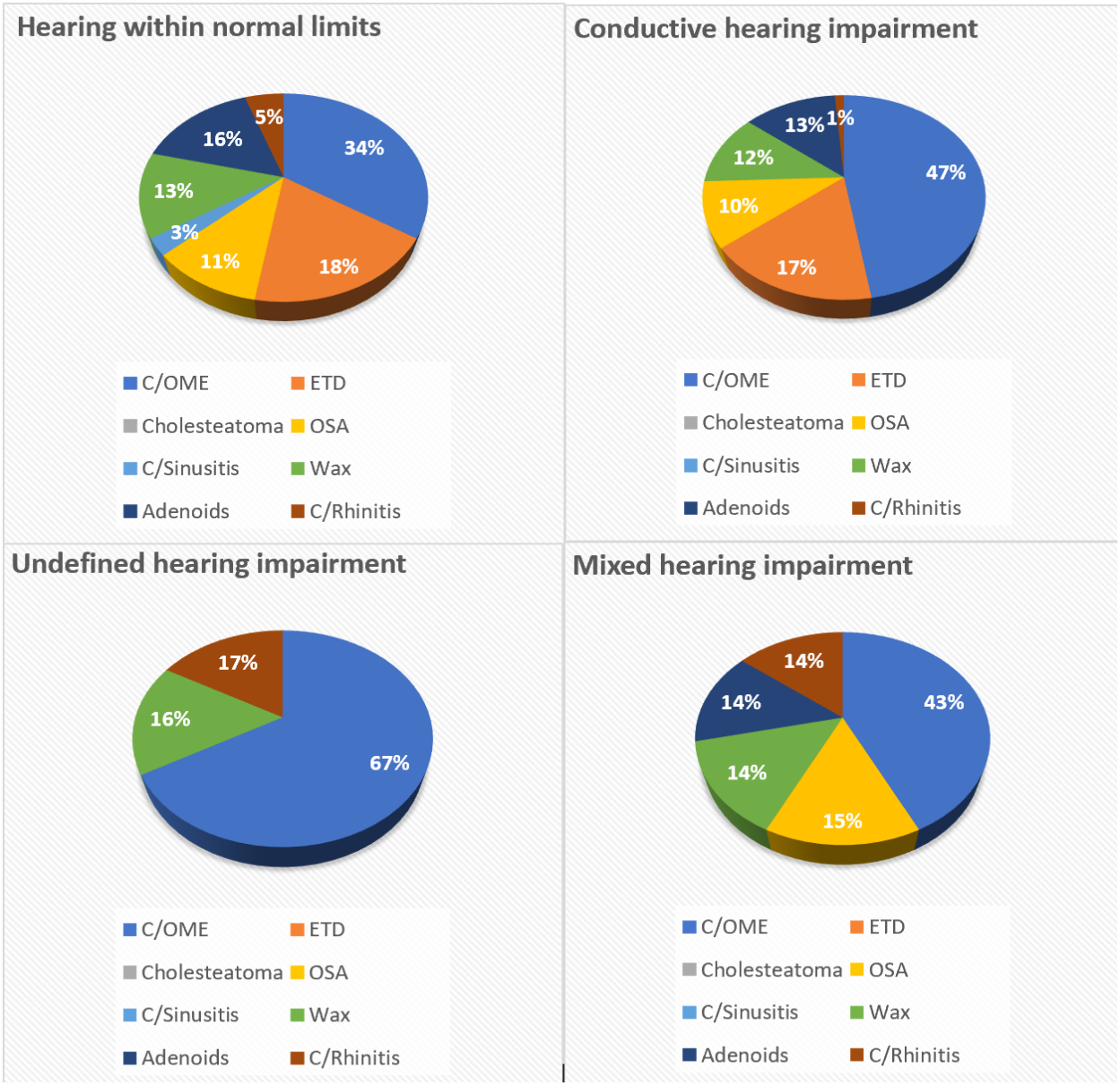


Figure 9 shows the occurrence of otolaryngological conditions in cases with hearing within normal limits and well as in cases of hearing impairment. Chronic otitis media and

Eustachian tube dysfunction were the most frequently occurring otolaryngological conditions, recorded in cases of hearing within normal limits and in cases of hearing impairment.

4.6. Hearing Surveillance practices

The third objective of the study was *to describe hearing surveillance practices undertaken for children with Down Syndrome at Sidra Medicine, Doha.* To determine audiology surveillance practices, the frequency of audiology clinic visits, waiting times between appointments and continuation of care per case was calculated. Of the 92 participant records included in the study, 17 (18.48%) records included single clinic encounters, and 75 (81.52%) records included more than one clinic encounter, with a maximum of nine visits and variance of 3.60 (m = 3.27, sd = 1.89).

For participant records with single visits (n=17), nine (52%) records revealed that the case was lost to follow-up because no follow-up referral for Audiology was provided despite the recommendation of continued audiology care as per ENT discretion. All nine of these cases were seen on referral from ENT. On completion of audiological testing, patients were sent back to ENT with the recommendation of continued audiology care as per ENT discretion. In cases in which records (n=75) revealed more than one clinic encounter at audiology, the majority 41 (54.67%) of records confirmed that patients were still actively managed within the Audiology department.

Table 5

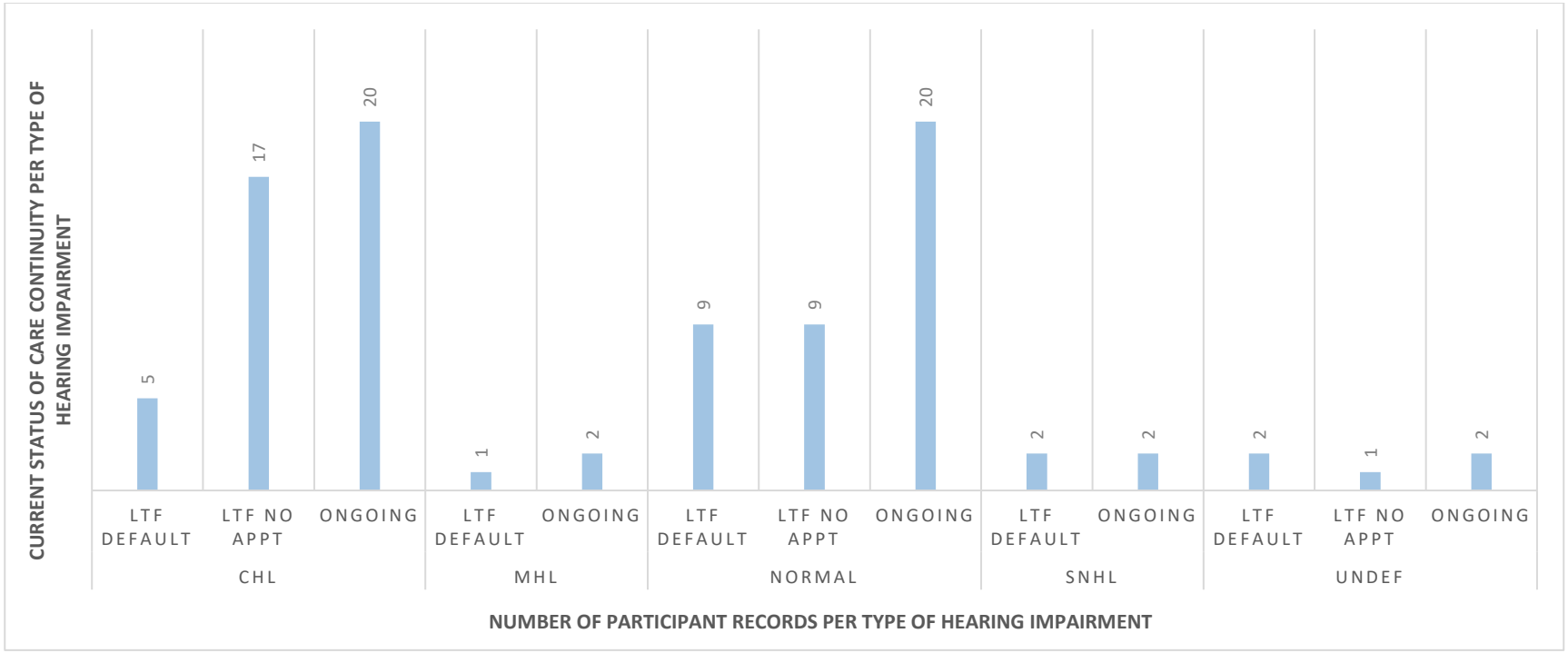
Continuity of care in patients with Down syndrome (N=92) in the audiology clinic

Visits	Lost to Follow up				Active	
	Defaulted		No Appointment			
1 visit	3	17,64%	9	52.94%	5	29.41%
>1 visit	16	21.33%	18	24%	41	54.67%

After removing records for single clinic encounters from the calculation, the average waiting time between clinic visits was calculated. The minimum wait time between appointments was 2 weeks and the maximum waiting time was 25 months with a variance of 22.40 (mean = 7.79, sd = 4.73). Table 5 shows the current status of continuity of care for the two groups of records.

Figure 10

Continuity of audiological care in patients with and without hearing impairment (N=92)



The average waiting time between appointments for patients actively under the care of the audiology team was 8.53 months. For those who defaulted appointments and were resultantly no longer under the care of the audiology team, the average waiting time was 7.40 months during the period they remained under the care of the Audiology department. Participant records reflected that 18 (19.6%) patients with hearing within normal limits were lost to follow-up (Figure 8). Hearing impairment was recorded in 28 (30,43%) records of participants who were lost to follow up and were no longer under the care of the audiology team. Of these, 22 (23.91%) had CHI, 1 (1,09%) had MHI, 2 (2,17%) had SNHI and 3 (3,26%) had hearing impairment which was classified as undefined.

4.7. Audiological and ENT management

The fourth objective of the study was *to describe the audiological and ENT management practices for children with Down Syndrome at Sidra Medicine, Doha.* A review of participant records reflected six management strategies commonly recommended by ENT and Audiology teams for patients with DS presenting with otolaryngological conditions and hearing impairment. Both teams function independent of each other however collaborate on patients with conditions placing them at risk for hearing loss. Typically, patients would be seen initially by ENT team and referred by the ENT physician to the Audiology team for assessment. Information regarding the management strategy utilized prior to the initial visit at Sidra Medicine was obtained from the case history information provided by the primary caregiver, and the subsequent management strategies utilized were obtained from the management plan documented in each participant record. The various management options were categorized on an assumed clinical progression in management from less intensive watchful wait periods without treatment, to more intensive surgical or assistive device recommendations. These were based on the recommendations of the attending Ear Nose Throat physician. The categories included:

- Grade 0 - Watchful wait period (recommended in asymptomatic/ mildly

symptomatic cases)

- Grade 1 - Medication prescribed for a predetermined treatment period
- Grade 2 - Adenoidectomy
- Grade 3 - Myringotomy and grommet insertion, Adenoidectomy with myringotomy and grommet insertion
- Grade 4 - Hearing aid or assistive listening devices

Table 6

Frequency of Management techniques undertaken on initial and subsequent clinic visits (N = 92)

Initial Management	*%*	*n*	Latest Management	*%*	*n*
Grade 0	38.04%	35	Grade 0	26.09%	24
Grade 1	41.30%	38	Grade 1	33.70%	31
Grade 2	1.10%	1	Grade 2	4.35%	4
Grade 3	19.56%	18	Grade 3	29.35%	27
Grade 4	0	0	Grade 4	6.52%	6

Table 6 reflects the frequency of use of the different management strategies. Management techniques undertaken prior to initial consultations at Sidra Medicine (i.e. made when treatment was sought at other facilities) or that which was recommended at Sidra Medicine for the first time (having not been managed elsewhere previously) was classified as "Initial Management" for the purposes of the study. In the category of Initial Management, medical management was recommended in 38 (41.30%) of cases. Watchful waiting was recommended in 35 (38.04%) of cases, despite 19 (54.29%)

of these cases having confirmed otolaryngological findings. Alternate management strategies were recommended in 19 (20.66%) cases.

The management technique most recently employed at Sidra Medicine during the course of a patient's care (within the study period) in the ENT and Audiology departments was classified as "Latest management" for the purposes of the study. In the category of Latest management, medical management and watchful wait was suggested for 31 (33.70%) and 24 (26.09%) of cases respectively. Surgical interventions were recommended for 33 (35, 87%) patients, and six patients with DS presenting with hearing impairment received hearing aids. Compared with Initial Management, medical interventions were recommended for seven less patients and watchful wait periods were recommended for 11 less patients. Compared to initial recommendations, surgical intervention was recommended for nine more patients, and six patients received hearing aids while no patients had been recommended hearing aids following their initial assessments.

Table 7

Hearing test results following the use of Initial Management (n =92)

		Type of Hearing impairment					Total
		CHI	MHI	NORMAL	SNHI	UNDEF	
Previous Management	GRADE 0	15	0	18	2	0	35
	GRADE 1	15	1	18	1	3	38
	GRADE 2	0	0	1	0	0	1
	GRADE 3	12	2	2	0	2	18
	GRADE 4	0	0	0	0	0	0
Total							92

Table 7 shows a comparison of previous management with initial audiometric findings and displays information on hearing status related to the various management options received. Conductive hearing impairment (ConHI) persisted in 15 (35.71%) cases after medical treatment was received. Additionally, eight (19.05%) records showed persistent ConHI after having had grommet insertion while four (9.52%) records showed persistent ConHI after having had adenoidectomy and grommet insertion. Of three patients with MHI and three with SNHI, no recommendations for hearing aid fitting or cochlear implantation had been made.

Table 8

Comparison of initial management with follow-up management strategies in patients with Down syndrome (n =92)

Initial Management	Latest Management	(Number of participant records)
SURGICAL MANAGEMENT	Watchful wait	0
	Medical Management	7
	Surgical Management	9
	Hearing aid	3
MEDICAL MANAGEMENT	Watchful wait	7
	Medical Management	16
	Surgical Management	13
	Hearing aid	2
WATCHFUL WAIT	Watchful wait	17
	Medical Management	8
	Surgical Management	9
	Hearing aid	1

Initial management was compared with latest management to determine the frequency of need for follow-up or secondary management options in each case (see **Table 8**). In 17 (48,6%) cases in which a watchful wait period was recommended, the same follow-up recommendation was made. In the remaining 18 (51.43%) cases, the presenting symptoms had worsened. Of these, medical management was provided in eight (44.4%) cases, surgical management was planned in nine (50%) cases, and a hearing aid fitting was recommended in one (5.7%) remaining case. In cases where medical management was initially recommended, seven (18.4%) were later recommended for a

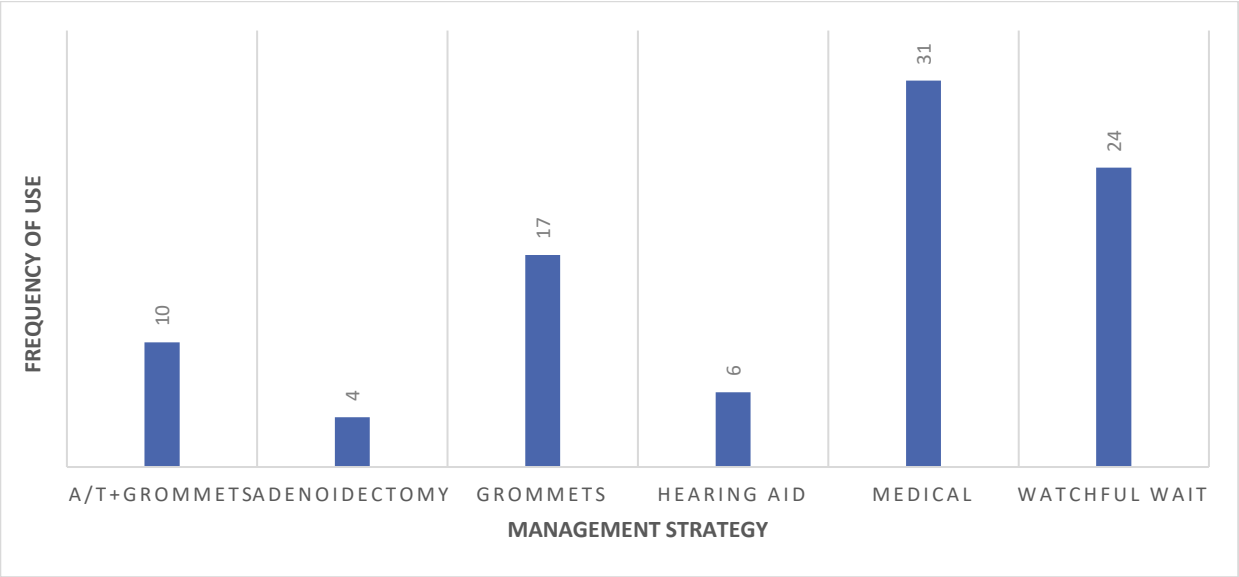
watchful wait period, 16 (42.1%) were followed up with further medical management and 13 (34.21%) had follow-up surgical interventions. In two (5.26%) cases, a hearing aid fitting was recommended. In 19 cases in which surgery had been the initial management option, seven (36.84%) recommendations for medical management were made due to recurring otolaryngological symptoms. Follow-up surgical intervention was recommended in nine (47.37%) cases and 3 recommendations for hearing aid fitting were made. Repeat grommet tube placement was recommended in seven cases, adenoidectomy with grommet insertion in one case and adenoidectomy in one case.

4.8. Relationship between management strategies and changes in audiological findings

The fifth objective of the study was *to determine the relationship between management strategies and audiological findings over time in children with Down syndrome at Sidra Medicine, Doha.* To achieve this, the frequency of use of the most recent management approach in each case was determined, following which the changes in degree of loss of hearing from periodic audiograms were used to determine the changes in hearing outcomes.

Figure 11

Use of recent management strategies in patients with Down syndrome (N = 92)



As per the categories outlined in Section 4.6, the frequency of use of the five different management approaches is shown (see **Figure 11**). Medical management and surgical management were recommended 31 (33.7%) and 31 (33.7%) of cases respectively. Watchful waiting was recommended in 24 (26.1%) cases. The recommendation for a hearing aid fitting was made in six (6.5%) cases.

Figure 12

Impact of management strategies on hearing outcomes in patients with Down syndrome (n = 79)



The different management strategies were compared with the latest, post-management pure tone findings to determine potential relationships between management styles and hearing status. Data from 13 (14.13%) participant records were excluded at this point owing to an absence of previous audiological data preventing the determination of changes in hearing outcomes. Figure 12 shows that all patients fitted with hearing aids showed improvements in hearing outcomes. Of these, three (50%) had CHI, two (33.3%) had MHI and one (16.7%) had SNHI. Surgical approaches resulted in improved hearing outcomes in 14 (51.9 %), with no changes in hearing observed in eight (29.6%) cases and further deterioration in hearing observed in two (7.4%) patients. Equal numbers of cases i.e. six (19.4%) had improvement in hearing and unchanged hearing outcomes following medical management. Hearing deteriorated in two (6.5%) cases in which medical management was recommended.

Following the removal of cases with hearing within normal limits and those in which changes in hearing outcomes could not be determined (i.e. only one set of hearing test results available in the patient medical chart), 52 cases remained. To investigate the possible relationship between recent management approaches and hearing outcomes, correlation analysis was employed. The Spearman's rho correlation coefficient between recent management and hearing outcomes was -0.451, indicated a moderate negative correlation between the two variables. The p-value associated with the correlation coefficient is < 0.001 ($p < 0.001$). This indicated that the correlation was statistically significant at the 0.01 level (two-tailed).

CHAPTER 5: DISCUSSION

5.1 Introduction

This chapter will discuss significant findings presented in the results chapter to contextualize the results and answer the research question; "What are the assessment practices and management strategies employed for paediatric patients with Down syndrome, who are at risk for otolaryngological manifestations and hearing impairment at Sidra Medicine in Doha, Qatar?". The chapter will begin with an introduction to the factors pertaining to hearing impairment (HI) in children with DS and highlight how the current study addresses knowledge gaps within the field in the local region, after which the results will be further discussed with reference to related literature.

Down syndrome (DS) is a recognized risk factor for HI. Studies have suggested that caregiver perception of HI and objective HI in children with DS are poorly correlated, substantiating greater advocacy for audiological care (Lau, Ko and Cheng, 2015). The contribution of otolaryngological manifestations to HI in children with DS, and the consequential impact of HI on their speech and language acquisition necessitates a multidisciplinary team (MDT) approach (Fortnum et al, 2014). Despite the obvious risks, DS is an under-researched topic in Qatar, specifically with regards to hearing in children with DS residing in the region. The current study aimed to explore the approaches to assessment and management utilized by the ENT and audiology teams responsible for the hearing care of children with Down syndrome at Sidra Medicine, Doha.

The results of the current study showed that Nondysjunction was the most common type of DS in the sample, which is consistent with widely reported data globally (Resende et al, 2023, Holcomb, 2024). Studies show considerable variation in the reported sex ratio for DS live births, however a large

study conducted over a 20-year period in Russia showed a male preponderance in Trisomy 21 (all types) with a sex ratio (SR) of 1.22 (Kovaleva, 2002). Similarly, the current study contained more records for male patients with a confirmed diagnosis of DS than females, and a SR of 1.191. The mean age of patients' mothers in the current study was 35.9 years, supporting the assertion that advanced maternal age is linked to higher incidence of DS (Newberger, 2000). The majority of patients (n=77) in the sample were of Arab descent, however this must be viewed in light of the fact that the majority of people residing in the country, both Qatari nationals and expatriates, are Arab Muslims. Notably, the oldest mother was 52 years of age and of Arab descent, which may support earlier reports from the Middle East suggesting that conception is expected to continue until menopause in Arab and Muslim women (Hamamy & Bittles, 2008, Fayed et al, 2017). The results reflected a notably high incidence of parental consanguinity in the sample, however its relation to the high incidence of DS in Qatar may be difficult to ascertain, as previous studies provide conflicting evidence on the relationship between consanguinity and DS. However, since Ray et al (2018) and Hashishe (1998) reported potential links between consanguinity and DS, further exploration in the area may be warranted especially because the practice of consanguinity remains integral to marriage practices within the Arab community.

Otolaryngological conditions in children with Down syndrome

The American Academy of Pediatrics reported a 50%-75% risk of chronic otitis media in children with DS. Other otolaryngological conditions commonly seen in children with DS include stenosis of the external ear canal which can result in wax impaction and preclude immittance measurements thereby delaying diagnosis of middle ear conditions, as well as Eustachian tube dysfunction with results from collapsing Eustachian tubes (Nash et al, 2014 Kreicher et al., 2018). Obstructive sleep apnea, chronic sinusitis, chronic rhinitis, and acute otitis media have also been reported (Kreicher et al., 2018) The results of this study were commensurate with these reports and

showed a high incidence of otolaryngological conditions, confirmed by immittance findings and/or examination by Ear, Nose and Throat (ENT) physician. The most frequently occurring conditions in the sample were chronic otitis media with effusion (OME), Eustachian tube dysfunction (ETD), adenoid hypertrophy and impacted cerumen. The study confirmed and highlighted the vulnerability of children with DS to persistent otolaryngological conditions. Chmielik et al (2023) reported that the health-related quality of life was compromised in children with recurrent otolaryngological conditions and required special consideration during clinical examination and management. Numerous scholars advocate for improved accessibility, streamlined audiological and ENT services and periodic multidisciplinary team (MDT) surveillance to mitigate the effects of otolaryngological conditions on hearing, communication and quality of life (Santoro et al, 2023, Mengoni and Redman, 2019, Mattaa et al 2011, Roizen, 2002)

While the current study did not investigate the underlying mechanism for the common otolaryngological conditions, it is well understood from existing literature that abnormalities of the upper airway and orofacial area place children with DS at risk for persistent otolaryngological conditions from infancy into later childhood (Chin 2014, Maris et al, 2014, Shott et al, 2001). Austeng et al (2013), reported that by age 10 the risk of OME reduced to 3-4%, from 10-20% in 2–5-year-old typically developing children. Comparatively, of 93% of children with DS diagnosed with OME 75% would go on to have chronic OME, persisting beyond the age of 6 years (Manickam et al, 2016, Maris et al, 2014). Mitchell, Holmes, and Turner (2018) reported an 80% likelihood of improvement of OME in 3 months in typically developing children, while OME in children with DS persisted in the same period and beyond.

The current study found that many of the children in the sample presented with multiple co-occurring otolaryngological conditions and that those under the age of six years of age appeared more at risk for persistent Eustachian tube dysfunction, adenoidal hypertrophy, and obstructive sleep apnea as well as high numbers of children from 0-5.11 years of age presenting with chronic OME and ETD. While other otolaryngological conditions investigated in the current study appeared to diminish with age, chronic OME, ETD and wax impaction continued into later childhood, although to a lesser extent. The high incidence of these conditions and their persistence into later childhood supports the need for frequent Audiological and ENT consultations, particularly when one considers their link to hearing impairment. While the management of otolaryngological conditions and prevention of associated hearing impairment (HI) is possible, greater vigilance of these conditions in children with DS is warranted due to their persistence throughout childhood and their impact on hearing and speech and language development outcomes.

Audiological profiles of children with Down syndrome

Previous prevalence studies confirmed HI variability ranging from 31%-81% in children with DS. Bull (2022) found that 75% of children with DS were at risk for hearing loss. Similarly, Shott et al (2001) reported that 81% of children with Down syndrome presented with hearing loss. All types of HI including conductive hearing impairment (ConHI), sensorineural hearing impairment (SNHI) and mixed hearing impairment (MHI) are seen in children with DS and are varied in severity and likely to fluctuate based on the etiology of the hearing loss (Laws & Hall 2014). In the current study, 50 of 53 patients presenting with HI on their initial assessment had co-occurring otolaryngological conditions, and statistical analysis confirmed a significant relationship between otolaryngological conditions and HI. Hearing within normal limits was found in 42.2% of patients on their initial clinic visit despite them having at least one confirmed otolaryngological condition. There were 15 patients who did not

have otolaryngological diagnoses on their initial visit, however 46,7% of them had at least one otolaryngological condition, with 71.4% also presenting with associated hearing impairment, on follow up sessions. These findings suggest that the occurrence of otolaryngological conditions in children with Down syndrome may fluctuate, as may the associated hearing impairment, substantiating close audiological and ENT monitoring. Targeted audiological surveillance following newborn hearing screening, with the use of suitable assessment procedures, is supported in the literature specifically in those populations at high risk for hearing loss (Fitzgibbons et al, 2024). Raut et al (2011) recommended surveillance throughout childhood using appropriate assessment procedures at timeous intervals, to determine the incidence, timings of presentation and type of hearing loss seen in children with DS. It has also been reported that the use of surveillance guidelines and care models may reduce loss to clinic follow-up (Fitzgibbons et al, 2024)

Moderate conductive hearing impairment (ConHI), associated with the aforementioned otolaryngological condition, was the most common type of HI identified in the current study. Mixed hearing impairments and SNHI were present in the sample, to a lesser extent than ConHI, consistent with findings of numerous other studies (Schrijver et al, 2019, Kreicher et al, 2018, Lau, Ko & Cheng, 2015, Park et al, 2012). Persistent, untreated ConHI compounds these difficulties, worsening auditory performance and negatively affecting receptive language, vocabulary, phonology, syntax, and auditory memory as well as speech intelligibility (Saksida et al, 2021, Chapman, 2000). In general, when compared with unaffected peers, children with ConHI score lower for hearing related quality of life measures (Smit et al, 2021). Early identification and management of otolaryngological conditions underlying ConHI may be the key to counteracting the effects of HI on speech and language development, socioemotional development, and academic progression. Early intervention for HI in at-

risk children with DS is supported by well-designed and adequately implemented hearing surveillance protocols (Manickam et al, 2015)

Audiological surveillance for children with Down syndrome

Adequate audiological surveillance is a fundamental component to the provision of effective hearing healthcare and maintaining quality standards in audiology. Lau, Ko and Cheng (2015) advocate for audiological surveillance in the DS population due to high prevalence, underestimation of and low parental awareness of hearing loss. The increased risk of recurrent otolaryngological conditions and HI in children with DS necessitates frequent review and expedited management. Concerningly, long waiting times and high lost to follow up/default rates were identified in the current study. Disrupted continuity of audiological care caused by high lost to follow up (LTF) rates which resulted from, in part, inadequate provision of follow up appointments. The absence of guidelines outlining surveillance timeframes and standardized care protocols for children with DS with HI may have contributed to the inadequate provision to follow up appointments seen in the current study. Other reasons identified for non-adherence to appointments which included "forgetfulness", "financial or transportation problems", "sickness" and parent and caregiver overcommitment "busy" (Fadzil et al, 2021). Long average waiting times for follow up appointments seen in the study may have, in part, been influenced by the Covid 19 pandemic from 2020 to 2021. Services in the Audiology department were interrupted and many patients missed scheduled appointments due to restricted movement during this period. Santoro et al (2023) also found that compliance to audiology appointments in patients with DS decreased from 58% to 45% during the Covid-19 pandemic, the lowest adherence any of the 5 medical services investigated in the study. Reasons identified for the decrease were the reduction in non-essential services and the hesitation on the part of the patient to attend appointments viewed as non-essential due to increased infection control concerns (Santoro, 2023). It could be understood that

during periods of uncertainty and limited access to healthcare services, caregivers may prioritize healthcare needs based on their understanding of what is more critical. Whicker (2020) have also reported that hearing is often under-prioritized compared to other seemingly more critical health concerns.

Children with DS have disproportionally reduced access to important healthcare services and are frequently affected by poorly coordinated care and disruptions to healthcare continuity (McCabe, Hickey and McCabe, 2011). This is concerning, although possibly not uncommon for the DS community, as a survey in Jamaica also reported a 30% adherence to hearing care guidelines in children with DS (Livingstone-Sinclair, Scott and Trotman, 2018).

Appointment non-adherence, while preventable, is associated with inefficiency of healthcare services, delays in appropriate management and lost opportunities for vital health education (Fadzil et al, 2012). For the audiologist, delays in follow-up appointments undoubtably impedes early intervention and affects optimization of hearing care service delivery. Of those patients who were LTF in the current study, 34% had been diagnosed with HI, thereby highlighting the relevance of and opportunity for facilitating improved appointment compliance and reducing LTF and default rates in patients with DS. One tool to support improved adherence to appointments is intensive family and caregiver counselling during initial encounters emphasizing the predisposition of patients with DS to conditions affecting middle ear function, the high likelihood of hearing impairment in childhood and the compounding effects of hearing impairment on existing speech and language development delays. One specific contributor to LTF in the current study was non-provision of follow up appointments. To remedy this problem, clinicians should reference standardized guidelines when determining follow up appointment requirements, thereby pre-empting the audiological needs of patients and facilitating

essential periodic surveillance. Such guidelines should include protocols for newborn hearing screening as well as targeted follow up at timeously intervals thereafter and may also suggest age-appropriate test procedures specific to the paediatric DS population. Lagan et al (2021) also showed that implementing a health surveillance clinic in Dublin improved adherence to clinic guidelines as well as improved opportunities for spreading health awareness. With regards to hearing surveillance in children with DS, audiologists retain the primary responsibility to promote and drive the hearing care of these high-risk patients.

Management of Hearing Impairment in children with Down syndrome

Hall et al (2018) found variations in clinical decision-making pertaining to the management of middle ear conditions and HI in children with DS were primarily guided by perceptions of professional remit, individual preferences and organizational factors. Hall et al (2018) identified the apparent difficulty of Audiologists and ENTs to differentiate between inherent speech and language, learning and behavioural challenges of children with DS and to those related to HI. The majority of related studies however emphasized the importance of management of HI resulting from middle ear conditions in children with DS, particularly in light of the compound effect untreated HI has on all domains of speech and language development, socio-emotional development, and academic performance (Nightengale, 2017, Kriecher, 2018, Hall et al, 2018, Roizen, 1993)

The management otolaryngological conditions and the resultant HI requires coordination between the Audiologist and the ENT physician, with various management strategy options available (Hall et al, 2018). The current study identified the options for management to include watchful wait/observation, medical treatments, surgical managements and the use of assistive devices. Following initial consultations with the ENT team, conservative approaches to management including a watchful wait period and medical treatments were most often recommended. These conservative

strategies, however, proved to be minimally efficacious with 71.42% of patients continuing to show signs of HI following either a watchful wait period or after receiving medical treatments. This is supported by literature highlighting that medical treatments were inferior to surgical interventions when managing upper airway obstructions and associated middle ear conditions in children with DS (Kavanagh, Kahane and Korden, 1986, Jacobs, Gray and Todd, 1996). Alternate management options, including surgery and utilization of hearing amplification devices, when dealing with middle ear conditions causing HI in children with DS are indicated.

Surgical options to manage underlying causes of hearing impairment in children with DS

The clinical decision making regarding surgical management of these cases is guided by the ENT physician. Surgical options included adenoidectomy and the use of grommets or pressure equalizing tubes (PET) or a combination of these two procedures. Mixed reviews on the efficacy and application of these procedures in the paediatric DS population have been raised. Price et al (2004) raised concerns regarding the efficacy of adenoidectomy in reducing middle ear related symptoms citing increased risk of complications and potential need for additional surgeries. Mitchell, Call and Kelly (2009) found higher rates of peri- and postoperative complication for routine otolaryngological surgeries in children with DS compared with their unaffected peers. It has been reported that while tonsillectomy, adenoidectomy and grommet placement is considered safe in children with DS, high rates of postoperative complications were higher and longer postoperative hospital stays were often required (Sait, Alamoudi & Zawawi, 2022).

Contrasting reports on the effectiveness of grommet or pressure equalizing tubes in the DS population have been reported. Sait, Alamoudi and Zawawi (2023) reviewed 21 articles reporting on the use of grommets/PET in children with DS and concluded that, while some studies showed

increased complications with PET placement, others found the procedure minimally complicated and highly effective in reducing the effect of HI. Higher rates of extruded and blocked PET are seen in children with DS due to more tenacious effusion resulting in the need for repeated insertions in up to 55% of cases (Shott et al, 2001, Sheehan and Hans, 2006). In some cases, stenosed ear canals prevent the use of PET (Sheenan and Hans, 2006). Alternative options for the management of HI in situations where repeated PET placement had limited benefit, or in which PET placement was not possible was recommended (Paulson, Weaver ad Macarthur 2014, Sidell et al, 2014, Sacks et al 2003). The findings of the current study supported the efficacy of grommet placement in alleviating HI in children with DS however repeated need for grommet placement was evident in many cases. Alternatives to medical treatments and surgery , such as hearing aid fittings, were seldom explored in patients with CHI in the study.

Use of hearing aids for conductive hearing impairment in children with Down syndrome

The study showed that hearing aids (HA) were recommended for 6 patients presenting with MHI and SNHI. No recommendations were made for HA fitting in patients with ConHI. Literature suggests that HA use in children with DS presenting with ConHI, who showed no improvement in hearing following surgical interventions, corresponded with improved quality of life measures (McDermott et al 2008, Nightengale et al, 2018, Sheehan and Hans, 2006). Conventional behind the ear (BTE) HAs should be considered as a first option based on cost, ease of use and limited invasiveness. Barriers to the use of conventional BTE HAs include stenosed ear canals, irregularly formed pinnae and compliance to HA use. The use of bone-anchored (BAHA) or bone conduction HAs in such cases was recommended by Sheehan and Hans (2006) and McDermott et al (2008) who reported high patient and caregiver acceptance and satisfaction with the use of BAHA devices.

The results of the current study were consistent with related literature and highlighted the vulnerability of children with DS to persistent otolaryngological conditions and HI, most frequently ConHI. It showed that despite limited success rates, recurrence of symptoms and limited improvement in hearing sensitivity, conservative management of otolaryngological conditions causing HI were favoured as a first line management strategy. The study also revealed that while more invasive surgical procedures were more frequently linked to improved hearing sensitivity compared with conservative treatment measures, otolaryngological conditions sometimes recurred, and hearing once again deteriorated. Based on the results, the option to recommend hearing amplification to remedy HI, even as a temporary solution, was not frequently explored. Hearing amplification, either by way of conventional BTE HAs or BAHA, may prove useful in mitigating the impact of HI resulting from persistent otolaryngological conditions in the paediatric DS population.

CHAPTER 6: SUMMARY, CONCLUSION AND RECOMMENDATIONS

6.1 Introduction

Children with DS face a range of medical conditions which threaten to compromise their quality-of-life outcomes. With the advancement of medical treatments and greater accessibility to healthcare services, even in remote areas, the life expectancy of people with DS continues to increase (Lulita et al, 2022). When considering the increased life expectancy in this vulnerable population, quality of life outcomes becomes more relevant. A significant contributor to quality of life is hearing. Good functional hearing is inextricably linked to speech and language development, communication and socialization (Grayson et al, 2019). Furthermore, hearing impairment has a negative impact on educational and vocational outcomes as well as socioemotional presentation in affected individuals. Careful consideration of the audiological care needs and provision of effective audiological care to children with DS is therefore essential in promoting their hearing related quality of life.

6.2 Summary of the Study Findings and Implications of the Study

6.2.1 Otolaryngological findings

The overwhelming majority of participant records reviewed for the current study contained findings of otolaryngological conditions as confirmed by atypical otoscopic examination and tympanometric findings. Recurrent otitis media with effusion, Eustachian tube dysfunction and impacted wax associated with small external ear canals was commonly diagnosed. It was not uncommon for participant records to reflect more than one co-occurring otolaryngological condition particularly int those aged between 0 – 5.11 years, highlighting the vulnerability of younger children with DS to otolaryngological conditions.

- *Implications for practice*

  Clinicians responsible for the otolaryngological and audiological care of children with DS should remain cognizant of the occurrence and persistence of otolaryngological conditions in this population. Regular assessment of middle ear function and close evaluation of the impact of strategies employed to manage otolaryngological conditions must be prioritized

6.2.2 Audiological Profiles

In addition to irregular middle ear findings, the current study confirmed that HI was prevalent in children with DS living in Qatar, with more than half the participant records included in the study reflecting a diagnosis of HI. Moderate ConHI was the most commonly diagnosed HI, resulting from the high incidence of otolaryngological conditions previously mentioned. Mixed HI and SNHI were also present, although to a lesser extent than ConHI, however with an incidence consistent with that previously mentioned in similar studies.

- *Implications for practice*

  With knowledge of the audiological profile of children with DS provided by the current study, the high risk for temporary and permanent HI, as well as the potential impact of HI on the process of speech and language acquisition also occurring within the early years of child development should not be ignored. Standard audiological care of children with DS must include routine periodic screening for HI and diagnostic evaluation for HI as warranted at intervals not exceeding 6 months, to minimize the consequence of HI on quality of life. HI should be anticipated, and adequate resources should be allocated to the management of HI in the population.

6.2.3 Audiological Surveillance

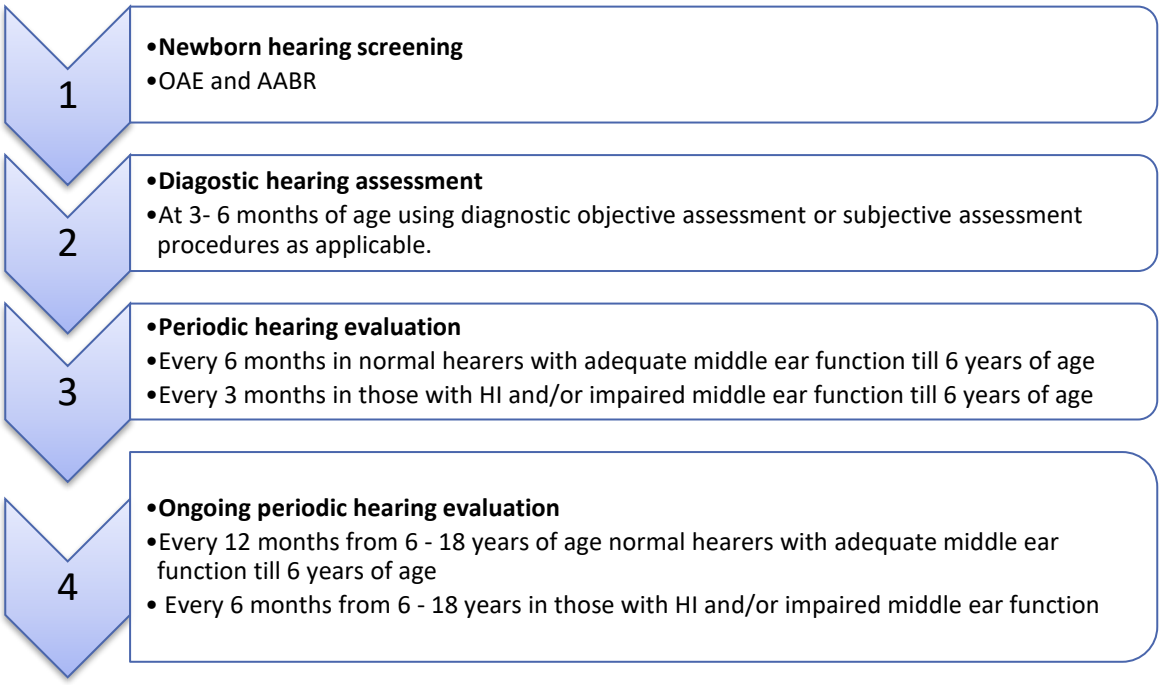
The current study showed long follow up appointment waiting times, which is potentially detrimental to the optimized audiological care of patients at risk for HI. Furthermore, high appointment default and lost to follow up rates were identified in the study, of which a common causative factor was clinicians' failure to provide patients with follow up appointment dates. Additional potential causative factors mentioned in other studies reviewed suggested the need for more clinician and parent/caregiver engagement as well as increased advocacy for audiological care.

- *Implications for policy*

  The study provided sufficient information to motivate policy makers to consider the development of standardized guidelines for the audiological care of children with DS in the region. Establishment of a task force responsible for the formulation of national guidelines to inform clinicians in the country of factors placing children with DS at risk for recurrent otolaryngological conditions and HI, as well as policy supporting the routine assessment of hearing in this special population would be invaluable to their audiological care. At an institutional level, Audiology protocols specifically designed to guide best practice in the appropriate hearing screening of infants born with DS, the ongoing surveillance for HI at timeously intervals in children with DS as they age as well as to provide suggestions for the effective management of HI in the population would be beneficial. Figure 13 shows a proposed protocol for the assessment of hearing in children with DS at the study site based on the existing literature reviewed for this study and with consideration of the outcomes of the study.

Figure 13

Proposed generic guideline to assess hearing in children with DS at the study site



- *Implications for practice*

  Further to policy making, strategies to support caregivers improve adherence to appointments to ensure adequate hearing surveillance is required. One such strategy could include the use of text messaging services or reminder calls prior to upcoming appointments. This may allow caregivers the opportunity to request a change of date should

they be unable to attend appointments and reduce default rates. Where available, educating caregivers on use of patient portals to access crucial health information including follow up appointments would be beneficial.

6.2.4 Audiology and ENT management in the DS population

Conductive HI observed in the current study was linked to otolaryngological conditions. Many of these conditions persisted after conservative treatments by ENT physicians. In most patients undergoing treatment for otolaryngological conditions which were also associated with HI, surgical approaches more often resulted in improved hearing outcomes when compared with more conservative treatment approaches. Recurrence of otolaryngological conditions post-surgical management was recorded in some participant records.

- *Implications for practice*

  Better collaboration between ENT and Audiology teams in the hearing care of children with DS may facilitate improved hearing outcomes in the population. More regular evaluation of hearing in patients with DS presenting with persistent otolaryngological conditions alongside ENT visits may heighten ENT physician awareness of associated HI in affected children and support more decisive clinical management. In cases where ENT management of underlying conditions resulting in ConHI has shown minimal restorative impact on hearing, Audiologists may recommend the use of hearing amplification devices, even as a temporary solution, to reduce the impact of HI on the speech and language and quality of life outcomes for children with DS. Special consideration should be given to the type of amplification device recommended, with suitable recommendations made by the audiologist based on the specific needs and the individual characteristics of the patient. For children

with DS presenting with MHI and SNHI, improving access to hearing aid more quickly will also support better outcomes for speech and language as well as quality of life.

6.3 Strengths and Limitations of the study

Due diligence was undertaken to ensure the integrity of the current study, however, as is not uncommon in research, some study limitations were identified. Table shows the strengths and weakness identified in the study.

Table 9

Strengths and weaknesses identified in the study.

STRENGTHS	LIMITATIONS
STUDY DESIGN	
The retrospective nature of the study ensured data was readily available for use, easy to access and that the study was affordable to conduct. The current study would provide a suitable foundation for future prospective studies for children with DS who have hearing impairment.	Some limitations owing to the retrospective nature of the study were identified. Many participant records originally reviewed did not include vital information required to meet the study objectives. Resultantly, many records were excluded from the study, thereby reducing the overall sample size. Utilization of data from medical records not originally collected for research purposes raised some researcher concerns about the accuracy of records, their completeness as well as whether the records are comprehensively recorded and necessitated close inspection of records before inclusion to ensure their reliability. With consideration of these factors, the study findings remain generalizable.
SAMPLE SIZE	
	The study was conducted at a fairly new site. Resultantly, utilization of clinical services was low at the start of the study period and increased over time. 92 participant records were included in the study, after those not meeting the study criteria had been excluded. The resultant sample size was relatively small compared with studies of similar nature

	however population constraints directly impacted the records available for inclusion. The small sample size limited the application of more complex statistical data analysis measures including multivariate logistical regression analysis, which may have been able to reliably measure relationships between variables in a larger sample.
DATA COLLECTION TOOL	
The tool utilized in the abstraction and collection of data was designed by primary researcher. This ensured that all information and variables required to meet the study objectives and answer the research question was available in participant records. Furthermore, the data was abstracted solely by the primary researcher using a standard guide, thereby eliminating misclassification bias.	

6.4 Conclusion

Locally conducted research is scarce in Qatar possibly because the healthcare system is still developing. While a few studies conducted locally confirmed that high incidence of DS in Qatar, a gap in knowledge of the local DS population is evident, specifically as it pertains to hearing and otolaryngology. The current study was the first in the country to explore hearing in the paediatric DS population. It revealed that a significant number of children with DS accessed Audiology and ENT services at the study site between the period 01 January 2018 to 31 December 2022, despite the test site being a new facility which was still refining and expanding its services. The study highlighted the importance of adequate access to effective and appropriately co-ordinated Audiological and ENT care and routine surveillance of children with DS for ear and hearing related conditions.

The study identified a high utilization of ENT and Audiology services amongst children with DS. This suggested that caregivers had sufficient awareness of the need for ear and hearing care, and the prioritization of ear related conditions in the overall care of children with DS. This awareness provides

a suitable platform for strong advocacy for audiological care, which can be undertaken not only by the audiologist, but also by primary care providers including paediatricians and primary healthcare workers.

The findings of this study supported the need to re-evaluate current approaches to management of otolaryngological conditions in the local paediatric DS population. Although not explored in great clinical detail as this was out of the scope of practice of the primary investigator, the study finding suggested that conservative approaches to the management of otolaryngological conditions (including watchful wait periods and medical treatments) were frequently ineffective in remediating symptoms and managing the underlying cause of these conditions. Since conductive hearing impairment of varying degrees was often associated with these otolaryngological conditions, efficient management thereof is of relevance to the Audiologist. This may support the need for improved collaboration between Audiology and ENT teams, allowing Audiologists opportunities to provide feedback on the HI associated with persistent otolaryngological conditions in children with DS which could support the ENT physicians' clinical decision-making process.

Approaches to the management of conductive HI associated with otolaryngological conditions may also need to be re-evaluated. The persistence of otolaryngological conditions and the complexities in their management results in continued temporary conductive HI, as confirmed by the study findings. The study revealed an opportunity to explore alternate, possibly temporary, hearing loss management strategies in these cases. One such alternative would be the issuance of suitable hearing aids, which would mitigating the burden of temporary HI on speech and language development, learning outcomes and hearing related quality of life in children with DS. This was confirmed by McDermott, et al

(2008) who reported high patient and caregiver satisfaction as well as improved quality of life outcomes in hearing impaired patients with DS subsequent to hearing aid fitting.

By evaluating current audiological practices, the study provided insights on potential shortcomings these practices and highlighted the need to formalize policy documents to guide the audiological care for this vulnerable group. With consideration of the outcomes of this study, recommendations for further research are presented.

6.5 Recommendations for future research

The current study highlighted the need for future research which could benefit clinicians and patients in the local context and supplement knowledge gained from research conducted outside Qatar. Considering the scarcity of research studies in Qatar, population-based studies as well as those which would inform policy and practice would be invaluable.

- *To explore the relationship between parental consanguinity and DS.*

  High rates of parental consanguinity in the region increases the risk for genetic disorders in their offspring. While reviewing relevant literature for the current study, conflicting reports on the association between DS and parental consanguinity were found. Further research into a possible relationship between the practice of consanguinity and the risk for DS would support improved pre-conception counselling in affected communities.

- *To evaluate the effectiveness of audiology protocols in improving audiological care in children with DS.*

  Upon formulation and implementation of institutional protocols, protocols should be evaluated for the efficacy in promoting better audiological care in children with DS and the need for possible amendment.

- *To investigate the impact of untreated hearing impairment on speech and language outcomes in children with DS.*

  While reviewing literature for the current study, little information could be found on the speech and language performance on children with DS who presented with hearing impairment. Such information would be valuable in guiding audiological care, promoting streamlined management of hearing impairment and supporting provision of speech and language outcomes in children with DS.

- *To evaluate hearing aid use in children with DS.*

  The current study, as well as others reviewed during the literature search, suggested that many children with DS presenting with hearing impairment who would benefit from hearing aid fitting had not received hearing aids. Research on the benefit of hearing aid fitting and its, impact on speech and language outcomes in children with DS would be useful. Potential barriers to hearing aid acquisition and utilization in this population including include cost implications, compliance with use, sensory aversions inhibiting hearing aid use should be investigated.

- *To investigate the involvement of multidisciplinary teams in the medical and rehabilitative care if children with DS.*

  Children with DS present with multi-organ system disorders which require extensive medical and therapeutic support from various health professionals. More efforts are required to investigate the need for better interdisciplinary collaboration in meeting the healthcare need of children with DS.

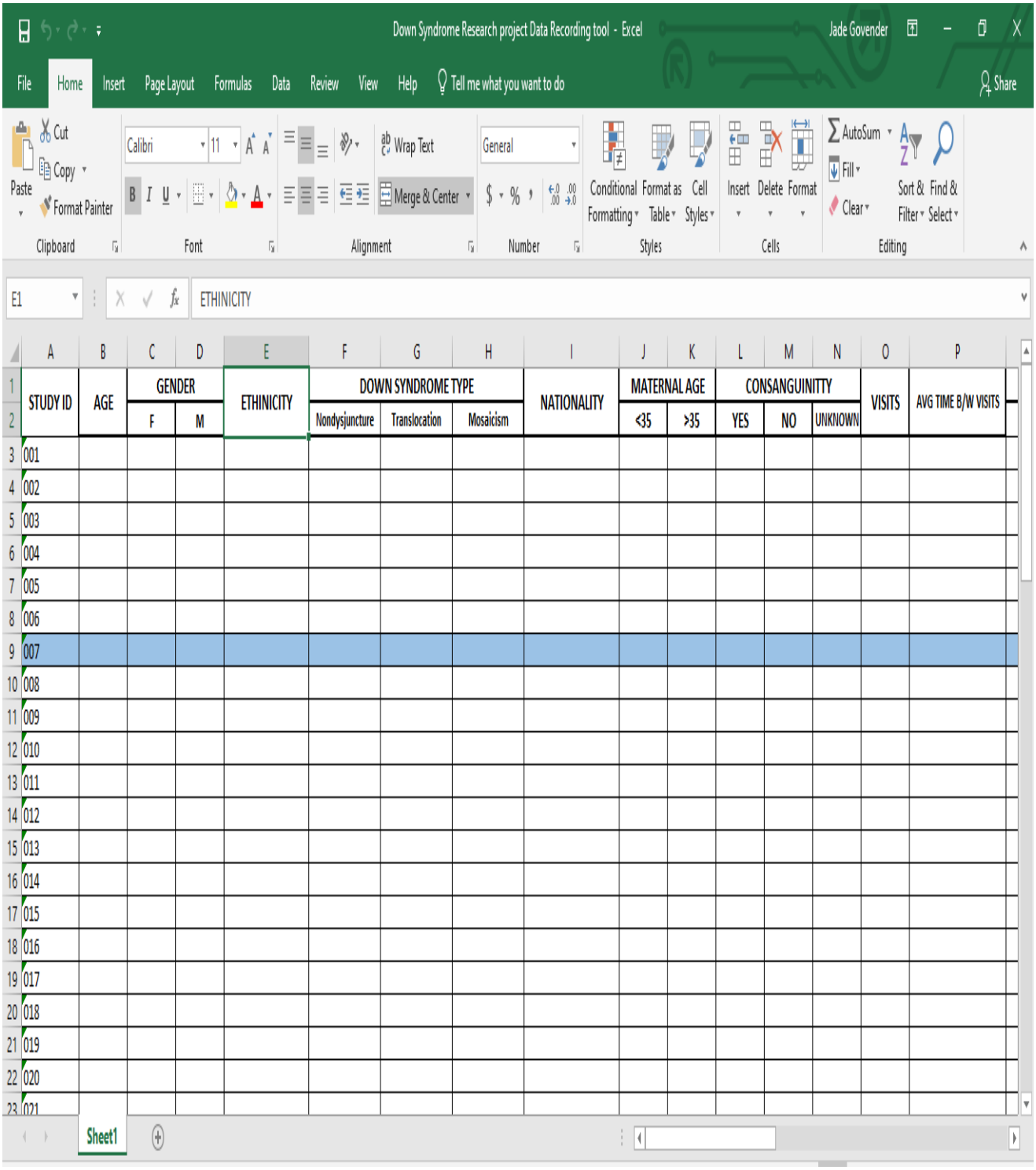
- *To develop a local DS registry in Qatar and support the establishment of patient/caregiver advocacy groups in the country.*

A national registry of children with DS in Qatar would be useful in tracking patients with the condition, addressing issues pertinent to their health and quality of life, increasing participation of children with DS as well as their parents/caregivers in meaningful clinical research opportunities. Down syndrome registries also benefit healthcare providers interested in research by supplying data, cohorts and relationships to better plan and coordinate clinical research initiatives.

APPENDICIES

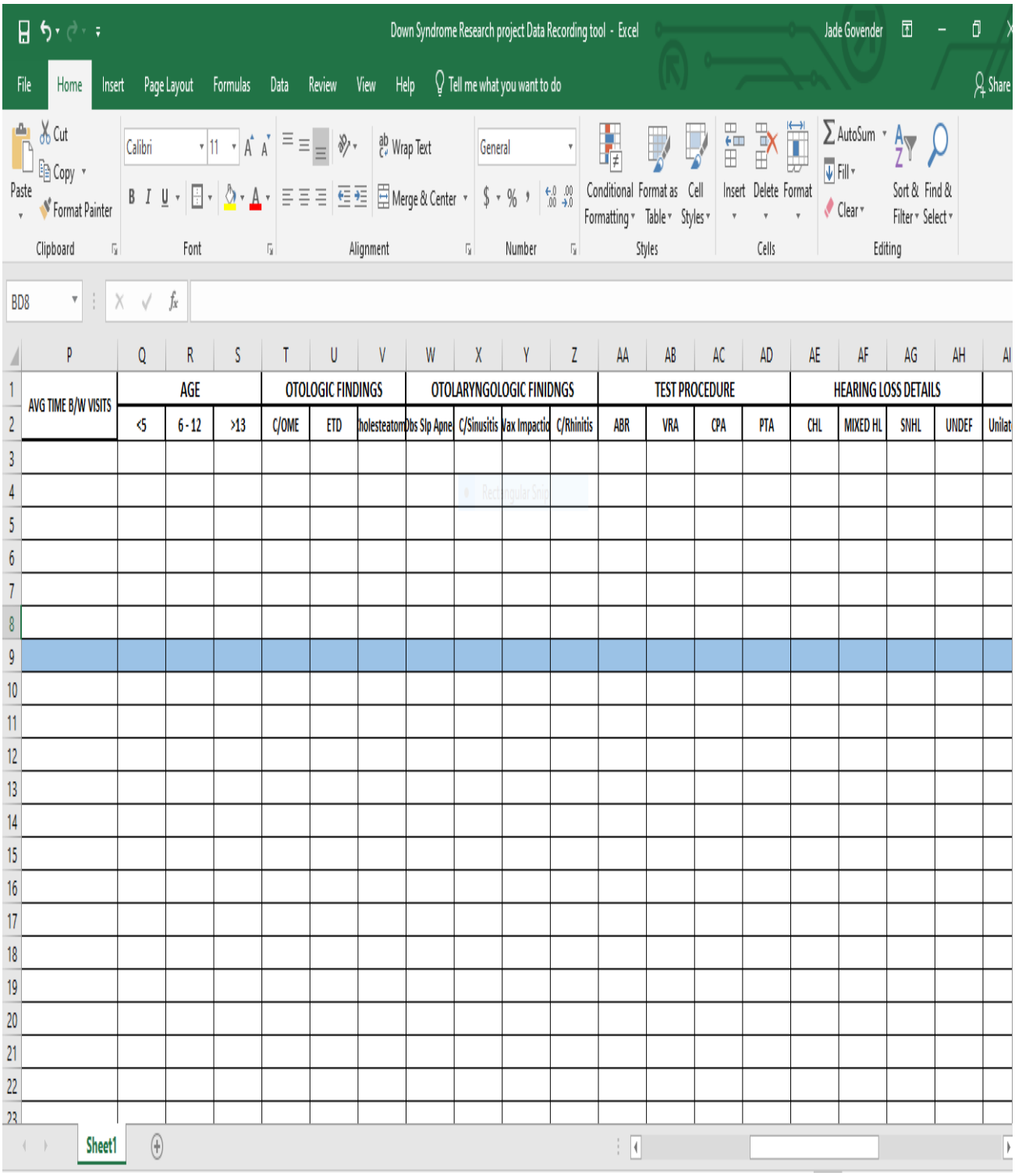
APPENDIX A1

Data Collection Tool section 1



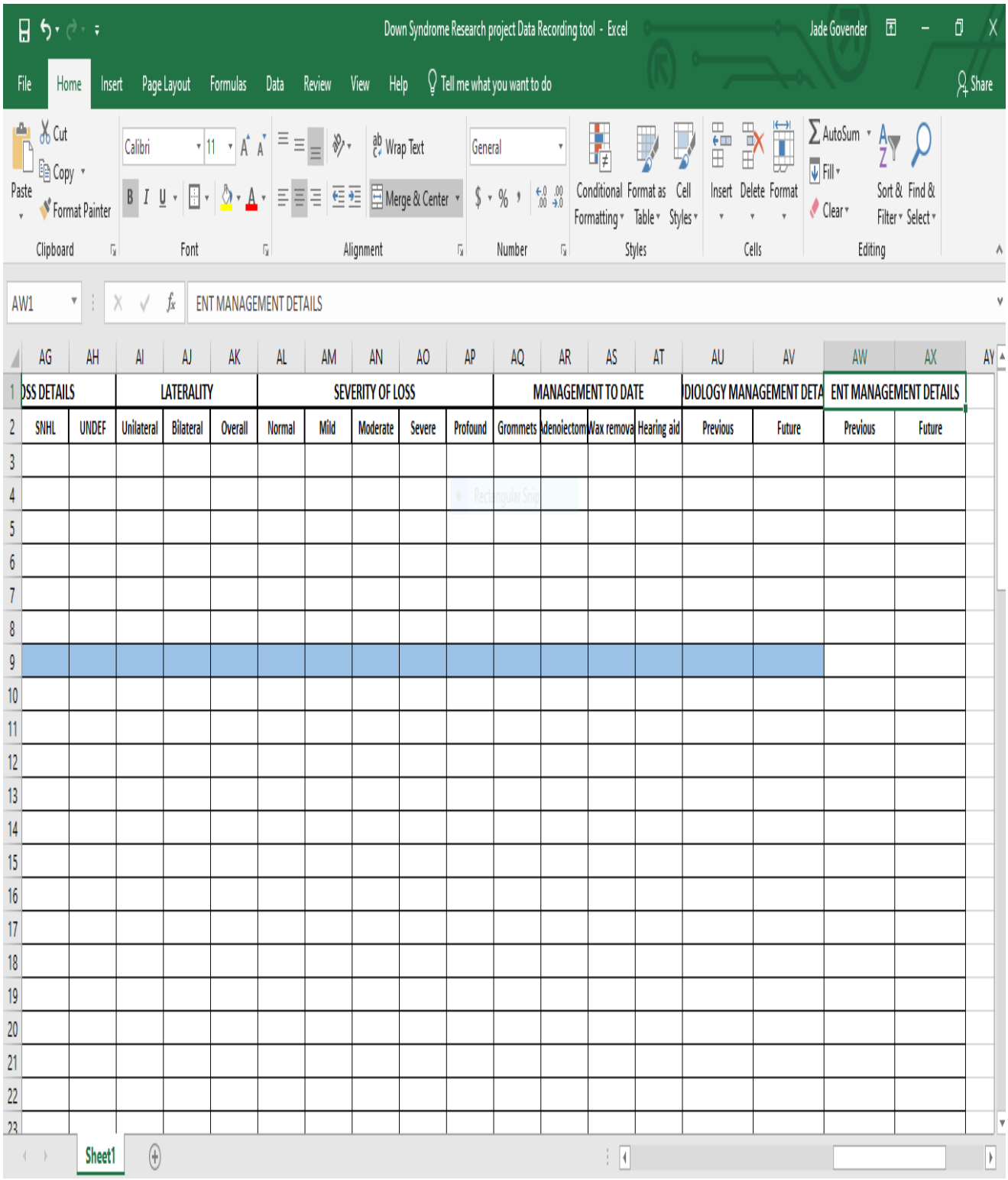
APPENDIX A2

Data Collection Tool section 2



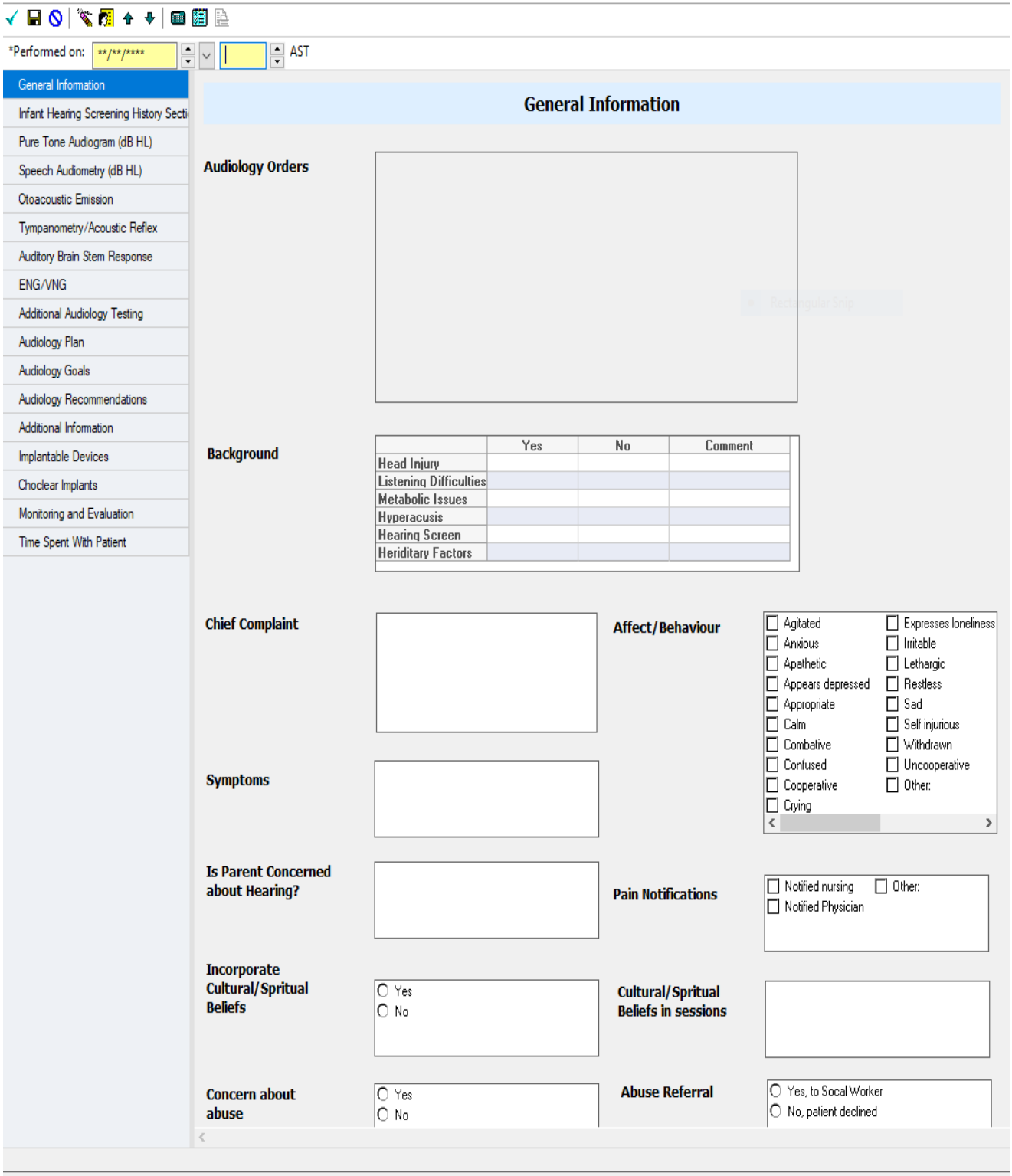
APPENDIX A3

Data Collection Tool section 3



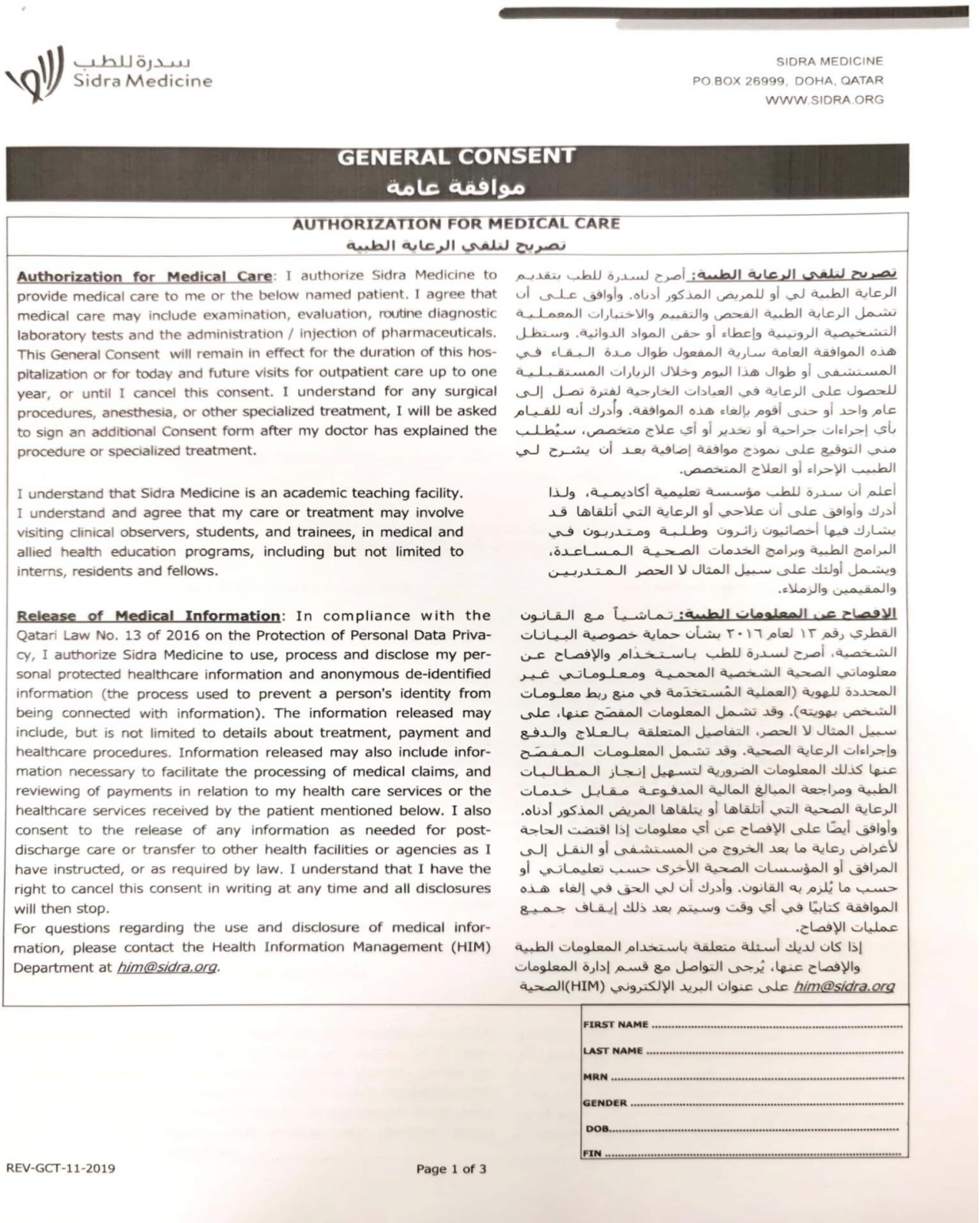
APPENDIX B

Audiology Reporting Template Cerner Powerchart



APPENDIX C1

General Consent for Treatment Page 1



سدرة للطب
Sidra Medicine

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WWW.SIDRA.ORG

GENERAL CONSENT
موافقة عامة

AUTHORIZATION FOR MEDICAL CARE
تصريح لتلقي الرعاية الطبية

Authorization for Medical Care: I authorize Sidra Medicine to provide medical care to me or the below named patient. I agree that medical care may include examination, evaluation, routine diagnostic laboratory tests and the administration / injection of pharmaceuticals. This General Consent will remain in effect for the duration of this hospitalization or for today and future visits for outpatient care up to one year, or until I cancel this consent. I understand for any surgical procedures, anesthesia, or other specialized treatment, I will be asked to sign an additional Consent form after my doctor has explained the procedure or specialized treatment.

I understand that Sidra Medicine is an academic teaching facility. I understand and agree that my care or treatment may involve visiting clinical observers, students, and trainees, in medical and allied health education programs, including but not limited to interns, residents and fellows.

Release of Medical Information: In compliance with the Qatari Law No. 13 of 2016 on the Protection of Personal Data Privacy, I authorize Sidra Medicine to use, process and disclose my personal protected healthcare information and anonymous de-identified information (the process used to prevent a person's identity from being connected with information). The information released may include, but is not limited to details about treatment, payment and healthcare procedures. Information released may also include information necessary to facilitate the processing of medical claims, and reviewing of payments in relation to my health care services or the healthcare services received by the patient mentioned below. I also consent to the release of any information as needed for post-discharge care or transfer to other health facilities or agencies as I have instructed, or as required by law. I understand that I have the right to cancel this consent in writing at any time and all disclosures will then stop.
For questions regarding the use and disclosure of medical information, please contact the Health Information Management (HIM) Department at *him@sidra.org*.

تصريح لتلقي الرعاية الطبية: أصرح لسدرة للطب بتقديم الرعاية الطبية لي أو للمريض المذكور أدناه. وأوافق على أن تشمل الرعاية الطبية الفحص والتقييم والاختبارات المعملية التشخيصية الروتينية وإعطاء أو حقن المواد الدوائية. وستظل هذه الموافقة العامة سارية المفعول طوال مدة البقاء في المستشفى أو طوال هذا اليوم وخلال الزيارات المستقبلية للحصول على الرعاية في العيادات الخارجية لفترة تصل إلى عام واحد أو حتى أقوم بإلغاء هذه الموافقة. وأدرك أنه للقيام بأي إجراءات جراحية أو تخدير أو أي علاج متخصص، سيُطلب مني التوقيع على نموذج موافقة إضافية بعد أن يشرح لي الطبيب الإجراء أو العلاج المتخصص.

أعلم أن سدرة للطب مؤسسة تعليمية أكاديمية، ولذا أدرك وأوافق على أن علاجي أو الرعاية التي أتلقاها قد يشارك فيها أخصائيون زائرون وطلبة ومتدربون في البرامج الطبية وبرامج الخدمات الصحية المساعدة، ويشمل أولئك على سبيل المثال لا الحصر المتدربين والمقيمين والزملاء.

الإفصاح عن المعلومات الطبية: تماشياً مع القانون القطري رقم ١٣ لعام ٢٠١٦ بشأن حماية خصوصية البيانات الشخصية، أصرح لسدرة للطب باستخدام والإفصاح عن معلوماتي الصحية الشخصية المحمية ومعلوماتي غير المحددة للهوية (العملية المُستخدَمة في منع ربط معلومات الشخص بهويته). وقد تشمل المعلومات المفصَح عنها، على سبيل المثال لا الحصر، التفاصيل المتعلقة بالعلاج والدفع وإجراءات الرعاية الصحية. وقد تشمل المعلومات المفصَح عنها كذلك المعلومات الضرورية لتسهيل إنجاز المطالبات الطبية ومراجعة المبالغ المالية المدفوعة مقابل خدمات الرعاية الصحية التي أتلقاها أو يتلقاها المريض المذكور أدناه. وأوافق أيضًا على الإفصاح عن أي معلومات إذا اقتضت الحاجة لأغراض رعاية ما بعد الخروج من المستشفى أو النقل إلى المرافق أو المؤسسات الصحية الأخرى حسب تعليماتي أو حسب ما يُلزم به القانون. وأدرك أن لي الحق في إلغاء هذه الموافقة كتابيًا في أي وقت وسيتم بعد ذلك إيقاف جميع عمليات الإفصاح.
إذا كان لديك أسئلة متعلقة باستخدام المعلومات الطبية والإفصاح عنها، يُرجى التواصل مع قسم إدارة المعلومات الصحية(HIM) على عنوان البريد الإلكتروني *him@sidra.org*

FIRST NAME .....
LAST NAME .....
MRN .....
GENDER .....
DOB.....
FIN .....

REV-GCT-11-2019

Page 1 of 3

APPENDIX C2

General Consent for Treatment Page 2

سدرة للطب
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Financial Consent / Assignment of Benefits: I certify that the insurance information given by me/guardian during registration is correct. I authorize Sidra Medicine to bill my insurance company and authorize my insurance company to pay for healthcare services received at Sidra Medicine. I also agree to pay all non-covered charges by my insurance company including deductibles, co-payments, co-insurance and non-covered services. I also understand that my financial status at Sidra Medicine may change due to additional non-covered services being provided by Sidra Medicine at any point of care or treatment and that my financial obligations towards Sidra Medicine may change accordingly.

الموافقة المالية/ تخصيص المستحقات: أقر بأن معلومات التأمين المُزودة من قبلي أو من قبل الوصيّ خلال عملية تسجيلي صحيحة. وأصرح لسدرة للطب بتقديم الفواتير المستحقة الدفع إلى شركة التأمين وأصرح لشركة التأمين بدفع مقابل خدمات الرعاية الصحية التي أتلقاها في سدرة للطب. وأوافق أيضًا على دفع جميع الرسوم والتكاليف التي لا تغطيها شركة التأمين بما في ذلك قيمة الخصومات والمدفوعات المشتركة والتأمين المشترك والخدمات التي لا تُغطيها شركة التأمين. وأدرك أيضًا أن وضعي المالي في سدرة للطب قد يتغير نتيجة الخدمات الإضافية غير المغطاة المقدمة من قبل سدرة للطب في أي وقت من أوقات تلقي الرعاية أو العلاج وأن التزاماتي المالية تجاه سدرة للطب قد تتغير وفقاً لذلك.

Valuables: Valuables should preferably be left at home or removed by the patient's relative/guardian. I further understand and agree that personal property brought into any Sidra Medicine facility is my responsibility and Sidra Medicine is not responsible for any loss or damage.

الممتلكات الثمينة: ينبغي ترك الممتلكات الثمينة في المنزل أو أن يقوم أحد أقرباء المريض/الوصي بإخراجها من سدرة للطب. كما أدرك وأوافق على تحمُّل المسؤولية عن الممتلكات الشخصية التي يتم إحضارها إلى أي من مرافق سدرة للطب ولا يتحمل سدرة للطب أي مسؤولية عن فقد أو تلف أي من الممتلكات الشخصية.

PATIENT / PARENT / LEGAL GUARDIAN
المريض/ولي الأمر/الوصي القانوني

My signature below confirms that:

- I understand the content of this general consent form
- I had an opportunity to ask questions
- I have been informed and provided a copy of my rights and responsibilities
- I understand that it is my responsibility to treat providers, staff, other patients with dignity, honor and respect and to follow the provider's rules and safety regulations.
- I also understand that my financial status could change from non-choice (the service provided at Sidra Medicine is not available in the public healthcare sector in Qatar) to choice (the service provided at Sidra Medicine is available in the public healthcare sector in Qatar) within my care or treatment and it is my responsibility to adhere to the plan of care and discharge, transfer and disposition at Sidra Medicine.
- I acknowledge that my refusal to adhere to Sidra's care plan, transfer (within Sidra Medicine or to other public healthcare facility), or discharge instructions, will result in changing my financial status at Sidra Medicine to choice services, and I will be charged accordingly.

يؤكد توقيعي أدناه:

- أنني أفهم محتوى نموذج الموافقة العامة.
- أنني قد أُتيحت لي الفرصة لطرح الأسئلة.
- أنني قد تم إخباري بحقوقي ومسؤولياتي وحصلت على نسخة منها.
- أدرك أنني مسؤول عن معاملة مقدمي الرعاية والموظفين والمرضى الآخرين بتقدير واحترام، فضلاً عن اتباع قواعد مقدم الرعاية ولوائح السلامة الخاصة به.
- كما أدرك أن وضعي المالي قد يتغير من غير اختياري (الخدمة المقدمة في سدرة للطب غير متوفرة في القطاع الصحي الحكومي في قطر) إلى اختياري (الخدمة المقدمة في سدرة للطب متوفرة في القطاع الصحي الحكومي في قطر) في إطار الرعاية أو العلاج الذي أتلقاه وأنني مسؤول عن الالتزام بخطة سدرة للطب بشأن الرعاية والنقل والتصريح بالخروج من المستشفى.
- أقر بأن رفضي الالتزام بخطة الرعاية أو النقل (من وحدة إلى أخرى داخل سدرة للطب أو إلى إحدى مرافق القطاع العام الصحية خارج سدرة للطب) أو تعليمات الخروج من المستشفى، سوف يؤدي إلى تغيير وضعي المالي في سدرة للطب واعتبار الخدمات الصحية المقدمة خدمات اختيارية مستحقة الدفع.

In compliance with Qatar Council for Healthcare Practitioners (QCHP) Fitness to Practice/Circular No. (2/2015), examination of a minor should be undertaken in the presence of his/her guardian or first degree adult relative: father, mother or both. If a first degree guardian is not available, then examination should be undertaken in the presence of a **second degree adult relative only: brother, sister, grandfather, or grandmother.**

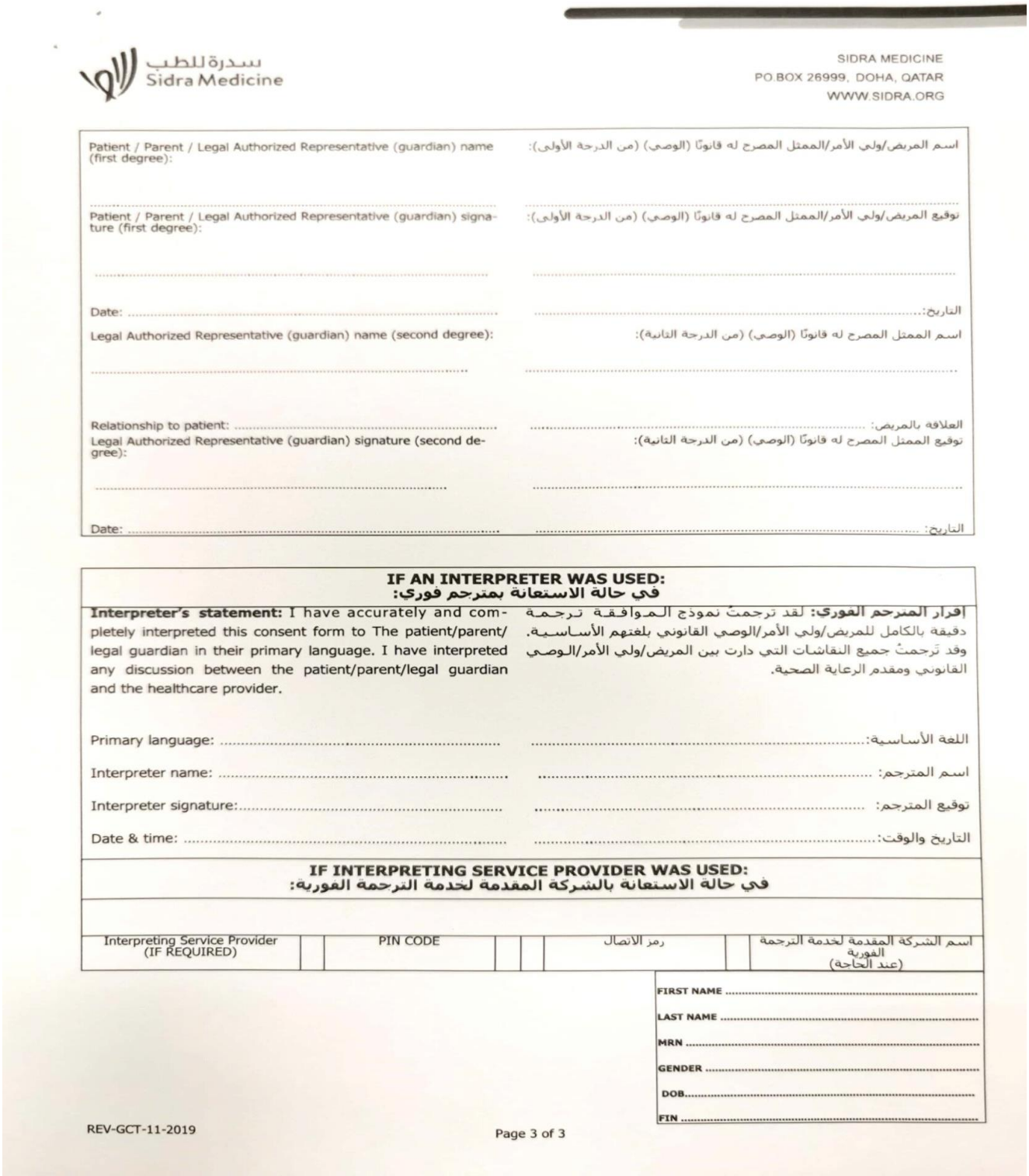
بالامتثال للتعميم رقم (٢/٢٠١٥) الصادر عن إدارة الكفاءة المهنية بالمجلس القطري للتخصصات الصحية، فإن فحص أي قاصر يجب أن يتم في حضور الوصي عليه أو قريب راشد من الدرجة الأولى: الأب، الأم أو كلاهما. وفي حال عدم وجود وصي من الدرجة الأولى، يجب إجراء الفحص في حضور **قريب راشد من الدرجة الثانية فقط: الأخ، الأخت، الجد، أو الجدة.**

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APPENDIX C3

General Consent for Treatment Page 3



سدرة للطب
Sidra Medicine

SIDRA MEDICINE
PO.BOX 26999, DOHA, QATAR
WWW.SIDRA.ORG

Patient / Parent / Legal Authorized Representative (guardian) name (first degree):	اسم المريض/ولي الأمر/الممثل المصرح له قانونًا (الوصي) (من الدرجة الأولى):
Patient / Parent / Legal Authorized Representative (guardian) signature (first degree):	توقيع المريض/ولي الأمر/الممثل المصرح له قانونًا (الوصي) (من الدرجة الأولى):
Date:	التاريخ:
Legal Authorized Representative (guardian) name (second degree):	اسم الممثل المصرح له قانونًا (الوصي) (من الدرجة الثانية):
Relationship to patient:	العلاقه بالمريض:
Legal Authorized Representative (guardian) signature (second degree):	توقيع الممثل المصرح له قانونًا (الوصي) (من الدرجة الثانية):
Date:	التاريخ:

IF AN INTERPRETER WAS USED:
في حالة الاستعانة بمترجم فوري:

Interpreter's statement: I have accurately and completely interpreted this consent form to The patient/parent/ legal guardian in their primary language. I have interpreted any discussion between the patient/parent/legal guardian and the healthcare provider.	**إقرار المترجم الفوري:** لقد ترجمتُ نموذج الموافقة ترجمة دقيقة بالكامل للمريض/ولي الأمر/الوصي القانوني بلغتهم الأساسية. وقد ترجمتُ جميع النقاشات التي دارت بين المريض/ولي الأمر/الوصي القانوني ومقدم الرعاية الصحية.
Primary language:	اللغة الأساسية:
Interpreter name:	اسم المترجم:
Interpreter signature:	توقيع المترجم:
Date & time:	التاريخ والوقت:

IF INTERPRETING SERVICE PROVIDER WAS USED:
في حالة الاستعانة بالشركة المقدمة لخدمة الترجمة الفورية:

Interpreting Service Provider (IF REQUIRED)		PIN CODE			رمز الاتصال		اسم الشركة المقدمة لخدمة الترجمة الفورية (عند الحاجة)

FIRST NAME .....
LAST NAME .....
MRN .....
GENDER .....
DOB.....
FIN .....

REV-GCT-11-2019

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APPENDIX D

IRB Approval letter

Tel: +974-4003-7747
Email: irb@sidra.org

Sidra IRB MOPH Assurance: IRB-A-Sidra-2019-0020
Sidra IRB MOPH Registration: IRB-Sidra-2020-009
Sidra IRB DHHS Assurance: FWA00022378
Sidra IRB DHHS Registration: IRB00009930

April 5, 2023

Approval

Dear Jade Evrille Govender,

On 5 April 2023 the IRB approved the following through 4 April 2024 inclusive.

IRB Number:	1859508
Protocol Title:	Hearing and hearing loss management in children with Down syndrome.
Principal Investigator:	Jade Evrille Govender
Type of review:	Initial Review
Sponsor/ Funding Agency:	None
Grant title and ID, if any:	N/A
Documents reviewed:	• Data Collection - Down Syndrome Research project Data Recording tool.xlsx (UPDATED: 10/5/2022) • Other - Jade E Govender _ Research approval letter.pdf (UPDATED: 10/5/2022) • Proposal - RA-200 Research Project Plan_ Jade Govender Hearing loss management in patients with Down syndrome.docx.pdf (UPDATED: 02/27/2023) • Sidra - IRB Application Form (UPDATED: 10/5/2022) • Training and Credentials
Level of review:	Expedited
Expedited Categories:	5
Pediatric Category:	Research does not involve greater than minimal risk
Informed Consent:	Waiver of informed consent is granted
Approved sample size:	70-80

Before *4 March 2024*, you are to submit a continuing review to request continuing approval or closure. If the IRB does not grant continuing review, approval of this protocol ends after 4 April 2024.

In conducting this study, you are required to follow Sidra's Policies and Procedures pertaining to Human Research Protection.

Tel: +974-4003-7747
Email: irb@sidra.org

Sidra IRB MOPH Assurance: IRB-A-Sidra-2019-0020
Sidra IRB MOPH Registration: IRB-Sidra-2020-009
Sidra IRB DHHS Assurance: FWA00022378
Sidra IRB DHHS Registration: IRB00009930

Other Institutions engaged in this human subject research must secure their IRB approvals and Assurance with MOPH for Protection of Human Subjects Involved in Research.

If you have questions or concerns, please call the IRB office at 4003-7747 or send an email to irb@sidra.org.

Sincerely yours,

Ayman Saleh, MD, MPH, FAAP
Chair
Institutional review Board
Sidra Medicine
t. +974 40036567

APPENDIX E

Ethical Clearance Certificate

UNIVERSITY OF THE WITWATERSRAND, JOHANNESBURG

R49 Ms JE Govender

HUMAN RESEARCH ETHICS COMMITTEE (MEDICAL)
CLEARANCE CERTIFICATE NO. M230964

NAME: Ms JE Govender **DEGREE:** MA
(Principal and Co-Investigators)

DEPARTMENT: School of Human and Community Development
Department of Speech Pathology and Audiology
University

PROJECT TITLE: *Hearing and hearing loss management in children with Down Sydrome in Qatar*

DATE CONSIDERED: 29 September 2023

DECISION: Qpproved unconditionally

CONDITIONS:

NOTE: If contact information regarding student study participants is required, please contact the Registrar's office <Nicoleen.Potgieter@wits.ac.za>

SUPERVISOR: Dr D Pillay; Professor A Kanji; Mr JM Andreas

APPROVED BY:
Professor P Ruff, Chairperson, HREC (Medical)

DATE OF APPROVAL: 12 January 2024 **EXPIRY DATE:** 11 January 2029

This Clearance Certificate is valid for 5 years from the date of approval. An extension may be applied for.

DECLARATION OF INVESTIGATORS

To be completed in duplicate and **ONE COPY** returned to the Research Office secretariat on the 3rd floor, Phillip Tobias Building, Parktown, University of the Witwatersrand, Johannesburg.

I/we fully understand the conditions under which I am/we are authorized to carry out the above-mentioned research and I/we undertake to ensure compliance with these conditions. Should any departure be contemplated from the research protocol as approved, I/we undertake to submit details to the Committee. **I agree to submit a yearly progress report** in the format available at https://wits.ac.za/research/researcher-support/research-ethics/ethics-committees/. This report is due annually, for the duration of the Clearance Certificate, beginning on the first anniversary of Date of Approval above. Unreported changes to the study may invalidate the clearance given by the HREC (Medical).

Signature of Principal Investigator Date

REFERENCES

Abbeduto, L., Warren, S.F. & Conners, F.A. (2007). Language development in Down syndrome: From the prelinguistic period to the acquisition of literacy. *Mental Retardation and Developmental Disabilities Research Reviews*, *13*(3), 247-261. https://doi.org/10.1002/mrdd.20158

Abdul Hadi, K. A., Salahaldin, A., Al Qahtani, A., Al Musleh, Z., Al Sulaitin, M., Bener, A., Chandra, P., & Alawi, F. (2012). Universal neonatal hearing screening: six years of experience in Qatar. *Qatar Medical Journal, 2012(*2), 42.

Al-Aama, J. Y., Alem, H., & El-Harouni, A. A. (2014). Otolaryngologicl issues in Down syndrome patients from Western region of Saudi Arabia. *Life Sci J, 11*, 1.

Almazrou, S. H., Alsubki, L. A., Alsaigh, N. A., Aldhubaib, W. H., & Ghazwani, S. M. (2021). Assessing the Quality of Clinical Practice Guidelines in the Middle East and North Africa (MENA) Region: A Systematic Review. *Journal of Multidisciplinary Healthcare, 14*, 297.

Al-Gazali, L., Hamamy, H., & Al Arrayad, S. (2006). Genetic disorders in the Arab world. *British Medical Journal*, *333*(7573), 831–834. doi:10.1136/bmj.38982.704931

Al Mosawi, A. (2022). Esquirol-Séguin-Down Syndrome Associated with Atrial Septal Defect plus Tricuspid Regurgitation. *Cardiology Research and Reports, 4*, 1-5. 10.31579/2692-9759/033.

Alanazi, M. Q., Albeshr, A. G., Alshahrani, H. A., Aldhawi, S. F., Alhabdan, Y. A., Alghrairy, L. A., ... & Alanazi, S. (2024). The Barriers of Conducting Medical Research in Saudi Arabia: A Review.

Andrade C. (2021). The Inconvenient Truth About Convenience and Purposive Samples. *Indian journal of psychological medicine*, *43*(1), 86–88. https://doi.org/10.1177/0253717620977000

Atqah, A., Bener, A., Hoffmann, G., & Sandridge, A. (2006). The pattern of Down syndrome among children in Qatar: A population-based study. *Birth Defects Research Clinical and Molecular Teratology*, *76*(8), 609–612. doi:10.1002/bdra.20290

Austeng, M. E., Akre, H., Falkenberg, E. S., Øverland, B., Abdelnoor, M., & Kværner, K. J. (2013). Hearing level in children with Down syndrome at the age of eight. *Research in developmental disabilities*, *34*(7), 2251-2256.

Barr, E., Dungworth, J., Hunter, K., Mcfarlane, M., & Kubba, H. (2011). The prevalence of ear, nose and throat disorders in preschool children with Down's syndrome in Glasgow. *Scottish Medical Journal*, *56*(2), 98–103. https://doi.org/10.1258/smj.2011.011036

Basonbul, R. A., Ronner, E. A., Rong, A., Rong, G., & Cohen, M. S. (2020). Audiologic testing in children with Down Syndrome: Are current guidelines optimal?. *International Journal of Pediatric Otorhinolaryngology*, *134*, 110017.

Belletti, D., Zacker, C., & Mullins, C. D. (2010). Perspectives on electronic medical records adoption: electronic medical records (EMR) in outcomes research. *Patient Related Outcome Measures*, 29-37.

Bull, M. J. (2011). Health supervision for children with Down syndrome. *Pediatrics*, *128*(2), 393-406.

Bull, M. (2020). Down Syndrome. *New England Journal of Medicine. 382*, 2344-2352 DOI: 10.1056/NEJMra1706537

Caples, M., Martin, A. M., Dalton, C., Marsh, L., Savage, E., Knafl, G., & Van Riper, M. (2018). Adaptation and resilience in families of individuals with down syndrome living in Ireland. *British Journal of Learning Disabilities, 46*(3), 146-154.

Centre for Arab Genomic Studies. (28 February 2013) Report on Down syndrome in the Arab world. 2013. 5 February 2023 from DownSyndrome-English123.pdf (cags.org.ae)

Centers for Disease Control and Prevention. (16 December 2022). Data and Statistics on Down Syndrome. 5 February 2023 from https://www.cdc.gov/ncbddd/birthdefects/downsyndrome/data.html

Chadha, S., Cieza, A., & Krug, E. (2018). Global hearing health: future directions. *Bulletin of the World Health Organization*, *96*(3), 146.

Chin, C. J., Khami, M. M., & Husein, M. (2014). A general review of the otolaryngologic manifestations of Down Syndrome. *International journal of pediatric otorhinolaryngology*, *78*(6), 899-904.

Ching, T. Y., Crowe, K., Martin, V., Day, J., Mahler, N., Youn, S., ... & Orsini, J. (2010). Language development and everyday functioning of children with hearing loss assessed at 3 years of age. *International journal of speech-language pathology*, *12*(2), 124-131.

Clark, C. M., Patel, H. H., Kanekar, S. G., & Isildak, H. (2017). Enlarged vestibular aqueducts and other inner-ear abnormalities in patients with Down syndrome. *The Journal of Laryngology & Otology*, *131*(4), 298-302.

Committee on Genetics; Health Supervision for Children with Down Syndrome. *Pediatrics* February 2001; *107* (2): 442–449. 10.1542/peds.107.2.442

Corder, J. P., Al Ahbabi, F. J. S., Al Dhaheri, H. S., & Chedid, F. (2017). Demographics and co-occurring conditions in a clinic-based cohort with Down syndrome in the United Arab Emirates. *American Journal of Medical Genetics Part A, 173*(9), 2395-2407.

Davies, B. (1996). Auditory disorders. In B. Stratford and P. Gunn (eds), New Approaches to Down Syndrome (London: Cassell), 100–121.

De Schrijver, L., Topsakal, V., Wojciechowski, M., Van de Heyning, P., & Boudewyns, A. (2019). Prevalence and etiology of sensorineural hearing loss in children with down syndrome: A cross-sectional study. *International journal of pediatric otorhinolaryngology*, *116*, 168-172.

de Graaf, G., Buckley, F., & Skotko, B. G. (2021). Estimation of the number of people with Down syndrome in Europe. *European journal of human genetics*, *29*(3), 402-410.

Earley, M. A., Sher, E. T., & Hill, T. L. (2022). Otolaryngologic Disease in Down syndrome. *Pediatric Clinics*, *69*(2), 381-401.

Elsanadiky, H. H., & Afifi, P. O. (2017). Universal neonatal hearing screening program in private hospital, Qatar. *Tanta Medical Journal, 45*(4), 175.

Fadzil, F., Idris, I. B., Kamal Nor, N., Ismail, J., Mohd Tamil, A., Mohamad Noh, K., ... & Ismail, R. (2021). Missed appointments at a child development centre and barriers to access special needs services for children in Klang Valley, Malaysia: A mixed methods study. *International Journal of Environmental Research and Public Health, 19*(1), 325.

Fayed, A. A., Wahabi, H., Mamdouh, H., Kotb, R., & Esmaeil, S. (2017). Demographic profile and pregnancy outcomes of adolescents and older mothers in Saudi Arabia: analysis from Riyadh Mother (RAHMA) and Baby cohort study. *BMJ open*, 7(9), e016501.

Fidler, D. J., Needham, A. W., & Schworer, E. (2019). Infant foundations in Down syndrome: Early constraints on cognitive skill acquisition. *International Review of Research in Developmental Disabilities*, *56*, 41-65.

Fitzgibbons, E. J., Driscoll, C., Traves, L., & Beswick, R. (2024). Detecting Hearing Loss Through Targeted Surveillance: Risk Registry and Surveillance Timeframe Recommendations. *Journal of Speech, Language, and Hearing Research,* 1-16.

Fortnum, H., Leighton, P., Smith, M. D., Brown, L., Jones, M., Benton, C., ... & Sutton, K. (2014). Assessment of the feasibility and clinical value of further research to evaluate the management options for children with Down syndrome and otitis media with effusion: a feasibility study. *Health Technology Assessment (Winchester, England), 18*(60), 1.

Ganek, H. V., Madubueze, A., Merritt, C. E., & Bhutta, Z. A. (2023). Prevalence of hearing loss in children living in low-and middle-income countries over the last 10 years: A systematic review. *Developmental Medicine & Child Neurology, 65*(5), 600-610.

Gearing, R. E., Mian, I. A., Barber, J., & Ickowicz, A. (2006). A methodology for conducting retrospective chart review research in child and adolescent psychiatry. *Journal of the Canadian Academy of Child and Adolescent Psychiatry*, *15*(3), 126.

Ghadersohi, S., Bhushan, B., & Billings, K. R. (2018). Challenges and outcomes of cholesteatoma management in children with Down syndrome. *International journal of pediatric otorhinolaryngology, 106*, 80-84.

Girotto, G., Mezzavilla, M., Abdulhadi, K., Vuckovic, D., Vozzi, D., Alkowari, M. K., ... & Badii, R. (2014). Consanguinity and hereditary hearing loss in Qatar. *Human Heredity*, *77*(1-4), 175-182.

Graydon, K., Waterworth, C., Miller, H., & Gunasekera, H. (2019). Global burden of hearing impairment and ear disease. *The Journal of Laryngology & Otology, 133*(1), 18-25. doi:10.1017/S0022215118001275

Hamamy, H., & Bittles, A. H. (2008). Genetic clinics in Arab communities: meeting individual, family and community needs. *Public Health Genomics*, 12(1), 30-40.

Harlor Jr, A. D. B., Bower, C., Committee on Practice and Ambulatory Medicine, & Section on Otolaryngology–Head and Neck Surgery. (2009). Hearing assessment in infants and children: recommendations beyond neonatal screening. *Pediatrics, 124*(4), 1252-1263.

Hart, O., & Yoost, J. L. (2025). Retrospective Chart Reviews: A Practical Approach to Designing, Collecting Data, and Writing the Manuscript. *Marshall Journal of Medicine*, *11*(1), 5.

Hickey, F., Hickey, E., & Summar, K. L. (2012). Medical update for children with Down syndrome for the pediatrician and family practitioner. *Advances in pediatrics, 59*(1), 137-157.

Holcomb, Alayna Marissa. "Oral Health Education and Dental Care Access of Families of Children with Down Syndrome in New Mexico." (2024).

Jawhari, B., Ludwick, D., Keenan, L., Zakus, D., & Hayward, R. (2016). Benefits and challenges of EMR implementations in low resource settings: a state-of-the-art review. *BMC medical informatics and decision making, 16*(1), 1-12.

Joint Committee on Infant Hearing. (2007). Year 2007 position statement: Principles and guidelines for early hearing detection and intervention programs. *Pediatrics, 120*(4), 898-921.

Junod, V & Elger, B., 2010. Retrospective research: what are the ethical and legal requirements? *Swiss Medical Weekly*. *140*, 13041

Kazemi, M., Salehi, M., & Kheirollahi, M. (2016). Down syndrome: current status, challenges and future perspectives. *International journal of molecular and cellular medicine, 5*(3), 125

Kennedy, C. R., McCann, D. C., Campbell, M. J., Law, C. M., Mullee, M., Petrou, S., ... & Stevenson, J. (2006). Language ability after early detection of permanent childhood hearing impairment. *New England Journal of Medicine*, *354*(20), 2131-2141.

Khalid-Raja, M., & Tzifa, K. (2016). Current demand of paediatric otolaryngology input for children with Down's syndrome in a tertiary referral centre. *The Journal of Laryngology & Otology, 130*(11), 995-1000. doi:10.1017/S0022215116008963

Kovaleva, N. V. (2002). Sex ratio in Down syndrome. *Tsitologiia i genetika, 36*(6), 54-69.

Kreicher, K. L., Weir, F. W., Nguyen, S. A., & Meyer, T. A. (2018). Characteristics and progression of hearing loss in children with Down syndrome. *The Journal of Pediatrics*, *193*, 27-33.

Lagan NC, Mc Grane F, Huggard D, et alImplementation of a health surveillance clinic for children with Down syndrome. *Archives of Disease in Childhood - Education and Practice* 2021;106:60-62.

Lau, W. L., Ko, C. H., & Cheng, W. W. (2015). Prevalence and parental awareness of hearing loss in children with Down syndrome. *Chinese Medical Journal*, *128*(08), 1091-1095.

Laws, G., & Hall, A. (2014). Early hearing loss and language abilities in children with Down syndrome. *International journal of language & communication disorders, 49*(3), 333-342.

Lehman, A., Leach, M., & Santoro, S. L. (2021). Delivering a new diagnosis of Down syndrome: Parent experience. *American Journal of Medical Genetics Part A, 185A*, 3615– 3622. https://doi.org/10.1002/ajmg.a.62408

Livingstone-Sinclair, K., Scott, C., & Trotman, H. (2018). Adherence to health management guidelines for children with Down's syndrome at the Bustamante Hospital for Children, Jamaica. *Tropical Doctor, 48*(4), 301-305.

Iulita, M. F., Chavez, D. G., Christensen, M. K., Tamayo, N. V., Plana-Ripoll, O., Rasmussen, S. A., ... & Fortea, J. (2022). Association of Alzheimer disease with life expectancy in people with Down syndrome. *JAMA network open*, *5*(5), e2212910-e2212910.

Määttä, T., Määttä, J., Tervo-Määttä, T., Taanila, A., Kaski, M., & Iivanainen, M. (2011). Healthcare and guidelines: a population-based survey of recorded medical problems and health surveillance for people with Down syndrome. *Journal of Intellectual and Developmental Disability, 36*(2), 118-126.

Malt, E. A., Dahl, R. C., Haugsand, T. M., Ulvestad, I. H., Emilsen, N. M., Hansen, B., ... & Davidsen, E. M. M. (2013). Health and disease in adults with Down syndrome. *Tidsskrift for Den norske legeforening.*

Martin, G. E., Klusek, J., Estigarribia, B., & Roberts, J. E. (2009). Language Characteristics of Individuals with Down Syndrome. *Topics in language disorders, 29*(2), 112–132. https://doi.org/10.1097/tld.0b013e3181a71fe1

Matkin, N. D., & Wilcox, A. M. (1999). Considerations in the education of children with hearing loss. *Pediatric Clinics of North America, 46*(1), 143-152.

McDaid, D., Park, A. L., & Chadha, S. (2021). Estimating the global costs of hearing loss. *International journal of audiology, 60*(3), 162–170. https://doi.org/10.1080/14992027.2021.1883197

McDermott, A. L., Williams, J., Kuo, M. J., Reid, A. P., & Proops, D. W. (2008). The role of bone anchored hearing aids in children with Down syndrome. *International journal of pediatric otorhinolaryngology*, *72*(6), 751-757.

Mengoni, S. E., & Redman, S. (2020). Health monitoring of young children with Down syndrome: A parent-report study. *British Journal of Learning Disabilities, 48*(1), 10-18.

Mitchell, R. B., Call, E., & Kelly, J. (2003). Ear, nose and throat disorders in children with Down syndrome. *The Laryngoscope*, *113*(2), 259-263.

Moeller, M. P. (2000). Early intervention and language development in children who are deaf and hard of hearing. *Pediatrics*, *106*(3). Available at: www.pediatrics.org/cgi/content/full/106/3/e43.sos

Newberger, D. S. (2000). Down syndrome: prenatal risk assessment and diagnosis. *American family physician,* 62(4), 825-832.

Nightengale, E. (2018). Hearing loss in children with Down syndrome. *The Hearing Journal, 71*(2), 10-12.

Nightengale, E. E., Wolter-Warmerdam, K., Yoon, P. J., Daniels, D., & Hickey, F. (2020). Behavioral audiology procedures in children with Down syndrome. *American journal of audiology, 29*(3), 356-364.

Omar, M., McCoy, J. L., McCormick, A. A., Vellody, K., & Chi, D. H. (2021). Repeat tympanostomy tubes in children with *Down syndrome. International Journal of Pediatric Otorhinolaryngology, 148*, 110811.

Park, A. H., Wilson, M. A., Stevens, P. T., Harward, R., & Hohler, N. (2012). Identification of Hearing Loss in Pediatric Patients with Down Syndrome. *Otolaryngology–Head and Neck Surgery*, *146*(1), 135–140. https://doi.org/10.1177/0194599811425156

Patel, T. A., Nguyen, S. A., & White, D. R. (2018). Down syndrome as an indicator for pediatric otolaryngologic procedures. *International journal of pediatric otorhinolaryngology*, *112*, 182-187.

Paulson, L. M., Weaver, T. S., & Macarthur, C. J. (2014). Outcomes of tympanostomy tube placement in children with Down syndrome—a retrospective review. *International Journal of Pediatric Otorhinolaryngology, 78*(2), 223-226.

Porter, H., Buss, E., Merchant, G. R., & Leibold, L. J. (2022). Observational Study to Preliminarily Characterize the Audiological Profile of Children with Down Syndrome. *Journal of Speech, Language, and Hearing Research*, *65*(11), 4498-4506.

Porter, A., Sheeran, N., Hood, M., & Creed, P. (2021). Decision-making following identification of an infant's unilateral hearing loss: Parent and professional perspectives. *International journal of pediatric otorhinolaryngology*, *148*, 110822.

Ramia, M., Musharrafieh, U., Khaddage, W., & Sabri, A. (2014). Revisiting Down syndrome from the ENT perspective: review of literature and recommendations. *European Archives of Oto-Rhino-Laryngology*, *271*, 863-869.

Ray, A., Oliver, T. R., Halder, P., Pal, U., Sarkar, S., Dutta, S., & Ghosh, S. (2018). Risk of Down syndrome birth: Consanguineous marriage is associated with maternal meiosis-II nondisjunction at younger age and without any detectable recombination error. *American Journal of Medical Genetics Part A*, *176*(11), 2342-2349.

Raut, P., Sriram, B., Yeoh, A., Hee, K. Y., Lim, S. B., & Daniel, M. L. (2011). High prevalence of hearing loss in Down syndrome at first year of life. *Annals of the Academy of Medicine-Singapore*, *40*(11), 493.

Rezayat, A. A., Nazarabadi, M. H., Andalibi, M. S., Ardabili, H. M., Shokri, M., Mirzaie, S., & Jarahi, L. (2013). Down syndrome and consanguinity. *Journal of research in medical sciences: the official journal of Isfahan University of Medical Sciences*, *18*(11), 995–997.

Riehle-Colarusso, T., & Oster, M. E. (2016). Down syndrome: changing cardiac phenotype?. *Pediatrics*, *138*(1).

Roberts, J. E., Price, J., & Malkin, C. (2007). Language and communication development in Down syndrome. *Mental retardation and developmental disabilities research reviews*, *13*(1), 26-35.

Roizen, N. (1997). Hearing loss in children with Down syndrome: a review. *Down Syndrome Quarterly*, *2*, 1–4.

Roizen, N. J., Magyar, C. I., Kuschner, E. S., Sulkes, S.B., Druschel, C., van Wijngaarden, E., Rodgers, L. Diehl, A., Lowry, R., & Hyman, S. L. (2014). A Community Cross-Sectional Survey of Medical Problems in 440 Children with Down Syndrome in New York State. *The Journal of Pediatrics*. *16*(4) 871-875, https://doi.org/10.1016/j.jpeds.2013.11.032.

Roper, R. J., & Reeves, R. H. (2006). Understanding the basis for Down syndrome phenotypes. *PLoS genetics*, 2(3), e50.

Sabrinna Silva Resende , A., Machado Miranda Leal Barbosa , S., Luísa Xavier Carvalho Andrade , I., Manoel Oliveira Costa , L., Moura Melo, S., Holanda Moura , E., Vinicius de Carvalho Souza , M., Andrade da Silva Junior , E., de Assis de Brito , R., & Folha Gois Moreira , K. (2023). Characterization of the manifestations of Down Syndrome in Brazil between 2016 and 2020: an epidemiological study. Seven Editora. Retrieved from http://sevenpublicacoes.com.br/index.php/editora/article/view/2000

Sait, S., Alamoudi, S., & Zawawi, F. (2022). Management outcomes of otitis media with effusion in children with down syndrome: A systematic review. *International Journal of Pediatric Otorhinolaryngology*, 111092.

Santoro, S. L., Howe, Y. J., Krell, K., Skotko, B. G., & Co, J. P. T. (2023). Health surveillance in a Down syndrome specialty clinic: implementation of electronic health record integrations during the coronavirus disease 2019 pandemic. *The Journal of Pediatrics, 255,* 58-64

Sanyelbhaa, H., Kabel, A., El, H. A. E. R. A., Sanyelbhaa, A., & Salem, H. (2017). The risk ratio for development of hereditary sensorineural hearing loss in consanguineous marriage offspring. *International journal of pediatric otorhinolaryngology*, *101*, 7-10.

Setia M. S. (2016). Methodology Series Module 1: Cohort Studies. *Indian journal of dermatology, 61*(1), 21–25. https://doi.org/10.4103/0019-5154.174011

Schworer, E. K., Ahmed, A., Hogenkamp, L., Moore, S., & Esbensen, A. J. (2022). Associations among co-occurring medical conditions and cognition, language, and behavior in Down syndrome. *Research in Developmental Disabilities, 126,* 104236.

Shearer, A. E., Hildebrand, M. S., & Smith, R. J. (2017). Hereditary hearing loss and deafness overview. GeneReviews®[Internet].

Sheehan, P. Z., & Hans, P. S. (2006). UK and Ireland experience of bone anchored hearing aids (BAHA®) in individuals with Down syndrome. *International journal of pediatric otorhinolaryngology, 70*(6), 981-986.

Sheffield, A. M., & Smith, R. J. H. (2019). The Epidemiology of Deafness. *Cold Spring Harbor perspectives in medicine, 9*(9), a033258. https://doi.org/10.1101/cshperspect.a033258

Shott, S. R. (2006) Down syndrome: common otolaryngologic manifestations. *American Journal Medical Genetics Part C: Seminars in Medical Genetics*, *142C*, 131-140

Shott, S. R., & Donnelly, L. F. (2004). Cine magnetic resonance imaging: evaluation of persistent airway obstruction after tonsil and adenoidectomy in children with Down syndrome. *The Laryngoscope, 114*(10), 1724-1729.

Shott, S. R., Joseph, A., & Heithaus, D. (2001). Hearing loss in children with Down syndrome. *International journal of pediatric otorhinolaryngology*, *61*(3), 199-205.

Sidell, D., Hunter, L. L., Lin, L., & Arjmand, E. (2014). Risk factors for preoperative and postoperative hearing loss in children undergoing pressure equalization tube placement. *Otolaryngology-Head and Neck Surgery, 150*(6), 1048-1055.

Sidenna, M., Fadl, T., & Zayed, H. (2020). Genetic epidemiology of hearing loss in the 22 Arab Countries: a systematic review. *Otology & Neurotology, 41*(2), e152-e162.

Sijeeni, A. S. M. (2016). *Understanding the experiences of mothers caring for children with down syndrome in Saudi Arabia* (Doctoral dissertation, Queensland University of Technology).

Silverman, W. (2007). Down syndrome: cognitive phenotype. *Mental retardation and developmental disabilities research reviews*, *13*(3), 228-236.

Stoel-Gammon C. Down syndrome. Effects on language development. ASHA. 1990 Sep 32(9):42-44. PMID: 2145841.

Tedeschi, A. S., Roizen, N. J., Taylor, H. G., Murray, G., Curtis, C. A., & Parikh, A. S. (2015). The prevalence of congenital hearing loss in neonates with Down syndrome. *The Journal of pediatrics, 166*(1), 168-171.

Tsou, A. Y., Bulova, P., Capone, G., Chicoine, B., Gelaro, B., Harville, T. O., ... & Whitten, M. S. (2020). Medical care of adults with Down syndrome: a clinical guideline. *Jama, 324*(15), 1543-1556.

Tucci, D. L., Merson, M. H., & Wilson, B. S. (2010). A summary of the literature on global hearing impairment: current status and priorities for action. *Otology & Neurotology*, *31*(1), 31-41.

Tweedie, D. J., Bajaj, Y., Ifeacho, S. N., Jonas, N. E., Jephson, C. G., Cochrane, L. A., ... & Wyatt, M. E. (2012). Peri-operative complications after adenotonsillectomy in a UK pediatric tertiary referral centre. *International journal of pediatric otorhinolaryngology, 76*(6), 809-815.

United Nations (2022, March), *World Down Syndrome Day, March 21*. https://www.un.org/en/observances/down-syndrome-day

van den Driessen Mareeuw, F. A., Coppus, A. M. W., Delnoij, D. M. J., & de Vries, E. (2020). Quality of health care according to people with Down syndrome, their parents and support staff-A

qualitative exploration. *Journal of applied research in intellectual disabilities : JARID*, *33*(3), 496–514. https://doi.org/10.1111/jar.12692

Wahab, A., Bener, A., & Teebi, A. S. (2006). The incidence patterns of Down syndrome in Qatar. Clinical Genetic Journal, *69*(4), 360–362. doi:10.1111/j.1399-0004.2006.00593.

World Health Organization. (2021). *World report on hearing. World Health Organization.* https://apps.who.int/iris/handle/10665/339913. License: CC BY-NC-SA 3.0 IGO

World Health Organization. (2022) Deafness and Hearing Loss. Retrieved January 2, 2023, from https://www.who.int/news-room/fact-sheets/detail/deafness-and-hearing-loss

World Health Organization. (2023) World Hearing Day 2023: Ear and hearing care for all! Let's make it a reality. Retrieved 15 October, 2023, from https://www.who.int/europe/news-room/events/item/2023/03/03/default-calendar/world-hearing-day-2023--ear-and-hearing-care-for-all!-let-s-make-it-a-reality

World Health Organization Regional Office for the Eastern Mediterranean (2013). *Summary Report on the Regional workshop on strengthening and integrating the ear and hearing care programme within primary health care and health systems* (Report No WHO-EM/CPB/017/E). IC_Meet_Rep_2013_EN_15190.pdf (who.int)

Wiseman, F. K., Alford, K. A., Tybulewicz, V. L., & Fisher, E. M. (2009). Down syndrome—recent progress and future prospects. *Human molecular genetics*, *18*(R1), R75-R83.

Yoshinaga-Itano, C., Sedey, A. L., Wiggin, M., & Chung, W. (2017). Early hearing detection and vocabulary of children with hearing loss. *Pediatrics, 140*(2).

Yüksel, F., & Uğur, C. (2021). Analysis of Newborn Hearing Screening Test Results of Children with Down Syndrome. *Turkish Archives of Pediatrics*, *56*(5), 458.