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Masters Research Report

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29-09-2018

A Sociological Analyses of the lived realities and Narratives of Women diagnosed and living with Turner Syndrome and how they conceptualise their gendered identities'

By

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Declaration

I declare that this report is my own work and has not been submitted before for any degree or examination in any other University.

Nadine van Jaarsveld

27 September 2018

Abstract

This study focuses on the psychological and social aspects that affect those diagnosed with Turners Syndrome. It analyses the narratives of these women, attending to how they negotiate and develop their own identities with particular emphasis being placed on their gendered identities and what it means to be living with a genetic condition and the implications thereof. By focusing on the narratives of these women I will explore the different factors and aspects that affect one's identity that we so often take for granted. I consider factors such as one's early childhood development as well as interpersonal relationships and the process of diagnosis. This argument is exemplified by the different aspects associated with Turners Syndrome specifically how these women develop their identities in relation to aspects of the syndrome such as infertility.

Dedication and Acknowledgments

No measure of gratitude could ever fully express how humbled and thankful I am for being given the opportunity to do a project that is so close to my heart. Every word typed and every sentence read was a truly enriching experience and I can honestly say that this project was a fulfilling journey from beginning to end.

To the Lord almighty for blessing me abundantly with so much love and support as well as the strength and courage throughout this project.

To my parents Coenie van Jaarsveld and Monique van Jaarsveld for taking every step with me and being right there by my side from day one, I could not have asked for better parents! You truly make the world a better place by just being in it.

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For the Women living with Turner Syndrome this one is for you!

Introduction

I have always been intrigued by this notion of the *feminine identity* and what we as a society define as a *feminine Identity*. The way in which we negotiate and conceptualise our identities is truly one of the most complex and fascinating social processes that has so many nuances and intricacies. One's identity works and operates on so many different levels.

My research looked at Turners Syndrome, a rare genetic condition, in which women are not necessarily fitting the biological profile of what is considered to be genetically female; however are still classified and socialised as women. When you initially approach someone, and ask them if they have heard about Turners Syndrome the usual response is either a simple 'NO' or a very blank stare. Truth be told in a world that is so medically advanced we actually know very little about Turners Syndrome, thus it is a condition that is both underdiagnosed, underreported as well as under researched. This is a clinical syndrome discovered in 1938 and results in a failure of secondary sexual characteristics and developments at puberty. Some of the more noticeable characteristics of Turners Syndrome include a small stature, webbed neck, congenital heart-disease as well as some renal abnormalities as written in Gravholt & Novo Nordisk, (2009).

Infertility is a defining symptom associated with Turners Syndrome since these women do not naturally produce any eggs in the ovaries or produce any of the necessary growth hormones. They are then considered to be/classified as infertile which is something that is potentially devastating for most women. This facet of infertility is something that will most definitely affect the way in which women with Turners Syndrome will view themselves in relation to other women as well as society, hence why the analysis of their narratives will give valuable insight into this journey these women take to define and redefine themselves.

Regardless of the fact that Turners Syndrome is not by any means life threatening it is however a very real condition that affects these women in a very tangible manner. These women are subjected to a lengthy process of diagnosis which involves numerous blood tests and treatments such as hormone replacement therapy. On further research, what alarmed me was how sparse the information on Turners Syndrome was. Currently we have the scientific journals and biological reports that provide us with information however no tangible research has been done from the perspective of those living with Turners Syndrome. I found myself

asking why is there so little research being done on Turners Syndrome? And where are the narratives of the affected women?

What defines me as a woman? How is a *feminine identity* negotiated within contemporary society? The *feminine identity* as we know it is not as simplistic as many perceive it to be especially when contextualized in the broader context of society. Embedded within the identity of women is the argument of biology and physiology, the physical characteristics which makes one a woman. From another perspective, the identity of women is also deeply entrenched within society and how society teaches us about the expectations and behaviours of women, thus defining their position within society. One such expectation of women is that of maternity and motherhood, because women can bear children it is therefore expected of them to become mothers thus femininity and maternity are equated into the same /ideal of what constitutes a woman.

This research is based on the premise that women are biologically capable of bearing children thus creating this social expectation of women becoming mothers which is actually a social role. Embedding the biological aspects of maternity into the socially created feminine identity. This is just one example of how we have constructed a feminine identity based on the biology.

In chapter one of this report I covered the fundamental aspects that drove this research project such as the aims and rationale. I identified my primary aim as wanting to contribute to research being done on Turners Syndrome and to demonstrate how important it is to include patient narratives and what they can contribute to research. The aim of this project was largely based on my own story and my own personal experiences with Turners Syndrome and using this to develop some of the more pertinent questions that arose while doing this project. While doing my own research on Turner's Syndrome I identified a potential research question when I considered what information was available and then saw that in context of the type of information I needed as both a patient living with Turners Syndrome as well as a researcher which developed into the rationale of this project. I chose to focus on the narratives of patients as a source of information to include a different perspective.

In chapter two, I wrote about the more practical elements of this research project. It is important to note that this project was qualitative in nature thus it was centred around the information gathered being as detailed and informative as possible.

Keeping the qualitative aspect in mind this research project consists of two major elements namely the narratives of each participant as well as my own narrative that I have incorporated throughout this project. To gather the narratives of the participants I have chosen to use interviews as this was the best means of collecting detailed narratives and really delve into the thoughts and experiences of the participants.

Chapter three is the literature review in which I began by covering literature on Turners Syndrome which included the initial report by Dr Henry Turner who discovered Turners Syndrome in 1968. The other literature covered includes reports and studies done by various medical professionals. What I aimed to do by including this literature was to create some context to the project by covering the basics of what Turners Syndrome is thus giving the reader some needed insight into the condition.

Once I had covered the literature on Turners Syndrome I shifted my focus to literature that discussed the different aspects of *sex* and *gender* and the different arguments pertaining to these two seemingly stable parts of our identities as human beings. And finally, the last part of the literature review centred around literature done on narratives and the different aspects of using narratives as an integral and very important research tool as well as the importance of these narratives and what they can contribute to research. What I had to be cognisant of was the fact that the narratives of the participants centred around their experiences as women, as well as their experiences as patients going through the process of diagnosis and treatment.

In Chapter four, I recorded my research findings and what transpired during the interviews with five participants. For this section, I divided the content of the interviews into five major categories which were early childhood and development, diagnosis and treatment, living with Turner's Syndrome, Turner's Syndrome and love, issues around gender and infertility and finally my last two sections focus briefly on support and my role as both patient and researcher. I developed my interview questions in relation to my own experiences with Turners Syndrome thus these categories are a reflection of my own narrative as well as the stories told by my participants. Each category also links to different stages of the participants' life and certain moments in their lives were being diagnosed with Turners Syndrome played a role and how they understand their condition from a patient's perspective.

Chapter 1:

Aim and Rationale

The core aim of this project was to raise awareness about Turners Syndrome and to add to current research by focusing on the construction of a gender identity particularly amongst women living with Turners Syndrome. I also aimed to show how important the participants' narratives are and what they can contribute particularly to our understanding of complex social constructs such as *gender*. My aim was also to provide future researchers and Turners Syndrome patients with balanced information about Turners Syndrome that includes the empirical facts and subjective narratives as a means of gaining a more holistic and deeper understanding of the condition. By furthering the study on Turners Syndrome my hope was that we start using words such as 'condition' or 'syndrome' when referring to Turners Syndrome since this in not by any means a disability or defect.

Problem Statement

As researchers doing empirical work on Turners Syndrome we rely on the knowledge of experts as well as genetic counsellors as a means of guiding us, thus patients diagnosed with the condition also rely on this very same information to understand the condition. It follows then that for patients and researchers the initial understanding of Turners Syndrome is highly medicalised. However, this is where biology has its limitation when referring to issues of *gender*. In the context of this study I understand *gender* to be referring to one's masculinity or femininity which includes the social and cultural elements of the male and female body. However we are now at a time in society where one needs to discuss *gender* in a more dynamic way and thus expand our understanding of *gender* as a concept and begin to include for example a discussion on intersexed bodies that have actually disrupted the normative masculine and feminine binary conceptions of *gender*.

We assume that one's *gender identity* is necessarily grounded in biology; however, we know that this is in fact not the case because *gender* as a construct of identity is far more complex. For instance, from the perspective of medical theory and physiology, the body is made up of genes, chromosomes as well as DNA. This also includes the anatomical parts which are used

to construct the phenotypical or normative male or female body. Once society recognises and accepts these definitions of what is considered genetically male and female this then extends to how we structure our identities, adjust our behaviour. Subsequent to the biological conceptions of *gender* ,society uses biology and a means of justifying the position of men and women within that society .Staying with this argument that is largely premised on the dominant medical discourse and biological science we have developed very specific male and female characteristics which leaves no room for any grey areas or different types of bodies that may disrupt or challenge the way in which biological sex is identified and how we then define and understand our *gendered identities*.

How would this genetic condition affect their understanding of themselves as women*?* There is something very unique about the position these women hold in relation to their *feminine identities* based on the fact that they do not meet the biological prerequisites of what is to be considered biologically female, yet they are socialised into a feminine role. The use of narratives will help us understand the complex process of developing a *gendered identity* which is precisely what makes Turners Syndrome a fascinating and intriguing case. The argument is that these women have a very weak, and in some instances, no biological base from which they can conceptualize an understanding of their bodies and their gendered identity. There is this constant tension between trying to understand their biological *sex* while still assuming the prescribed female *gender* role which does change at different stages in their lives when one considers factors such as age and the knowledge they have about their condition.

For this project, I will argue that such issues of *gender identity* should be understood from within the context of the experiences of my participants and the way in which they understand their condition. Thus, we need to analyse the more social and psychological underpinnings of how these women formulated their identities and understood their place in society and how these changes and develop at different stages of one's life. In order to understand how women affected by the syndrome construct their *gender identities* is imperative to attend to patients' narratives.

The reference to a *patient narrative* is based on the fact these women live with the diagnosis of a medical and genetic condition and it is from this medical condition that their narratives emerge. The narrative approach provides a window into gaining understanding of what we

know little about, that is the psychosocial dimensions of the experience of living with Turners Syndrome. In addition, by exploring process of medicalisation of the condition I hope to understand how different aspects of one's identity work in relation to a diagnosis and current medical knowledge with particular emphasis being placed on Turners Syndrome.

However, what seems to be lacking in the literature from your medical journals and textbooks are the psychosocial effects and implications of such genetic conditions. It would be interesting to see how my respondents feel about the hormone therapy and what effect it has had on their lives. In relation to the idea of narratives one must question whether the participants will be speaking from the perspective of a patient within the context of therapy and how this has impacted on the development of their identities. What role would clinical therapy or treatment play in a narrative of these participants? This just reiterates the notion of needing narratives that give us the patients' perspective, as well as the psychological and social nuances and effects of hormone therapy.

Rationale:

What I found most alarming is that no empirical research has been done from the patients' perspective. I found myself asking why is there so little research being done on Turners Syndrome? And where are the narratives of these women? This is an area study which needs more research, specifically on how women –at various stages of their lives- diagnosed with the syndrome understand and conceptualize their *gendered identities*.

A major gap in the literature on Turners Syndrome is a framework that links two often opposing fields of research, the social construction of *gender* and the biological definitions of *sex*. The potential of using patients' narratives is that it offers a practical way of viewing how one's biology and society interact. This highlights how gendered identity is developed and forms part of a process that involves multiple facets and elements.

We must also look beyond the confines of medical and physiological discourses because as much as medical knowledge is useful and informative it can also be very limiting and restrictive, especially when we consider woman with a genetic condition who is trying to grasp the magnitude or severity of her condition. Thus, this notion of the socially created, normative and *gendered identity* should be grasped and analysed from within the narratives

of these women diagnosed with Turners Syndrome. This would essentially give us, researchers, a more concise and accurate way of analysing the way in which one constructs an identity that includes both the objective truths about the condition as well as consider the subjective story and journey of the individual.

A major component of this research is based on the fact that I myself have Turners Syndrome which in this instance makes me both a participant as well as a researcher. My idea was to one day, broaden the scope for research done on Turners Syndrome so that we don't just analyse such conditions from a purely biological perspective but that we can integrate how we study genetic conditions and include patients' narratives.

At the age of sixteen I was diagnosed with a rare genetic condition known as Turners Syndrome. I distinctly remember walking into the gynaecologists consulting room and almost immediately was asked if I had ever been tested for something known as Turners Syndrome and without much hesitation I went for blood tests immediately. The gynaecologist very briefly explained what Turners Syndrome was and mentioned that I had a missing x chromosome which resulted in infertility which was the extent of the knowledge provided. It was my mom and dad who actually did further research on this condition that really helped me make sense of what was going on with my body. I had made peace with my diagnosis until my third year at varsity when I was really started questioning my own feminine identity and femininity. My personal journey with Turners Syndrome as well as all the other brave women living with the condition helped me realise that there is more to one's identity than meets the eye. What I can add here is that questioning my own feminine identity brought me out of my comfort zone and really challenged me to think differently about how I viewed my body. What makes this study on Turner's Syndrome unique is the perspective these women can offer when we include their narratives and experiences in this study.

With regards to their *gendered identity*, these women provide a very interesting position and narrative about how they have come to understand themselves and their place within society with particular reference to how their condition challenges normative conceptions of we understand as *gender*. What I also had to consider in this study was a lifetime perspective as some of the participants were diagnosed at birth with Turners Syndrome while others were only diagnosed much later on in life.

I inquired about the impact this has had their understanding and conceptualizing of their gendered identity. What this study would contribute is a more dynamic understanding of how we understand the concept of *gender* by using the narratives of women with Turners Syndrome as an example of how not all bodies fit the very strict categories we as a society have developed.

My contribution here would be to produce knowledge on the experience of a group of individuals who share similar diagnosis; prior to my study the participants had no point of reference from other women living with the same condition. Where I can extend on the study done by Matebese from the University of Cape Town in 2008. Matebese focused broadly on the psychosocial elements of Turners Syndrome whereas I wanted to go into more detail and focus on how Turners Syndrome patients dealt with interpersonal relationships, marital, sexual relationships and infertility, how they coped at school as well as the support and coping mechanisms they used or that was offered to them.

These narratives can supplement the already dense bio-medical descriptions of Turners Syndrome and focus more on the information directly from the .A final element of my rationale would be to argue that the cultural and societal dynamics here in South Africa will contextually add a very different perspective on issues such as *gender* and how this is influenced by culture thus the experience of living with a genetic condition. To have a project focused here in South Africa will indeed be unique for local patients as well as those researching the condition.

Objectives:

A general objective is to critically assess, through the narratives of the participants, the different aspects of how a *gendered identity* is constructed and re-negotiated and discuss this with reference to Turners Syndrome.

The specific objectives of my study are

1. To gather the narratives of 5 women diagnosed with Turners Syndrome at different stages of their lives,
2. To focus on how these women have (re) constructed their *gender identity* vis a vis a medical diagnosis.

Chapter2:

Methodology

At the very heart of the Qualitative research method is this idea of gaining a deeper understanding of a specific population according to Denzin and Lincoln, (2013). What Qualitative research does is use multiple disciplines as a means of conducting research on various subject matter. It also provides us with data that shows the interconnections and relations between humans' social settings and theoretical concepts which was also mentioned by Denzin and Lincoln, (2013). Qualitative research is based on the interpretation of meaning that emerges from the participants of the research project and how these meanings link to certain discourses which all forms a part of the qualitative method and process. There is thus no set theory or paradigm and it does not belong to specific discipline as understood by Denzin and Lincoln, (2013). In terms of design qualitative research, it is flexible in so far as it can adjust/adapt to the settings and methods of a specific research project. When collecting qualitative data uses everything from "semiotics, narrative, content, discourse, archival -even statistics, tables and graphs" (Denzin and Lincoln, 2013.)

Qualitative research is unique based on the fact that it focuses on real life individuals, scenarios and situations thus providing a detailed analysis of attitudes, thoughts, behaviours and experiences according to Harding, Nettleton and Taylor, (2002) and Denzin & Lincoln, (2013). What qualitative research adds is an analysis of behaviours, attitudes and characteristics we are specifically interested in, where the focus lies within the "richness of detail" (Harding, Nettleton and Taylor.2002). Because I am interested in this process of gendered identity formation and experiences of women I worked within a Qualitative research paradigm.

With reference to my research I looked specifically at theory as well as the narratives that give us insight into the process of how one conceptualises and understands gendered identities. I considered and analysed gendered identities from within the context of a very specific genetic condition and I interrogated how this impacted on the narratives of the participants. Through the qualitative research my aim was to uncover the in-depth narratives and meanings that these women associated with the process of diagnosis and their condition which would ultimately influence their narratives and their individual understanding of their

gender identity and how it is constructed. The Qualitative research that I undertook used ethnographies, interviews as understood in Harding, Nettleton and Taylor, (2002).

An Ethnography or the ethnographic method is essentially a way in which researchers analyse people's behaviour in everyday contexts and situations according to Hammersley, (1998) as oppose to the traditional scientific research environment. First made popular by Anthropologists the data collected from an ethnography is based on observing the participants or through informal conversations which means the information that is collected is usually raw and researchers use this to analyses and interpret meanings and even experiences as understood by Hammersley, (1998) as well as Gay y Blasco & Wardle, (2009).

When using ethnography as part of your study you put yourself in a unique position in relation to your participant, in this instance you as the research are not the expert but your participant is the one with the valuable insights. Many researchers would describe the ethnographic method as an outsider looking and analysing the thoughts and lives of their participants. Using the ethnographic method, you must let go of your own presumptions and assumptions about a group of people in order to effectively learn anything about them.

The challenge when performing an ethnographic study lies in the fact that you have to approach your study with no preconceptions or hypotheses according to Gay y Blasco & Wardle, (2009) and Hammersley, (1998). Remaining objective is essential, in relation to my research project, objectivity means that you must look at how these women live with Turners Syndrome form their own perspective. What I must be mindful of as a researcher is that I have also been diagnosed with the exact same condition as my respondents thus I must not let my own personal views and experiences affect those of my respondents. Thus, I must ask how I would remain objective in light of having a reflexive position as both a woman living with Turner's syndrome and a researcher who is studying the condition. This research project however focuses more on using an autoethnography.

An autoethnography is an approach whereby the researcher uses elements of both ethnography as well as a personal biography as a means of analysing personal experiences in order to understand how these experiences relate to theoretical concepts and constructs (Anderson, 2006). Essentially the researcher becomes an active participant of the actual research project. According to Anderson, (2006) some of the key features of an autoethnography is analytic reflexivity, a visible narrative of the researcher, dialogue with research participants and informants as well as clear discussion of theory and discourses.

When writing an autoethnography, an author will write about past experiences which is narrating their version of a story and their understanding. In writing, the author would interview other participants as well as consult with supplementary sources of texts such as photographs and diaries, to help with the research which complements the ethnographic method of research very well. Central to my methodology is the gathering and analysing of the narratives of my participants and their social realities about living with a genetic condition.

In the reading by Latham, (2017) the definition of an autoethnography initially used by Stacy Holman Jones ,Tony Adams and Carolyn Ellis who define autoethnography as "purposefully commenting on or critiquing culture and cultural practice , making contributions to existing research, embracing vulnerability with purpose and creating a reciprocal relationship with audiences in order to compel a response" (Latham,2017).This definition encompasses everything my project and research is about which is challenging and questioning one's gender identity while contributing to existing research on Turners Syndrome and embracing the condition and using it as a tool to drive knowledge creation and research.

If I consider my own journey with Turners Syndrome, my positionality is that of I am both a researcher as well as a participant based on the fact that I am researching a condition that affects me personally and is something I am living with. This affords me as a researcher a certain level of self-reflexivity or "analytic reflexivity" (Anderson.2006) which allows me to take this a dual role as participant and researcher. I thus hold a very different position within my study because I am living with same condition as the participants, meaning I have my own experiences and interpretations of living with Turners Syndrome that will inform or influence different parts of my research, as well as being the researcher that is analysing the experiences of my respondents as well as my own position as a woman living with the syndrome allows me to conduct autoethnography in my research.

This is something very relevant to my personal story as I have taken my condition and used my journey as inspiration for this master's research project, however I had to ensure that I do not impose my experiences on the participants. I have moments throughout this research project where I am the researcher conducting the interviews and writing the report and then I also integrate my own story and experiences with Turners Syndrome alongside the stories told by each participants as a means of exemplifying my own narrative as both patient as well as researcher.

Narratives

The narrative research method is a term that best describes research that focuses on the written and/or spoken word of individuals and their lived experiences according to Frank (2013). Another term used for narrative are "accounts of the immediate"(Gay y Blasco & Wardle, 2009) or first person accounts where the information is coming from your source first hand which means that your data will be "immediate"(Gay y Blasco & Wardle, 2009).Since narratives fall under the qualitative and ethnographic research method they can provide a type of knowledge that is rich in detail allowing us as researchers the ability to comprehend and understand our participants story (Charon, 2001).

I have previously argued that the narratives of patients actually have the potential to give us insights into the psychosocial and often personal aspects of having a condition such as Turners Syndrome. These narratives considered the participant's stories about being diagnosed with Turners Syndrome and how these different elements intersect and impact the way in which these women see themselves and construct their identities. I would argue that these narratives are an interphase between the medical process of diagnosis and these women's own narratives about their gendered bodies, their identities and themselves. These patient's narratives are often very "grounded in experience and encourage reflection" (Greenhalgh & Hurwitz, 1999). Narrative can actually serve as a very useful analytical tool that can encourage a more holistic approach to treatments (Greenhalgh & Hurwitz, 1999) in the context of the medical field. Thus we speak of a patient narrative based on the fact that each of my participants are giving a narrative from within the context and understanding of their medical condition.

With any medical condition, illness or disease we have this inclination to rely on empirical science and facts as a means of understanding the condition without considering any of the social implications or the context of the patient let alone how the patient understands the condition. My goal for this research project was to gather the narratives of five (5) women diagnosed with Turners Syndrome and to understand how this genetic condition has impacted on their everyday lives since being diagnosed and what impact this diagnosis has had on their understanding of their gendered identities.

I would also like to understand how my participants interpreted their medical diagnosis and how the condition was initially explained to them by their doctor or endocrinologist. As part of collecting the narrative I had to add some contextual element to the study by looking at my

participants' life stories and background from their early childhoods which helped me to understand the perspective of my participants or in the relation to their narratives. These narratives collected in this project is what is referred to as a *patient narrative* which is something discussed by Rita Charon when she writes about turning to a "narrative knowledge'' (Charon, 2001) that we as researchers can use to interpret the significance of meanings and stories and their relevance which from the perspective of the researcher could possibly be a link between the motivation to be a part of the Turners Syndrome South Africa support group and their need of these women to tell and share their stories.

I then moved to the narrative of their diagnosis and gathered information pertaining to how the participants were diagnosed and by who, what was entailed in the diagnostic process and how they felt about being diagnosed with a genetic condition. Once I had discussed the background and process of diagnosis I then asked my participants questions relating to their gender identity and how they understood their femininity or feminine identity in relation to Turners Syndrome. Taking the above into consideration one interview per participant was sufficient to gather a comprehensive a narrative. My assumption when starting with this research project was that one's understanding of what Turner's Syndrome is and how their diagnosis was explained to each individual shaped the narratives of each of the participants.

The very notion of telling one's story about Turners Syndrome was a very empowering and very therapeutic moment for my participants as well as for me, because these interviews as well as the Turners Syndrome South Africa support group allowed us the opportunity to talk someone with the same condition that we could relate to and someone that understands the perspective.

Interviews

I decided to use an interview as research instrument which is written on at length in Harding Nettleton and Taylor, (2002), or more specifically a "qualitative interview" (Groleau et al.2006) as my primary method of data collection. In the context of my study the interviews focused on collecting qualitative information which was in the form of the narratives of five women living with Turner's Syndrome thus the interviews were centred around the experiences and stories of my participants. The structure of the interviews was predominantly framed by each individual participant as each participant's narrative and experience of Turner's Syndrome was unique. From my experience the interviews where more conversational when guided by the participant.

However, if there was indeed a comment or point made by my participant that I felt needed further discussion or clarification I addressed those after the initial discussion keeping the interview more relaxed, which adds to the flow of discussion. I chose the interview method because I felt that it would complement my research question as well as the qualitative method. When one chooses to research attitudes, meaning and experiences it is important for you as a researcher to be in the same space as your participant building that positive report and personal connection needed when dealing with a personal subject such as identity and personal experiences. In the context of my study the interviews were led by my participants and the narrative of each participant and I respected the information that the participant chose to share.

A key aspect that I paid particular attention to during the interviews was the practical side of living with Turners Syndrome where I looked at a range of topics from early childhood development, diagnosis and treatment and I even added a section on interpersonal relationships. From a researcher's perspective the questions that centred around living with Turners Syndrome provided me with valuable content and perspective of each participants. I really wanted to collect a narrative that fully grasped the nuanced aspects living with this genetic condition and how this diagnosis was perceived. I wanted a frame of reference that went beyond my own interpretation and understanding of my experiences. Essentially what I wanted to know was how drastically Turners Syndrome changed the lives of each of each participant and their perceptions of themselves.

Themes for the Interview

- Background and participant history
- Process of diagnosis
- Interpretation/understanding of diagnosis
- Social life and Adjustment after diagnosis
- Understanding their genetic Difference
- Understanding their Gendered identity
- Support

Sample of participants

For this research project, I selected a sample of five (5) women between the ages of 18 and 50 who have all been diagnosed with Turners Syndrome at different stages. I selected my sample by using a purposive sampling method, meaning I selected this group of women because they share a common trait or in this instance a shared diagnosis of a rare genetic condition .Despite the fact that my sample is focused on a particular condition, the distribution of information amongst the ages of the respondents as well as the narratives of their diagnosis experience and those narratives produced a compelling comparative aspect to the research.

The participants were all recruited from my Turners Syndrome South Africa Facebook page which I started in 2015 and the Turner's Syndrome WhatsApp group which I created in June of 2016. My initial idea when starting the Facebook page was to create a platform for women that were diagnosed with Turners Syndrome to share their stories and experiences on the page however after numerous emails and direct Facebook messages I found that these women wanted to connect and talk directly with other women diagnosed with Turners Syndrome thus the Facebook page prompted a WhatsApp group that became a support group for these women.

Each member initially contacted me on a voluntary basis as Turners Syndrome South Africa is listed as a support group on the Genetic Alliance South Africa website with my contact details, as the women contact me I subsequently added them to the group. We communicate on a weekly basis on WhatsApp sending messages of support to each other but also sharing our experiences thus creating a very positive and comfortable environment for each member. I sent out a general invitation on the WhatsApp group inviting the members to participate in

my research project and all the responses were very positive and five members on the group were willing to participate. I scheduled the interviews with my participants either via a Skype interview online or at a restaurant or coffee shop were the participants felt at ease and comfortable, it is imperative that the interviews happened at a place where the participant felt at ease.

Participants:

- Participant 1 (50yrs.-married - diagnosed at 17yrs-no children)
- Participant 2 (18yrs- single-no children-diagnosed at 6yrs)
- Participant 3 (26yrs-married-diagnosed at Birth)
- Participant 4 (27yrs-engaged-diagnosed at birth)
- Participant 5 (42yrs-married- diagnosed at 17yrs-adopted daughter)

Ethics

I obtained permission and ethical clearance from the Wits University non-medical ethics committee as well as the Humanities faculty by means of an ethics application form before I conducted the interviews. This is particularly necessary since my research topic deals with very personal and sensitive issues. I fully briefed each of my participants on my research topic and ensured that they understood their involvement in the project and rights as a participant. Eliciting sensitive information and personal stories required informed consent and a consent form signed by participants before I conducted my interviews and recordings.

I also had to provide each participant with a support information document that had the numbers of various doctors and organisations that each participant could contact should they require further assistance and support or counselling. I maintained transparency and open communication with the participants and ensure that they were fully briefed on their involvement and participation in this research and that I will be recording the interview and taking notes during the interviewing process which will be used in this project.

I respected any requests by participants to remain anonymous as some of the participants might have felt embarrassed about the condition or may have been bullied or teased which caused the participant high levels of anxiety or stress that made them feel uncomfortable

sharing their narrative. I had to ensure that any personal or harmful information in the interviews remains confidential between myself as the researcher and the participant. With delicate or private information, it remained my responsibility to check that the information is kept and stored safely, locked and secured to ensure that it remains confidential. I also gave each participant the option of using a pseudonymous instead of their real names to ensure that they are protected and to ensure confidentiality however they consented to me disclosing first names and their ages that was used throughout the project.

I was cognisant of the fact that the participants may have felt uncomfortable or vulnerable as some of the details might be quite intimate or even evoke emotions of being bullied or teased at school for example. Thus I ensured an open dialogue with the participants should they at any stage have felt uncomfortable they could stop the interview. As researcher it was also my responsibility to create a safe space where my participants felt protected and comfortable while sharing their stories. During the interviews, the participants were aware that they could communicate whether they felt comfortable answering any of the questions and should they at any stage have felt the need they may have requested that I stop the interview process.

As a researcher, I focused on the ethical principle of no harm and endeavoured to protect the identities of my participants and fully brief my participants to the best of my abilities so that they are not harmed in anyway. It is also my responsibility that my participants were treated with the necessary dignity and respect throughout the process and that should they require any form of counselling that they are given the necessary help and contacts which I subsequently provided should they feel the need to talk to a psychologist or genetic counsellor. On a more practical note had to ensure that my interview questions were not vulgar or insensitive. I remained in contact with the participants and informed them about any progress made on the research project and sent them a copy of the project should they request one. I have the responsibility as a researcher to adhere to the ethical code of the university and to uphold the high level of professionalism that the university prides itself on.

Looking in retrospect at the interviews with my participants one issue that I must mention was it was particularly difficult to stay impartial or objective while doing the interviews. I had to really keep my emotions in check and stay focused throughout the interviews. I also noticed that I had to constantly remind myself that I was in fact a researcher and not a participant, thus during the interviews I was there to listen to the participant and wanted the interview to centre around their narratives and not my own story .I consider my story as a

supplementary element that is considered alongside the narratives I have gathered from each participant. What the interviews allowed me to do was actually look at my own experiences from a different perspective because I was now a part of a group of women that I could relate to. After meeting each participant, I gained a level of respect for these women and their positivity and optimism because they taught me that Turners Syndrome can become your biggest drive to succeed and not an obstacle.

Chapter 3:

Literature review

This literature review begins by providing an in- depth discussion of the genetic condition referred to as Turners Syndrome as a means of giving some context to the analysis and to the research project at hand. Since Turners Syndrome is a rare chromosomal condition this serves as an evident justification and motive for doing this project. I began this review by firstly defining Turners Syndrome and then discussing the historical research which has been done to date. I have also discussed the different elements that surround Turners Syndrome including the physiological as well as psycho-social elements.

The second part of the literature serves as a theoretical extension of the discussion on *sex* and *gender* identity which is a critical part of understanding Turners Syndrome with regard to the social aspects of one's identity. Finally, I have added a discussion on patient and illness narratives which is a core component of my study considering the fact that I was only able to find two pieces of literature that engage with the stories of patients and their experiences with Turners Syndrome. By analysing Turners Syndrome, we are afforded the opportunity to critically assess concepts such as *sex* and *gender* which are traditionally concepts we don't question or challenge and take for granted however what we will see from the literature is how contextual and fluid these concepts actually are. We are also given a chance to consider the narratives and perspectives of patients as a means of critical analyses.

Turners Syndrome

I begun by defining Turners Syndrome, the associated characteristics and side effects, as well as all of the associated health risks. The very notion of something being a 'syndrome' implies that this condition comprises of multiple symptoms which occur together in a consistent manner which is precisely what Dr Henry Turner an endocrinologist from the University of Oklahoma in the United States of America observed in 1938. I was fortunate enough to find the original report by Dr Turner in which he mentions a previously unreported condition which he observed in some of his female patients. Dr Turner mentions several of his patients having a webbing of the skin in the neck area as well as a deformity of the elbow or 'cubitus

valgus'(Turner.1938) which he initially likens to 'Klippel-Feil Syndrome'(Turner.1938) however what made this condition different/unique was the noticeable retardation of sexual development. I noted from the report that all of Dr Turner's patients were between the ages of 15 and 23 years hence the condition could only have been diagnosed when his patients had reached puberty and thus we have the condition known today as Turners Syndrome.

Turners Syndrome is a genetic syndrome that is sometimes also referred to as "gonadal dysgenesis" (Fausto-Sterling, 2000) where females do not have the second x chromosome which is associated with secondary sexual characteristics such as regular menstruation and breast development. Females diagnosed with Turners Syndrome are short in stature and don't naturally produce the hormone Oestrogen according to Fausto-Sterling, (2000) and don't experience puberty. Apart from the genetic complications according to Gravholt, (2001) Turner's Syndrome is associated with a multitude of medical problems such as cardiovascular disease, insulin resistance and Ischaemic heart disease.

Dr Turner was the first endocrinologist to write a formal report that documents Turners Syndrome we actually have very little historical background knowledge which is where Professor Andrew Zinn in 1999 plays a crucial role as he explored some of these historical aspects. Zinn, (1999) mentions three of the most probable earliest reported cases of Turners Syndrome which date as far back as Giovanni Battista Morgagni (1682-1771) a well-known Italian anatomist. A second case was reported in 1925 by Seresevskij and a third case reported in 1930 by German geneticists Otto Ullrich.

The work done by Andrew Zinn, (1999) coincides very closely with another core piece of literature which is an information book edited by Claus Gravholt, (2009) were the focus is also on this very biomedically oriented explanation of Turners Syndrome. In the initial chapters what Gravholt & Novo Nordisk, (2009) do is add a more psychological and social dimension to the discussion of Turners Syndrome which adds new depth and perspective to the understanding of the condition. For example, in chapters four and five there is a discussion on transitions and puberty which is something neither Zinn, (1999) nor Turner (1938) cover to that extent of detail which is what makes Gravholt & Novo Nordisk, (2009) a welcome addition to the literature.

What I can give some critique on is that the information in this book caters for a very specific audience thus I struggle to find ways to link it to a South African context and experience of women with Turners Syndrome very unclear sentence. maybe just delete and start with next

sentence. Turners Syndrome is a condition that remains the same no matter where in the world you are. however the experiences that different women may have in different parts of the world will differ depending on factors such as the information given was provided by medical professionals such as endocrinologist, access to treatment in a functioning health care system and support societies which is a challenge here in South Africa where there are only a very limited number of diagnosed patients and a general lack of knowledge and willingness to talk about Turners Syndrome long sentence. Another important aspect of Turners Research in South Africa is the cultural stigmas caused by infertility based on the fact that Turners Syndrome affects women from any ethnic, cultural or social background.

At this present moment in South Africa the number of confirmed diagnosed cases of Turners Syndrome is unknown based on the fact that the Department of Health does not focus on gathering information on congenital and genetic disorders and conditions. However, what I can say from having spoken to the women on the Turners Syndrome South Africa WhatsApp group is that Turners Syndrome is an anomaly that affects women of any racial, ethnic or cultural classification at random. I have spoken to women from all racial and cultural backgrounds on the Turners Syndrome Whatsapp group that currently has 19 active members. I have also engaged with women on the Turners Syndrome Facebook page that has 72 followers at present.

Subsequent to this I found an article published in 1971 which focuses on Turners Syndrome in the "Bantu Population"(Grace.1971) which happens to be one of the few South African articles I have found pertaining to Turner's Syndrome apart from the published dissertation by Matebese, (2008) which was project done at the University of Cape Town that centred around the more psychological and social aspects of Turners Syndrome where nine participants living with Turners Syndrome were recruited from the Groote Schuur Hospital to partake in an interview.

The chromosomal analyses for Turners Syndrome only really started in 1950's (Zinn, 1999) and by the mid-1960's could we do empirical chromosomal testing or a "karyotypic diagnosis" (Zinn,1999) to determine that Turner's Syndrome actually involves a complete or in some instances a partial missing sex chromosome which is referred to as "mosaicism" (Zinn,1999).According to the article by Ibarra-Ramirez and Martinez-de-Villarreal,(2016) it was in 1959 that doctors confirmed the "Chromosomic base"(Ibarra-Ramirez and Martinez-de-Villarreal,2016). Now that genetic studies have advanced we could develop a phenotype

for Turners Syndrome and are able to detect and treat Turners Syndrome much earlier however the report by Ibarra-Ramirez and Martinez-de-Villarreal, (2016) state that 15-30% of Turners Syndrome cases are not reported until after the age of 12.

Considering that Turners Syndrome directly implicates these women's sexual development it follows then that 98% of these women with Turners Syndrome are infertile according to Zinn (1999). The research report by Zinn, (1999) mentions other complications associated with Turners Syndrome such as the psychological and educational issues for instance attention-deficit-hyperactivity and issues of socialisation and low self- esteem, which reinforces the need to research the experiences and narratives of these women since the focus of literature was bio medically oriented.

On further research, I found that a large portion of the articles about Turners Syndrome actually write about growth hormone therapy and Oestrogen replacement therapy which is in fact a reality for most women living with the condition. According to Rosenfeld, (1997) the use of hormone replacement therapy dates as far back as Dr Henry Turner in 1938 who used this therapy on this first group of Turners Syndrome patients in the USA Rosenfeld, (1997) further evaluates historical data pertaining to the use of growth hormones and found that the growth hormones are indeed effective, which is something I can confirm since I was on hormone therapy for a year.

There were however there were questions raised as to what age therapy should start and how the dosage should be regulated as well as some of the psychosocial impacts of therapy. In a study done in Taiwan by Hsu et al, (2008) they tested growth hormone treatment on children aged 9 months to 15 years and found that in fact the younger participants actually reached a relatively normal adult height stating that early detection and treatment for those women diagnosed with Turners Syndrome "is advisable, so that puberty can be induced at the appropriate age" (Hsu et al.2008).

The argument by both Hsu et al, (2008) as well as Rosenfeld, (1997) focuses on how growth hormone treatment can aid those living with Turners Syndrome even with something as simple as reaching a normal adult height based on the fact that short, stature is something very prevalent for women with Turners Syndrome Ibarra-Ramirez and Martinez-de-Villarreal even described hormone replacement therapy as "Fundamental for these patients" Barra-Ramirez and Martinez-de-Villarreal, (2016).

This view is supported by Hjerrild et al, (2008) who did an entire study on the clinical treatment of Turners Syndrome and found that although early detection and treatment is beneficial the correct dosage of "female sex steroids" (Hjerrild et al.2008) is still uncertain and left to the discrepancy of the endocrinologists and doctors. Both Hjerrild et al (2008) as well as Hsu et al, (2008) agree that there is a need to include the psychosocial aspects of treatment.

The vast majority of the women on my Turners Syndrome WhatsApp group have been on some form of hormone therapy or medication with varied levels of growth and success. This substantial literature on hormone replacement therapy and clinical treatment by authors such as Hjerrild, Mortensen, & Gravholt, (2008) and Doherty et al, (1997). There was in fact very little written on the more social implications of being diagnosed with a genetic disorder.

The writings of Smith, (1999) and Clark, (2015) help us to understand how Turners Syndrome is far more than just a purely genetic biological condition, but that the effects extend further, and has an impact on all aspects of these women's lives. Smith, (1999) notes how the medical and physical aspects of Turners Syndrome are much easier to treat and deal with because they are obvious which relates directly to the literature on hormone replacement therapy and treatment and how patients received this treatment to help with their growth. Brook, (1986) writes a very comprehensive and practical piece on how one can manage Turners Syndrome in three stages after the initial diagnosis namely "Discussion with parents, Management of growth and Induction of puberty" (Brook,1986). I have noted how most of the articles actually relate directly to doctors intervening.

There appeared to be this stigma that Turners girls have some form of mental impairment however the contrary has been noted by both Turner, (1938) and Zinn, (1999). The problem with stigmas associated with one's intelligence and mental capabilities is that they create certain misconceptions about the condition that it hinders the process of a proper and balanced diagnosis and a positive prognosis by endocrinologists. Another huge gap in the literature is based on the fact that the "scientific study of Turners Syndrome is quite young" (Smith, 1999) thus we need a study that doesn't just focus of the genetic makeup of these women but how this condition actually affects them in a tangible and real way which is further explored by Smith, (1999) where she uses the example of ageing in her article. I can argue that we most definitely need more substantive research done on Turners Syndrome that includes the patient's stories. This type of research will include a platform where Turners

Syndrome patients share their experiences and narratives which will ultimately be beneficial for physicians and patients alike, giving the type of research done on Turners Syndrome a new dynamic and perspective.

In South Africa one of the most relevant research was done by Matebese, (2008). he also did a qualitative study where the sole focus of the research was based on the psychosocial difficulties relating to Turners Syndrome which was very helpful and added a comparative element to my own work as well the extended the arguments by Smith, (1999) and Clark, (2015). Noted in the study by Matebese, (2008) was self-esteem issues and negative body image amongst girls with Turner's Syndrome where they had difficulty relating to friends and even engaging socially.

"Sex and Gender"

The debate that surrounds trying to understand and define *sex* and *gender* can literally involve one sitting for hours contemplating the distinctions ad arguments. When one tries to understand this seemingly simple process of drawing the distinctions between what is understood when one refers to *sex* and *gender.* A common misconception that society has is that *sex* and *gender* actually equate to the same thing and that they that fall under one homogenous category or that *sex* and *gender* are "coextensive" (Mikkola, 2008) concepts. However, what the literature alluded to is the fact that *sex* and *gender* are actually separate concepts that relate to two different aspects of one's identity. This binary notion of *sex* and *gender* where *sex* is something grounded in nature and biology while *gender* is grounded in culture, behaviour, identity and one's position within society according to Mikkola, (2008).

What is starting to develop here is an argument that is infinitely more complex than what society actually perceives it to be. The binary definition or conception of *sex* and *gender* needs to be seen in relation to other factors such as society and psychology and how they also influence our understanding of *sex* and *gender.* When one includes society and culture in a debate it challenges the robust and structured biological argument that makes our understanding of *sex* and *gender* far more dynamic. In a bid to try and understand *sex* and *gender* we need to go back to basics and analyse what we know about *sex* and *gender* and what has historically, socially and even culturally influenced heterosexuality (two sex system) and gender normativity (normative/socially accepted behaviour for men and women.

Professor Anne Fausto-Sterling argues that a body's *sex* is too complex to view in a narrow/restrictive fashion. Fausto-Sterling suggests that one should rather note how there are "shades of difference" (Fausto-Sterling, 2000) when one discusses *sex* and *gender*. Traditionally scientists, biologists and medical professionals adopt a very binary and dichotomous view of what is defined as *sex*. We would define someone as '*male*' or '*female*' based on observable, physical, physiological, anatomical characteristics. For example being a women or female is our *sex* which in the normative sense is denoted by "breasts and a vagina" (Fausto-Sterling,2000). Another critical aspect to one's *sex* that relates very closely to Turners Syndrome is the argument of genetics and chromosomes.

Historically the study of genetics and the human anatomy and biology was at the forefront in defining what is scientifically defined as *man* or *woman*. Essentially, doctors and medical scientists have justified their position based on biology and scientific fact. Once medical science was taken as empirical truth or fact which is often referred to as "biological determinism" (Mikkola, 2008). Society adopted the notions of biological determinism and integrated these scientific facts into their definition and separation of men and women.

As a growing field of study psychology along with feminism and feminist theory gained momentum and subsequently it was in fact Feminist scholars that introduced us to the argument for a social constructivist view of *gender*. Social Constructivism argues that *gender* is socio-culturally defined while *sex* is something natural and defined by one's biology. From this argument the sex-gender binary was introduced. From this feminist tradition emerged theorists such as Anne Fausto-Sterling and Judith Butler who argued the point that *sex* is also social.

Margaret Wetherell also noted the idea of social constructivism and states that individual social identities should be regarded as projects that are constantly being made and remade. Since these women are living with a genetic condition that affects multiple facets of their identities and lives one can argue that *gender* is something that is constantly being negotiated and interpreted. At this point we can assert that a gender identity is based on 'collective understandings' of socially significant categories according to Wetherell, Maybin, & Stevens, (1996). These "collective understandings" that Wetherell et al., (1996) refer to all develop from the dialogues, experiences and observations that we as individuals would use as sources of information to understand/comprehend notions of *sex* and *gender*.

What I have attempted to broadly outline above is the argument that *sex* and *gender* are concepts that lie at the borders of biological and medical knowledge as well as the social and cultural norms and values. In a more recent article Latham, (2017) describes *sex* as being "made" or "produced", (Latham.2017) which reinforces this notion that *sex* and *gender* do not have to be exclusively bound to the biological definitions and understandings, but that they can be attributed to particular "contexts, practices and encounters" (Latham.2017).

My research shows how both, biology and culture play a role but it appears that cultural/social play a more prevalent role insofar as the research participants are not *female* in terms of *sex* but identify as *women* in terms of *gender*. Here I would briefly like to mention the theory of "Gender Socialisation" (Mikkola, 2008) that argues the premise one acquires and learns appropriate gender specific behaviour. Thus, the argument focuses on the aspect of nurture and how individuals are raised.

Another theorist of social learning theory who writes about the social and personal aspects of *gende*r is Michael Kimmel who in 2009 wrote a piece on the different sources of social information where he mentions parents, siblings, family, friends, schools and media as people and places which we interact with and learn from as primary sources of information about our *gender*. After reading Wetherell et al., (1996) and Kimmel, (2009) I can clearly see how one's *gendered identity* forms part of a dual process that is not only "historical and social" (Kimmel.2009) but also "personal, private and individual" (Kimmel,2009) .After reading books published by both Anne-Fausto Sterling, (2000) and more recently Judith Butler, (2004) I began questioning and grappling with this notion of femininity and identity. At the very crux of the debate that Fausto-Sterling and Butler present is that there is a definite social dimension to our gendered identities and that there is a stark difference between what one understands as *sex* and what we then label as *gender*.

Taking the above debates into consideration one can begin to conceptualise what Anne Fausto-Sterling means when she says "Labelling someone a man or a woman is a social decision" (Fausto-Sterling, 2000). Biology and science only account for one aspect of the feminine identity and experience. Hence why we as researchers need another perspective and understanding of the feminine identity that is located outside of scientific and biological discourses to give a more social account of the experiences and realities of women. When one works within the discourse of *gender* it involves the process of socialisation by which we as

members of an assigned male or female *sex* learn socially acceptable behaviour as well as the expectations and characteristics that accompany that biological label.

Anne Fausto-Sterling who is a biologist by training, but much later in life became a historian of science. For this project, her book 'Sexing the Body' (Fausto-Sterling, 2000) is a pivotal piece of literature that forms crucial part in the understanding of constructs such as *sex* and *gender*. In the first chapter of her book Fausto-Sterling, (2000) challenges our understandings of what we as society consider and understand to be normal and what is natural much in the same way as Butler (1990) and Kimmel, (2011). Keeping with this common thread of trying to understand the social and biological arguments for defining *sex* and *gender*, Fausto-Sterling also mentions the traditional binary model of having two sexes (male/female) and two genders(masculine/feminine).

Fausto-Sterling grabs one's attention by first challenging one of the most iconic and loved professions in the world which is sport. To be more precise Fausto-Sterling (2000) focuses on one sport in particular, which in recent times as also come under fire, which is the world of athletics. This discussion and example really brings to the attention how fragile the concept of *sex* and *gender* actually is and how something such as ambiguous genitalia or even a genetic disorder such as Turners Syndrome can disrupt our understanding of *sex* as a concept. Fausto-Sterling essentially takes one of the most patriarchal and gender divided platforms in the world and completely disassembled it by showing how these notions of *sex* and *gender* are actually very fluid in nature and highly restrictive and ultimately not very practical.

When one is limited to being either male or female it restricts people's understanding the what true diversity there is in the world, if we construct a world with such rigid ideological tools then we will fail to acknowledge and appreciate the differences. As with the initial sporting example if we continually divide bodies into exclusively male and female sports we risk excluding a potentially excellent sportsperson from participating. When we think about the situations and examples that are outside of these perceived societal norms, such as Turners Syndrome, we should challenge our way of thinking about such bodies and how we understand them, however after reading Fausto-Sterling the aim is not to create a completely new set of norms and discourses but rather to recognizing the various ways in which human beings can exist and interact with each other.

Fausto-Sterling, (2000) brilliantly dissects the nature versus nurture debate from both the human and natural science perspectives. She approaches the debate from a multiple disciplinary approach and includes politics, history, philosophy as well as sociology in her critical analyses of the debate. At the heart of her argument was the idea that one's biological-sexual characteristics do not necessarily equate with his or her *gender* which was a very profound notion. Fausto-Sterling, (2000) argues that a gender identity arises neither universally from nature nor nurture but rather from the way society views and understands human beings within communities. She poses some very important questions as she critiques both scientific and medical research, citing examples from the natural sciences and daily life to reinforce her arguments. It is this type of engaging dialogue that allows us to discover what it means to be human.

For Fausto-Sterling, (2000) a human being cannot be defined by his or her genitalia but is ultimately a consequence of his/her culture, history, purpose, and values whereas Butler, (2004) would argue from the perspective of society and power dynamics. The idea of gender ambiguity or gender fluidity is socially unsettling to many people since it completely disrupts the normative gender binaries. Fausto-Sterling, (2000) argues that within this instability of discourse with regards to the notions of *gender* is what has resulted in medical and scientific faculties to justify their research and in construction concrete concepts of *sex* which are the individual's anatomical attributes, and gender which is the internal conviction/understanding of one's masculine and feminine identity. Fausto-Sterling, (2000) adds more to the realm of ethics and morals, and then extends this to include the more political and social arguments that necessitate thinking about human bodies in a multidisciplinary way.

Butler is widely recognised for her work on the notion of gender performativity in her book ‘Gender Trouble’ (Butler, 1990). What Butler does in this book is extend the discussion by authors such as Kimmel, (2011) and Wetherell et al., (1996) and critically analyse how one construct a gendered identity. Butler links categories of *sex* and *gender* to specific structures of power which is also something noted by Latham, (2017).

According to Butler, (1990) and most of the above-mentioned theorists the concept of a gendered identity is often assumed to be a natural expression of the human anatomy or anatomical features, thus we as a society consider a gendered identity to be something innate. Since we lean heavily on biological and physical facts to inform our gender the assumption is that when we construct our gendered identities we tend to overlook and misinterpret the more

poignant social and public aspects that form a part of this very complex and dynamic process. In her theoretical arguments, Butler, (1990) alludes to how the social and biological aspects of the process of creating a gendered identity is actually at times even counterintuitive. What I am alluding to here is the fact that by drawing distinctions between a women's *sex* and *gender*.

Butler's work has aided us in thinking critically about the ways in which we view notions of *sex* and *gender* by including a discussion on the role of power and the power dynamics and the structures of power that perpetuate/create a specific gender identity. This argument by Butler, (2004), Kimmel, (2011) and Wetherell et al., (1996) is exemplified by Turners Syndrome as a genetic condition because these women do not have those essential biological and physiological characteristics which for many are at the core of our gender identities. Hence why I can argue that in this instance *gender* is in fact something that extends past biology and shows how we would have to include Butlers' notion of "performativity" (Butler,1999). What Butler means by his performativity is that *gender* develops through a "stylized repetition of habitual acts" (Bultler.1999) that reinforces the idea of *sex* and *gender* by means of dressing a certain way or having certain mannerisms. Thus, by constantly reinforcing such accepted behaviour we create the impression that gender is in fact something natural.

We can thus assert by using authors such as Judith Butler and Margaret Wetherell that we as a society do not simply create our own notions of *gender*, for gendered identities are always produced within social contexts of society and by communities by means of socialisation; we are assimilated into a specific gendered role thus assuming that identity. With reference to the discussion on the literature about Turners Syndrome we saw how Dr Turners' patients were all adolescents meaning that before they found out about their diagnosis they had to be socialised into the female role since the characteristics of Turners Syndrome only became apparent much later in life. This reiterates the point that Butler makes about how we are never truly able to remove ourselves from thinking about gender ideologically. This is of course where the challenge lies for academics, researchers and even women living with Turners Syndrome to disrupt the current systems of power that reinforce the current *gender* discourses and ideologies in society.

For this research project the pivotal question in relation to *sex* and *gende*r was weather biology had an exclusive role in this equation or where there were other factors that one needs to consider when trying to solve this conundrum. What each author has tried to do is grapple with is how one defines and reconstructs these notions *sex* and *gender* and how this had changed the "normative ideals of certain bodies" (Butler, 2004) After engaging with the literature and various theorists what remains an important factor for me is the role of society and the vast multitude of social forces that sculpt and mould our lives as members of society.

What I must acknowledge is how *sex* and *gender* as concepts have changed over time and will thus continue to change as society develops. What also became apparent is that *sex* and *gender* mean different things to different societies that these social forces play a very active role in different parts of society. Anne Fausto- Sterling uses the example of Ancient Greeks using a "one-sex model" where it was argued that men and women were of the same sex and it wasn't until the late 1700's that society actually moved to a "two-sex model". What I found most helpful was how each author brought their own interpretation and argument and yet make it accessible to understand and engage with. This definitely made me as both patient and researcher think very differently about my project as well my own condition, it really pushed my understanding and forced me to think more dynamically and critically.

"Telling Stories" – Narratives

A third part of my study was the research done around the narratives of my participants. If I can very briefly explain that within the context of this research project I was particularly interested in the entire process of how each of my participants developed their individual narratives starting from early childhood right through to the harrowing diagnostic process they had to endure as well as the more nuanced elements such as their views on their gendered identity as well as infertility and how these different elements culminated and work together to influence their narrative. This process of collecting and interpreting such narratives would provide us as researchers with a different analytical perspective that we can use to help understand our participants.

When one considers the narratives pertaining to the medical field and medicine in general initially the stories were framed in relation to the patient's experiences of their relationship with doctors and other healthcare professionals as well as the scientific truths and knowledge provided by these doctors. However, in more recent times Sociologists and Anthropologists

are using the patient's experience of “suffering” and “repair” (Frank, 2013) to help understand how they developed a narrative in relation to their illness or condition as a meaning of finding and understanding the “meaning of illness” (Kleinman.1998).

When one considers Arthur Kleinman, who wrote extensively on illness and healing, was also one of the first scholars to do academic work on narratives, medicine and culture. Professor Arthur Frank of Calgary University in Canada, author of the brilliant book “The Wounded Storyteller” (Frank, 2013) is another scholar that used his own narrative as a foundation for his academic works. As researchers we can now take the academic writings of scholars such as Kleinman and Frank as well as the experiences and interpretations of illness and conditions from research participants and use those as analytical facts in their own right. What we must note is that narratives are substantially different as sources of information in so far as they are focused on individual experiences and are shaped by one's memories and emotions according to Groleau et al., (2006).

Thus, the inclusion and validity of such narratives that focus more on the perspective of the patient has been somewhat of a contested subject as narratives are often criticised as being too subjective and lacking in empirical data. If one were to follow the discussion of the literature on Turners Syndrome for instance it appears that the literature provided by endocrinologists and geneticists seems almost synthetic in so far as it only provides a basic notion of the symptoms and the diagnosis, thus showing an basic level of understanding and by providing an explanation that is not inclusive of the patient.

A narrative gives us the opportunity to understand how the ‘patient’ copes with the more “internal psychological and external social dynamics” (Groleau et al,2006) of Turners Syndrome. In short, the reading by Greenhalgh and Hurwitz, (2006) also mentions that a narrative provides a framework from which doctors and patients can approach the patient's problems holistically meaning they will take different aspects into consideration during diagnosis and treatment.

A narrative would give the researcher much more information and insight into the “mutilated and polysemous” (Groleau et al.2006) world of illness and medicine, as well as giving us the opportunity to understand our participants. One must also be mindful of the fact that the experiences and narrative of patients is always told from a specific social position and from a specific point of view according to Groleau et al., (2006). Often narratives are very reflective

and grounded in the patient's experiences and interpretation according to Greenhalgh and Hurwitz, (2006).

In his book Frank, (2013) focuses on his own experiences with cancer as a starting point to his discussion and narrative. For Frank a narrative has the potential to change our current conceptions of illness from being bio-medically centred to one that is more patient centred. Narratives will make information about the illness more accessible meaning that patients would be able to relate to their illness on a different level. If I relate this to Turners Syndrome, having the narratives of other women living with Turner's Syndrome will not only help patients understand the condition it will also help patients not feel as isolated. I must also mention at this point that this would also help me as both and patient as well as researcher understanding my own narrative and experiences. Taking this very dynamic notion of narration into consideration Frank, (2013) proposes three major types of narratives in his book.

The first is the restitution narrative which is based on the premise that your health will be restored and you will get better or as Frank quotes "Yesterday I was healthy, today I am sick but tomorrow I will be healthy again"(Frank, 2013). This is a very positive outlook on illness and sees a sense of hope that the body will be repaired. From within the context of the medical realm endocrinologists may not be able to provide the missing genetic material but they can provide the necessary hormones to treat some of the symptoms such as the growth hormone therapy, which I have discussed and previous sections.

The hormone therapy treatment does give the patient a sense of hope since short stature is something all women with Turners Syndrome must live with. In this context medicine might provide these women living with Turners Syndrome with a sense of hope or restitution from their condition. I had to ask myself some critical questions such as would I be able to find a narrative that links to the role that medicine plays in the lives of these women? Do doctors and endocrinologists facilitate a restitutive narrative?

The second narrative is the chaos narrative which unsurprisingly is the least popular narrative. Essentially a chaos narrative is heard when a patient's illness becomes too intense and the patient is totally overwhelmed. Frank refers a narrative of chaos as "the anti-narrative of time without sequence, telling without mediation and speaking about oneself without being fully able to reflect on oneself"'(Frank.2013). The chaos narrative is usually very intense and disjointed and often doesn't have a sequence or order. In the context of a chaos narrative the

illness or life in general will not get better. The chaos narrative usually evokes a sense of anxiety and shows how vulnerable and frail humans actually are.

A quest refers to a person's journey, thus a quest narrative would involve the journey a patient takes to learn more about their condition and what they gain from this experience. According to Frank, (2013) and Groleau et al., (2006) there is something to be learned from being ill and from being diagnosed with a condition. Within the context of Turners Syndrome many of these women are on a quest to learn something from their diagnosis and to look at their diagnosis "holistically" (Greenhalgh and Hurwitz.2006). This is something very relevant to my personal story as I have taken my condition and used my journey as inspiration for my masters' project , however I must ensure that I do not impose my experiences on the participants.

From the above discussion, we can see how narratives can actually be useful however there is one point of clarification which is needed and that is define when we as researchers use a patient's narrative and when do we use a woman's narrative. This is imperative that we make this distinction because we need to understand the context of the narrative. When referring to Turners Syndrome and the process of diagnosis that will be the patient's narrative and point of view as it involves the patient's experience of medical doctors as well as a discussion on understanding the condition. When we speak of a gendered identity this will be the woman's narrative and how they experience their femininity in relation to the genetic condition. The difference lies in the context of the discussion.

An excellent example of this type of narrative that also intersects with *gender* and *sex* while being a patient in a clinic is the piece by Latham, (2017) were the author documents and writes a narrative about their experiences in a clinic as a transsexual and how the clinic either does not know how to attend to transsexual bodies of completely ignores transsexuals, in this instance the power lies with the clinic defining *sex* and *gender*. This is similar to the intersexed bodies mentioned in Holmes, (2009) were the doctors in a Dutch clinic decided the *sex* and *gender* of the babes found to have ambiguous genitalia.

Now that we have a better understanding of what narratives are and what they entail we can see that they have a distinct purpose in relation to research. In an article titled "Literary narrative in medical practice" (Kottow and Kottow,2002) it is noted how certain subjects and practices in the humanities are slowly being introduced into the study of medicine.

In the article Kottow and Kottow, (2002) argue that a well read and researched doctor adds a new level of understanding and empathy to the study of illness and sickness especially when it comes to the involvement and consideration for patients. Essentially narratives have the ability to "inform sense, stimulate empathy and compassion" (Kottow and Kottow,2002) amongst doctors thus allowing the patient space to speak and to be more involved especially when it comes to treatment. Rita Charon speaks of a "narrative competence" (Charon,2001) meaning that doctors should have the skills necessary to acknowledge and absorb these narratives and then react to the story.

In the journal "Critical intersex" (Holmes,2009)' edited by Morgan Holmes there is a chapter written by Margriet van Heesch where she talks about the dangers of the diagnosis and surgery of intersex bodies and why many doctors choose to withhold the information about the intersex bodies and corrective surgery which caused many of the patient's psychological and emotional trauma. What I understand from reading these articles is that there is a need for a more holistic and inclusive method of research and analyses which is precisely what a narrative does with respect to including patients and understanding their perspective and context. As I mentioned earlier in the case of Turners Syndrome there is a woman's narrative and there is a patient's narrative which is important to acknowledge when considering treatment options as well as their social context and position. In my opinion Turner's Syndrome exemplifies how different narratives can actually intersect and they are sometimes complementary or contradictory.

In summary, I have covered a vast spectrum of literature which I subsequently divided into two main parts. One half of the literature dealt specifically with the genetic condition known as Turners Syndrome. I used multiple authors and included the original report written by Dr Henry Turner in 1938 to frame the discussion around Turners Syndrome. The literature helped me define what Turners Syndrome is and the different aspects which all form a part of the condition as well as the historical developments that have all contributed to our current literature and knowledge. The literature was excellent at outlining and discussing the different physiological and genetic characteristics of Turners Syndrome however I was only able to find two really substantive pieces that relate to the more psycho-social elements of Turners Syndrome.

This was where my narrative approach to the study of Turners Syndrome would extend and supplement this discussion and bring an entirely new perspective to how we study such conditions. The second half of the literature was more focused on the theoretical discussion on the notions of *sex* and *gender* and two theorists in particular Anne Fausto-Sterling and Judith Butler dismantle. These two theorists brought to our attention just how dynamic and fickle the concepts of one's *sex* and *gender identity* actually are. And finally, I discussed the concept of illness narratives introduced to us by Arthur Frank which had bought an entirely new perspective to the table and could potentially change the way in which we study disease since most the literature is based on medical science and knowledge we can change this notion to studies being more patient centred.

Chapter4:

Research Findings

4.1 Early Childhood and Development:

The first participant I interviewed was Erna (50) who lives in Vereeniging in the South of Johannesburg. Like all women with Turners Syndrome Erna is a petite lady, from the initial greeting and introduction one already gets the impression of someone who is warm and sincere. What made a huge difference was the fact that both I as researcher as well as my participants have Turners Syndrome which added a crucial element of positive rapport and trust between us, making a more comfortable atmosphere during the interview. The initial questions in the interview focused more on the participants childhood and childhood experiences. The reason as to why I started the interview with such questions was based on the fact that these childhood experiences are foundational in understanding the participant and gives much insight into their lives and gives context to their narrative.

Erna described her childhood as follows “I had a stable childhood and grew up with a brother and sister. My grandparents on my mom’s side came to stay with us when I was six years old. Yes, I would describe my childhood as stable and peaceful” (Van Nie Kerk, E.2017). She also recalled how growing up there weren’t any major incidents that could have alluded to the fact that she might have a genetic condition such as Turner's Syndrome however, she did mention how people would constantly point out how short she was which was no call for concern at that point in her life thus her family didn’t think too much about this. Erna did really well at primary school and adjusted very well and had no problems making friends or interacting with peers. She had her very close group of friends and told me that her philosophy when it comes to friends she prefers quality over quantity.

She did mention that she struggled with maths as a subject at school saying that “I did struggle with maths but the rest was okay. Remember that during the 60’s and 70’s there were not any support groups and we didn’t really know about Turners Syndrome so for me I just struggled with maths.” (Van Nie Kerk, E.2017) which is something that a vast majority of women with Turners Syndrome do struggle with nonverbal cognitive subjects such as

maths as discussed in Zinn,(1999). This was also a particular problem I personally struggled with my entire school career and had to take maths literacy in High School. For Erna the turning point was in high school at the start of puberty, she recalls how her experience of puberty was very different to that of her friends because she was so much shorter thus she couldn't relate to their experiences and said that "My dad and gran weren't very tall people so we didn't we think too much about the height and attributed my height to that." (Van Nie Kerk, E.2017).

My second interview was with Cayleigh (18) a student from Centurion. Cayleigh is a very intelligent young lady with a passion for music and is a very gifted musician and can play three instruments and almost immediately one can feel that energy and passion when you speak to her. Cayleigh describes her childhood as "Very happy, there were a few down moments but more up moments than down moments. Exciting and adventurous, happy girl growing up" (Rounciville,C.2017). Growing up with two loving parents and a younger brother she was a content girl growing up. Cayleigh was a very energetic young girl and mentioned how she liked walking around during class and struggled to sit still which was not a problem at home but it did cause some serious friction with teachers at school which later resulted in her parents taking her to A.D.D specialist in Pretoria and eventually lead to her Turners Syndrome diagnosis in grade one which was that critical moment that started her journey.

Cayleigh also mentioned how she didn't have any problems with making friends as such but chose to keep her group of friends very small and close since she struggled with episodes of anxiety growing up and recalls being bullied at school "Bullying defiantly, fighting with friends, teachers in my matric year specifically. I remember this boy Kegan who would kick my chair and another boy said I smelt bad" (Rounciville,C.2017). In terms of her schooling and academics Cayleigh was strong besides for maths which is a common trait shared by many diagnosed with Turners Syndrome. Maths was something she had to work very hard at right through primary school and high school. As mentioned before Cayleigh is a talented and gifted musician that can play three instruments which this is where she found her comfort. Cayleigh's parents told her about her diagnosis when she was fourteen when they felt she was ready and gave her the option of when she wanted to go to the doctor and start treatment.

My third interview was with Britt (26) from Johannesburg who is a laboratory assistant at Lancet Laboratories. I got the distinct impression that she was highly motivated and intelligent. Despite being diagnosed at birth Britt described her childhood as follows “I would say that I pretty much had a sheltered childhood. I went to the same primary school and high school which I think made it easier for me. Because kids grew up knowing that I was different and I went through my entire schooling career with them which made it easier on me because they knew where I was coming from.

So, for them it wasn’t that much of a shocker. I loved my childhood. I didn’t feel different because I was never made to feel different. Being the oldest sibling also made it easier because they didn’t expect anything different from me”. (Carlse,B.2017). Britt was raised by both parents and has two younger siblings which were all contributing factors that worked together to form the stability needed in the household. Britt recalls being a happy girl that didn’t have any trouble making friends since she attended the same primary and high school and had the same group of friends, thus she didn’t have any trouble socialising.

Britt was a strong student who excelled in math and science and later completed her BSc at the University of Pretoria. However, she mentions that there were moments growing up that she did feel different and she knew that something was different about her she just didn’t know what until her parents told her about her diagnosis when she was thirteen. The Turners Syndrome diagnosis did actually answer some questions and as Britt describes it “Everyone has a something, now my something had a name” (Carlse,B.2017). Growing up I can completely relate to this idea of needing to know why I was so different and finally having so many questions answered and the relief those answers bring.

My fourth interview was with Carla (27) from Pretoria who was also diagnosed at birth. Carla grew up with two siblings and both parents and recalls having the quintessential happy and fulfilling childhood or as would say “I am very fortunate in that I had a close to perfect childhood” (Farreira,C.2018) and this which can be largely attributed to her parents knowing about her diagnosis from the start which motivated them to give Carla a normal and stable childhood which is a sentiment shared by Britt who was also diagnosed at birth. In both instances parents endeavoured to treat their daughters as normally as possible while they were growing up and didn’t treat them any differently.

Carla attended a regular primary school and didn't have any problems making friends or socialising with her peers and teachers, she also partook in numerous extra mural activities. However, she does mention that she was teased at school for her height and being shorter than her classmates. She went through a rough time from about the ages of nine until she turned eleven where her self- esteem and confidence was heavily affected which is something she battles with to this day because she is very conscious about how she acts around others and in public.

Being in an all-girls high school made a huge difference because she felt more comfortable in that setting. However, she did find that she would compare herself to her friends and found that her relationships with men and dating became a problem. What helped Carla a great deal was that her parents gradually started introducing the idea of Turners Syndrome to her until she was old enough to understand the finer detail. Her parents explained her diagnosis to her and once she was old enough she started doing her own research. Regardless of the issues with confidence Carla didn't struggle academically at all and after matric studied at the University of the North West and became a dietitian.

My fifth and final interview was with Liezel (42) from Kwa- Zulu Natal. As in the case of the previous participants Liezel also had a very happy and stable childhood and only recalls fond memories growing up with her mom and dad at home. During the interview Liezel did mention how her height was always a massive problem since she was always the smallest girl in her class which did result in some teasing and name calling in primary school. In one instance she remembers how the grade seven pupils would carry her around school.

Liezel did attend three different schools in different provinces which did impact the way in which she approached people and made friends, generally Liezel doesn't have a problem with making finds and communicating or socialising however, she finds it difficult to approach people at times. Liezel is another participant that was diagnosed at sixteen. She recalls how her confidence was affected in high school when her friends were starting puberty and she could not relate to her peers because she was not developing which is similar to the experiences of Erna and even shares similarities with my own story.

4.2 Diagnosis and Treatment:

In contrast to my first set of questions relating to the experiences each participant while growing up, my second set of questions focuses more on the actual process of diagnosis and the types of treatment available to those managing the different symptoms associated with Turners Syndrome. Here I would like to argue that the process of diagnoses is as much a personal journey as it is a daunting process of medical inquiry. The narratives that each participant presents are their own interpretation and experience of the process of diagnosis and should be viewed from the perspective of the individual thus placing emphasis on the context of the individual. Furthermore, factors such as access to information consulting with medical doctors, and when one is diagnosed with Turners Syndrome have been considered since it would implicate the type of treatment the participant received and the impact the diagnosis had on their lives and their families. This narrative pertaining to the diagnosis is imperative in analysing how the participants understood their diagnosis as well as what is Turners Syndrome.

Erna recalls how the reason why she actually went to the doctor with her mom was due to an irregular menstrual cycle when she was fourteen. In reality only, a small percentage of women with Turners Syndrome actually develop a regular menstrual cycle. According to Erna the doctor ran multiple tests for a range of different conditions and illnesses thus it was more by a process of deduction that she was diagnosed with Turners Syndrome. Once she received a diagnosis the doctor immediately prescribed hormone replacement therapy which was a daily injection which she is still on today only now instead of an injection she takes a hormone tablet. Almost immediately Erna embraced her diagnoses and started doing some research that would allow her to understand her condition.

Liezel has a very similar experience in the sense that her height was also never really a point of concern until she turned sixteen in high school and didn't show any signs of development or growth. As a precautionary measure Liesel's mother took her to a doctor who referred them to an endocrinologist that began the chromosome testing that eventually confirmed a positive diagnosis for Turners Syndrome. Liezel was also put onto hormone replacement therapy immediately but describes her initial reaction to her diagnosis as a type of denial when she says "It was as if I didn't want to know, I wanted to forget" (De Beer,L.2018). She also didn't tell anyone about her diagnosis and kept it to herself.

Being diagnosed at the onset of puberty is a common experience for a vast majority of women based on the fact that Turners Syndrome directly implicates one's growth and development however, there are instances where doctors and other medical practitioners are able to detect Turners Syndrome at birth or much earlier, as was the case for Britt and Carla where both diagnosed at birth. Britt mentions how her blood pressure resulted in the doctor running numerous tests which revealed her Turners Syndrome diagnosis, however her parents only told her that she had Turners Syndrome at the age of thirteen and she remarkably doesn't suffer from any of the associated medical problems such as thyroid and heart problems as well as hearing difficulties besides having the excess skin or webbing on her neck surgically removed.

This webbing of the neck affects only approximately 23% of girls with Turners Syndrome according to Zinn, (1999) and it depends on the individual whether they would want the excess skin on the neck removed since this is something very personal and very aesthetic or cosmetic. From a psychological perspective the removal of the excess skin can help develop that girls confidence and self -esteem. However, this does change from individual to individual and is dependent on a number of factors such as money and access to the correct doctors. Britt was in the fortunate position that afforded her the opportunity to have the surgery done as a means of boosting her confidence.

Carla was diagnosed when her mother's doctor noticed something abnormal on her sonar scan. According to Carla the doctor was not sure whether the scan showed the baby having either Downs Syndrome or Turners Syndrome and as a result they urged Carla's mom to abort the pregnancy but thankfully she refused the abortion. When Carla was born the doctor confirmed the Turners Syndrome diagnosis unfortunately she does suffer from other associated health problems such as a heart condition that needs regular check-ups as well as Scoliosis which is a severe curvature in the spine which had an impact on the medication and hormone treatment she was prescribed to help stimulate growth.

Carla described her experience with doctors and the diagnostic process thus far as "intense and traumatic" (Farreira,C.2018). Despite being diagnosed at birth Carla suffers from many of associated health complications such as Scoliosis, cardiovascular and hearing problems thus during the interview Carla mentions how most of her childhood was spent in doctor's rooms or in hospital. She saw multiple specialists and went for surgery to correct the scoliosis which caused a complication for the growth hormone treatment. Taking Carla's story and

expanding on her experiences, I can see how Turners Syndrome affects each woman differently thus it has various degrees of impact and effect and will affect each woman individually. For example, some women might be born with the psychical characteristics but not have any heart or hearing problems. The reactions by each individual can also be attributed to their circumstance and upbringing. Carla for instance was born into a family that could afford the treatments and surgery because they had Medical Aid.

Cayleigh initially went to the doctor because her nursery school teacher insisted that Cayleigh was hyperactive since she was such an energetic child. This started an intensive process of visiting multiple specialists and medical practitioners until Cayleigh's parents finally took her to a paediatric endocrinologist that did a bone age analyses which then confirmed that Cayleigh had Turners Syndrome and was subsequently prescribed hormone replacement injections. Cayleigh remembers how "The whole process of diagnosis was very rough and that just added to the anxiety. To this day I am terrified of needles. Going for blood tests every four months is a huge story" (Rounciville,C.2017).This is another instance where Cayleigh's family had access to information and specialists such as endocrinologists Her parents also only decided to tell Cayleigh about her diagnosis when she started high school at fourteen and gave her the option of when she wanted to continue with the treatment which included taking oestrogen hormone tablets.

What I have covered on this section is the process of diagnosis and the participants' experiences of being diagnosed and treated for Turners Syndrome. The next section that follows looks at what it is like living with a genetic condition.

4.3 Living with Turners Syndrome

The preliminary questions of the interview pertaining to early childhood and their experiences relating to the process of diagnosis laid the foundation for much of the interview. This provided me with valuable insight and the necessary context needed to gain a more comprehensive understanding each of my participants. However, my interviews did not just stop at childhood and diagnosis, I went one step by further going beyond diagnosis and treatment because I wanted to know what it was like living with this genetic condition and how this diagnosis was perceived. I wanted a frame of reference that went beyond my own interpretation and understanding of my experiences. Essentially what I wanted to know was how drastically Turners Syndrome changed the lives of each participant and their perceptions of themselves.

Each participant had their own individual struggles after their diagnosis, apart from the broad spectrum concerns such as infertility, self-esteem, acceptance and personal development. Sometimes even the most mundane tasks such as driving a car or trying to reach for something in the cupboard can be a cause of major frustration because it makes one feel helpless in a sense and you do feel like you have to depend on either having a step ladder or asking others to help with chores around the house. However, I would like to mention that in this instance the frustration was caused by the short stature.

Another point that was raised was the idea of buying clothes as being short you can never really buy close that fit and constantly need to have pants shortened or wear clothes that complement your height. During the interview Carla and I had a good laugh because at one point she mentions that “I was constantly losing things and went through I don't know how many pencils in grade one” (Farreira,C.2018) which is some I could relate to personally because I was also one of those pupils that forgot stationery or a jacket at school which I later found out is fairly common amongst women with Turners Syndrome.

For me personally I have always had a challenge with my weight so buying clothes has always been something that has bothered me which is a stark contrast to the participants who are all petite. These practical everyday tasks where a particular concern for Erna, Carla and Liezel. However, for Britt living with Turners Syndrome had a very unique perspective when she says “I didn’t want to look like a syndrome kid because I didn’t feel like one. And I was scared that I looked like one because I didn’t feel like one and that was my biggest thing and I still deal with that. I don’t want to look like a syndrome child”(Carlse,B.2017). From my

perspective, after interviewing the participants, I can argue that this group of women exemplify the idea that you cannot be defined or limited by Turners Syndrome.

As I have mentioned before, Turners Syndrome is not in any way life threatening or debilitating. The narratives of these participants are testament to how one can take a condition and turn it into something very positive. For example, Carla and Britt have university degrees from the University of Pretoria and North West University, and all of the participants have driver's licenses and have stable employment. Britt excelled at maths and science at school which are the subjects that girls with Turners Syndrome find difficult and Cayleigh can play musical instruments which are a fine motor skill that she had worked hard at and perfected, she also performs with a band regularly and is perusing a degree in Music.

The most poignant statement made by one participant that really summarised what is was like living with Turners Syndrome comes from Britt who said "Everybody has a something, only difference is my something has a name" (Carlse,B.2017). This, statement really highlighted and eloquently captured the general consensus of the participants which is that Turners Syndrome is not a disease or something that one should be ashamed of.

Each participant there was something that they have achieved or a passion they are pursuing. For example Erna who is currently a Sunday school teacher, does community service with an animal therapy group known as *Paws for People* where her dogs are used as part of therapy at old age homes and hospitals. As mentioned before Cayleigh has just finished her matric and is pursuing her passion for music and performs on stage with a band regularly. Britt has a degree in human genetics from the University of Pretoria and is working at Lancet Laboratories. Carla has a degree in dietetics and is a qualified dietitian and is part of a very successful practice. And finally, Liezel is a mom and wife and considers that to be her greatest achievement despite the challenges of Turners Syndrome. For Liezel specifically, her daughter is her pride and joy thus the infertility was a huge challenge that she had to overcome despite her husband supporting her. This was something she felt she had to deal with personally.

For a vast majority of the participants there were no major issues as many felt that they were not any different from other members of their communities. When I asked my participants if they felt any different and the responses varied from some being concerned about their height while others felt a sense relief that they now knew what was wrong with them, Erna

mentioned that “Not that I can remember but what I do know was that I was always the shortest in the group in both primary school and high school.

There was never a problem with bullying except in primary school where they teased me about my glasses. Because there weren’t any support groups or much written about Turners Syndrome I was only diagnosed much later in Life at seventeen so we didn’t really realise that there was something different or wrong with me. My dad and grandmother weren’t very tall people so we didn’t we think too much about the height and attributed my height to that.” (Van Nie Kerk, E.2017). Thus for Erna she didn't really let the height affect her which is very similar to Cayleigh who said “I just knew that I had this and that I had to live with it. A lot started making sense and I realised why I was a little different to my friends.” (Rounciville,C,2017) so for her and her parents it was almost a relief that she knew what was wrong so that her parents knew how they could fix it and what they had to do to make her life as ‘normal’ as possible.

I noted that Britt was very comfortable in her skin however her major concern was “I don’t feel like I have a syndrome to be honest because I still do what people who don’t have Turners do. My biggest thing is that I don’t want to walk down the street and someone says ‘oh she has this syndrome .I didn’t feel any different at all, for me, I don’t know if it was the way my parents raised me but I thought ‘ah! Cool’ this is awesome I have something no one else has and something that makes me more unique and that my unique has a name” (Carlse,B.2017).

This shows how she really embraced her difference however she was concerned with looking like the phenotypical women with Turners Syndrome. For Carla it was the reaction to her height that really caused problems and according to her “Obviously the height thing made me look different and children can be nasty and when someone doesn't understand something and my friends babied me for a long a time because I was small which did make me feel different” (Farreira,C.2018).

4.4 Infertility

Apart from the more obvious features associated with Turners Syndrome such as the short stature and webbed neck; infertility is something that all women with Turners Syndrome struggle with. This infertility is a result of the body not producing sufficient hormones such as oestrogen and progesterone which affects the body's natural ability to produce and release eggs from the ovaries. Apart from the production of hormones the body also does not develop a regular menstrual cycle. From a more personal point of view infertility is something that I have grappled with and have had to work through in more recent years.

Since infertility is a reality for women affected by Turners Syndrome; I assumed that this would actually form the majority of the discussion during the interviews since fertility and having children is such an intrinsic part of the society in which we live. What I noted was that the participants were comfortable speaking about their infertility. I truly got the sense that they had made peace with their condition and being infertile. The participants almost seemed optimistic because immediately when I started speaking about infertility four of my participants went into a discussion about the alternative options available to women who can't fall pregnant naturally.

Erna spoke about how she also initially had to learn to accept the fact that she was infertile. In her instance what made this process exceedingly difficult for her was her family and in particular her mom and grandmother. She recalled having a conversation with her mom shortly after being diagnosed where her mom told her that she should not tell anyone that she was infertile which is something that she will never forget because, it was as if the infertility should be hidden. Erna also mentioned how shortly after getting married her grandmother came to visit and while she was sitting outside talking to her gran she suddenly realised that she could not continue the family lineage and give her grandmother any great grandchildren which still affects her at times. After trying for an adoption which was a process that Erna described as "difficult"(Van Nie Kerk, E.2017).

The application and screening process for the adoption took almost seven years without any success. Erna and her husband decided to approach an orphanage to volunteer as foster parents for one of the children, which meant that they could look after the child on weekends and holidays. Essentially what this provided the child with was a sense of security and family and to this day the little girl that Erna and her husband looked after all those years ago is still in constant contact with them and still refers to them as mom and dad.

When I asked Erna about whether she and her husband ever considered fertility treatment or egg donations she simply said that it was too expensive and that many women have approached her willing to donate their egg cells but for Erna this is complicated and expensive. She distinctly recalls a women that she worked with her approaching her offering to be a surrogate for Erna and her husband and all that Erna thought was "It will never be my child" (Van Nie Kerk, E.2017) .If one were to reflect on this we can understand why Erna opted for the foster care rather than egg donations because she felt that she was never going to have that biological connection to that child.

Cayleigh has just finished matric thus children are not her primary focus at this stage however what really impressed me about Cayleigh was how mature and optimistic she was when we spoke about infertility saying that "Like, honestly if I can have kids on my own I would prefer that but otherwise I would rather adopt a kid and give them a home which is something I would love to do since there are so many kids who don't have a home" (Rounciville,C.2017). For Cayleigh it is all about being comfortable in your own skin and accepting the infertility because it is something you cannot change. Being diagnosed with Turners Syndrome at a young age also contributed to the positive approach Cayleigh has to having children in future because her parents always emphasised and focussed on raising Cayleigh with values such as love and acceptance.

Britt was another participant that did not allow the notion of infertility to define her and affect her. What really fascinated me about Britt was that during the interview she mentioned that. "It is obviously something that I have thought about. It's funny because even as a kid before I found out that I couldn't have my own children I remember playing the Sims computer game and I always wanted my Sims to adopt. I have always liked the idea of adoption. So maybe it was me protecting myself unconsciously, or the bodies' way of knowing what it can and cannot do. So, for me adoption was something I have always wanted to do even before I found out about the diagnosis. Your body knows what it can and cannot do. My Fiancé is very much for the adoption idea, said I was willing to try the IVF root if he wanted something connected to him. He always said that infertility is something that could affect anyone and cannot be held against me. Wes would say it won't make me love you anymore or any less. I told Wes on the second date and I just asked him if this was something that he could deal with and if we could carry on with this relationship because I cannot pretend to be something I am not and told me as long as I wasn't dying he couldn't care if I could have kids or not" (Carlse,B.2017).

Essentially what Britt suggested is that your body knows its natural limitations. It was as if she found acceptance by embracing her body and by accepting her infertility even before she knew that she had Turners Syndrome. She recalled how from a young age she was always fascinated by the idea of adoption and still firmly believes that adoption will play a role in her future. Similar to the previous two participants Britt also felt her family was the biggest challenge because it was as if they took this infertility personally and blamed themselves.

On further analyses what Britt is saying is that egg donations and surrogacy are not the only option available to young women who are infertile, you can find acceptance and comfort in the fact that there are many women who actually struggle to fall pregnant that do not have Turners Syndrome. This also links to how young women at present are more connected and have access to information, thus their priorities have changed with regards to having children and their perspective of having children has also changed. They are more focused on their careers and their education as opposed to their grandmothers and mothers who saw family and children as a priority hence why Britts' family took the news of her diagnosis personally.

Carla has also accepted her infertility and feels that a woman's worth should not be determined by how fertile she is but rather by the strength of her character. Carla and I had a discussion about how the role of women in society is slowly changing and the reality is that women are actually choosing to study rather than start a family straight away. Another interesting point that Carla raised was that there are women who struggle to fall pregnant or who are infertile who don't have Turners Syndrome thus she found acceptance or solace in the fact that infertility is not something exclusive to this genetic condition.

The participant who was most affected by her infertility was Liezel who was devastated when she found out that she couldn't have children. As mentioned before in the previous section Liezel initially didn't take her diagnosis very well and as a result she didn't research Turners Syndrome any further after she was diagnosed. Liezel only really started doing some research on her condition once she was married and naturally she was shocked to find out that she was infertile. However, Liesel's story did have a much happier ending when she and her husband decided to adopt a baby girl who is now in high school and Liezel says "I can't imagine life without her" (De Beer,L.2018).

What I can add here is that the narratives pertaining to infertility were actually very positive and uplifting. This is what is referred to as a narrative focused on hope and restitution. What is worth noting is that the vast majority of my participants are opting to adopt instead of egg

donations. At first glance adoption seems less traumatic in the sense that one won't have to rely on doctors. With egg donations there is the risk of the procedure might not work whereas with adoption you giving a child a chance to have a happy fulfilling life which ties into the idea of wanting to give that child hope.

4.5 Turner Syndrome and Love and Relationships

Four of the participants are all in loving and committed relationships and have found partners that have accepted them for who they are. Erna, Liezel and Britt are all happily married and Carla is engaged. During the interviews I noted how each participant had struggled with confidence and self- esteem thus intimate relationships become a challenge especially when one needs to tell your long-term boyfriend, fiancé or husband that you have a genetic condition that affects your fertility. In the case of Erna, she was instructed by her mother not to not tell her future husband about her Turners Syndrome diagnosis until after they decided to get married but added "You know, I always say that it is so amazing because when I was sitting there in the doctor's rooms I immediately thought 'Lord the man that accepts me will accept me for who I am and will marry me regardless. You obviously need to deal with this but it wasn't such a big deal for me. I immediately worked through it and dealt with it. You have a normal life 'finished end klaar' the only thing you don't have is kids" (Van Nie Kerk, E.2018).

Liezel on the other hand only told her husband about her diagnosis after they had gotten married and started doing some research and found out that she was infertile which she describes as a devastating moment for her and her husband. I can draw from my own personal experience of telling my fiancé, who is now my husband that sharing your diagnosis with someone that you potentially would like to start a family with takes and immense amount of courage. I remember being so nervous about telling my husband because I thought he would be disappointed by the prospect of not having any kids yet when I told him he almost immediately googled Turners Syndrome on his phone and started reading articles.

I think I speak for myself as well as for the participants when I equate this anxiety of revealing your diagnosis to your partner with the fear of being rejected because you are afraid that your partner will not see you as feminine enough if you cannot have children.

4.5 Gender

When one takes the above narratives and discussion into consideration there is one crucial aspect that brings them all into one conversation. While doing the interviews I noted how each participant negotiated their individual identities, and more importantly their *gendered identities* starting from early childhood right through to when they were diagnosed with Turners Syndrome and how that affected their identities. From my point of view the notion of *gender* is something that permeates to the different parts of how we develop our identities.

With Turners Syndrome in particular it is interesting how the participants actually had a very stable idea of who they are regardless of when they were diagnosed. None of my participants ever questioned their femininity and a statement that really stood out for me was during the interview with Erna when she said "There is nothing I can do about it. I am the type of person that if I cannot change it then I accept it. I never felt like less of a woman, my mom wouldn't allow it. She always said we must dress neatly and put on some makeup, I was a bit of a tomboy" (Van Nie Kerk, E.2017).What is very interesting in this statement is how her mother was getting her to 'perform' her assigned gender by getting her to act more feminine, which ties back to the discussion on *sex* and *gender* by Fausto-Sterling and Butler where we initially questioned how 'natural' *sex* and *gender* where in the first place. This also links to how the older generations understood their own femininity thus wanted to pass that on to their daughters. For Erna's mom for example it was all about appearing feminine.

I noted and was not surprised to find that the participants were all raised as girls despite their diagnosis. Besides the diagnosis and missing genetic information there was no other apparent reason that these participants would not have been raised as women otherwise, even if they were diagnosed at birth or at puberty this would have not changed the fact that they were raised and socialised into a female role. From the context of the participants there was no grey area with regards to being female. The one aspect that is put into question is the issue of infertility It was almost as if each participant separated their femininity and their identities from the Turners Syndrome and treated the two as completely different aspects of their identities.

Thus, what I can deduct from the interviews is that your identity as a female is not necessarily coupled to being diagnosed with Turners Syndrome, making this argument

unnecessary. The question and argument pertaining to *gender* I have struggled with and almost became something personal to me. I based my argument largely on the premise that infertility played a central role to ones identity; however infertility is only one aspect of one's feminine identity. A point that came across clearly was the argument that these participants defined themselves as women despite being diagnosed at birth or at puberty because they were raised as girls by their parents. It was important to note that infertility, which I assumed would be a majour factor in how the participants viewed themselves however based on the interviews the argument around ones *gender identity* being linked to infertility, did not actually play such a significant role.

I can argue that *gender* is in fact a conscious decision that we actively participate in and subscribe to so that we can ensure our place within a community so that we are not marginalised and separated from our community. This is exemplified in the interviews when the participants speak about their childhood and how they were raised by their parents. From a personal perspective this can also be attributed to each participant being in hetero-normative intimate relationships. As members of society we are firstly taught how to behave as either a male or female, and as we grow older we constantly adjust our behaviour to fit that definition and understanding. In this case each of the participants regardless of being diagnosed at birth or at puberty have never questioned or doubted their femininity. This just reiterates the point that they are female, thus that is how they have understood themselves and how society always viewed them as female, and by behaving and acting in a certain manner and conforming to certain social norms and expectations associated with being a female(eg: getting married)

What I observed while speaking about infertility was that the participants had separated their infertility from their feminine identity. They did not consider infertility as an essential part of their identity because they knew about the alternatives available to them; many spoke about adoption or IVF. What also contributed to this was having a support system that did not purely focus on the fact that they were infertile. I can hence argue that having children and fertility one aspect of the feminine identity but is however not an exclusive part of one's feminine identity. There are a multitude of other aspects to ones feminine identity that one could choose to focus on.

4.6 Support

I have mentioned before in the methodology chapter of this report that each participant was recruited from the Turners Syndrome SA WhatsApp group which is a support group I started in 2014. The main idea behind starting the Turners Syndrome SA WhatsApp and Facebook groups was initially to try and connect with other women who had the same genetic condition as I had. As the group gained momentum I soon realised that there was a major lack in support for these women. At this point I would like to clarify that when I use the term support I am referring to the guidance and assistance given by doctors, gynaecologists and endocrinologists as well as by psychologists or genetic counsellors.

During the interviews a common experience shared by all five participants was that doctors offered a very basic type of support and would often refer to a medical text book that explained what Turners Syndrome was, or would then urge their patient to go and research Turners Syndrome on the internet. Apart from the support given by doctors what was also noted was that none of the participants were referred to either a genetic counsellor or psychologist. Instead they were told to see an endocrinologist for further treatment which is very necessary however the support an endocrinologist can offer is limited.

Hence why I started the WhatsApp group as a means of helping women with Turners Syndrome talk about the more personal and emotional challenges faced by someone who is diagnosed with a genetic condition. When I was diagnosed at sixteen all I wanted was a space or platform to talk to someone and share my experiences and to talk about how Turners Syndrome affected my life and the lives of my family. A support group places emphasis on the individual's narrative and focuses on what that person has experienced which I argue plays a vital part in analysing illness and disease from a completely new perspective. Speaking to each participant I understand that patient support groups provide a space where these women feel comfortable and can relate to other women with the same condition.

Another point raised during the interviews was that the families and partners of women with Turners Syndrome should also be involved in the process of support as many of the participants mentioned how their parents or grandparents felt guilty or responsible for the diagnosis. In terms of finding information most of the participants actually did their own research in their own capacity or in the case of those diagnosed at birth the parents also did their own research either on the internet or in a textbook. All of my participants commented on how sparse the information given by doctors was as Britt mentioned "Once I was

diagnosed my parents looked for a support group and they how we found Jo-Anne and Emma. There were one or two meetings but I felt like I didn't really need it because I had my friends and family. I felt like I didn't need that outside support because I had enough support. My Family are my rock and If anything, I would like more support for them. Because they if they can handle it I can handle it and because they were so strong I was able to be strong."(Carlse B.2017).

For the younger participants information was much easier to acquire and the time in which they were diagnosed was much more accepting as opposed to the two older participants that struggled for information and trying to find information while their diagnosis was shrouded in silence and secrecy as Erna would say "The doctor didn't tell me much. It was almost a situation of here is your diagnosis, goodbye! In those years they didn't even know about Turner Syndrome and if they did they wouldn't talk about it." (Van Nie Kerk, E.2017).

For each of m participants family was absolutely critical in giving support and played a vital role in proving the emotional support needed to cope and thrive despite being diagnosed with this condition. Apart from the emotionally support families provided a sense of normality and continuity need to develop a positive self-esteem and outlook in life so that they can become a fully functional and integrated part of society. For me personally I know my family plays an integral part in motivating me and providing me with the support I needed to succeed at school as well as in my tertiary education which is something that Britt, Caleigh as well as Carla mention during their interviews. Another crucial aspect was that each of the participants came from a stable home and a secure middle class family that could afford medical and treatment. Thus having the financial and monetary resources to see expensive doctors and specialists plays a role in how you as a patient would interpret and manage your condition. All of the participants were fortunate enough to have access to treatment, which made the process of accepting the diagnosis much easier.

4.7 My Role as the Patient and as the Researcher

Initially when I started with this research project I was very nervous about how my role would be defined within the context of this report. Since this project is an academic and analytic work which requires the researcher to be as objective and impartial as possible when conducting their analyses. How was I going to connect my own experiences and ideas about Turners Syndrome to the narratives and experiences of the participants without any bias? And how was I going to remain impartial while interviewing my participants when this project is something very personal to me as a researcher?

Initially I wanted to use my own story and experiences as a source of inspiration for the project and completely distance myself from the role of someone diagnosed with Turners Syndrome because after all, I was now a researcher that was committed to her academic work. However, I soon realised that in fact I can contribute just as much to this project as both researcher and participant based on the fact that I have what is referred to as "Analytic Reflexivity" (Anderson,2006) meaning that as a researcher I was cognisant/conscious of my connection to the research topic and the effects I have on each participant since we share a diagnosis.

According to article written by Leon Anderson,(2006) from Ohio University this notion of analytic reflexivity is driven by the researchers desire to understand themselves as well as their participants by using a dialectic conversation where you as the researcher look at your own actions and perceptions with reference to the conversations and narratives of your participants. Thus, there was a way in which I could include my own thoughts and narrative with the narratives of these participants.

What I found was that the participants were actually very receptive once I told them that I was diagnosed with Turners Syndrome. The participants were more comfortable talking to me once we had established that common ground. Because Turners Syndrome only affects a very small percentage of the population one never actually considers the fact that you might actually meet someone with the same condition but once you do there is an immediate similarity that connects you and you know that you can relate to that person. This also played a huge role in establishing a positive connection with each of the participants because to them I wasn't just a researcher, but someone that they could relate to and that understands them.

I approached this project with the hope that as much as I could learn from each participant that they could also learn from me and that I could have a positive impact on their lives by just sharing our stories and experiences. There were moments during this research project where I had to remind myself of my position as both patient and researcher and recognise when I was Nadine doing research and when I was Nadine the patient with Turners Syndrome.

Once I started listening to the participants and drawing parallels to my own experiences and my own struggles with confidence and low self -esteem I felt a sense of peace and hope knowing that I wasn't alone. This project really forced me to do some serious introspection and self- evaluation. And in this process, I learnt how to see my own opinions and experiences from within the context of a broader society and community of women living with Turners Syndrome that I was now a part of. This has honestly been one of the most rewarding and therapeutic and cathartic experiences. Not only has this project taught me about research and Turners Syndrome but it has also taught me a lot about myself and has certainly given me a new perspective.

Chapter 5:

Discussion of Findings and Concluding thoughts

In this chapter I will discuss some of the observations and arguments made each participant which will also include my own analyses and interpretations. My hope is that the narratives of these women and the findings of this project actually create a platform for discussion for other women diagnosed with Turners Syndrome as well as for patients with other conditions and that its shows that there is strength in their narratives.

My first observation is in relation to the more practical elements of using narratives as a means of analyses. From my experience of doing the interviews I noted how the narratives of the participants operate on two levels that actually work in unison to inform the participant narrative. The participants also seemed to place an emphasis on shared experiences. During the interviews they would constantly refer to an article that they have read or would asked me what my experiences have been as someone who is also living with the same condition.

Each narrative was informed by shared experiences of each participant living with Turners Syndrome, such as the notion of infertility or being on hormone therapy treatment.The participants also spoke from their own personal and individual experiences particularly with reference to their experiences with their diagnosis and doctors and how they interpreted Turners Syndrome and how they chose to process the information that they had just been given. Another deeply personal matter that was discussed was the notion of infertility and each participant had their own way in which they interpreted the fact that they were infertile.

The two older participants had a particularly important role here as they had already gone through the motions of adoption and caring for a foster child. They had a very different idea of what infertility meant for them as a parent to an adoptive or foster child as oppose to the younger participants. What each individual narrative alluded to was the idea of context and perspective of a narrative. The participants' background and upbringing was imperative as a means of understanding their story and how they would interpret certain questions differently.

From the perspective of the researcher the context of a narrative helps you understand and gain a better insight into each of each of your participants. To analyse the context of a narrative is a means of collecting important details and information that helps you understand why your participant chose to focus on a particular topic or why the participant has a particular point of view and what factors influenced a particular instance. With reference to this research project the notion of context would extend to a discussion on how each of my participants viewed Turners Syndrome in that particular moment.

What was particularly interesting was the different experiences between the younger and older participants. Firstly, one has to consider the broader contextual elements such as the historical context and period that informed the experiences of the older participants. This was particularly interesting as both older women were diagnosed at a period in time where medical technology was not as advanced thus there was no chance of being diagnosed with something like Turners Syndrome at birth or in utero. I noted a very stark contrast between the older and younger participants was their reaction to their diagnosis. The younger participants were more accepting and comfortable with the diagnosis whereas the older participants were either in complete denial or had to take time to accept their diagnosis. This relates to the generational aspect were younger participants had access to more information and better health care making the situation easier.

Even the information on Turners Syndrome available to patients and doctors during the 60's and 70's was also scarce and very limited which had an impact on how participants reacted as patients and how their families reacted to the diagnosis. This lack of information and awareness about Turners Syndrome was also detrimental in so far as there also was not sufficient support available to these women. There weren't any support groups or ways of contacting other women with the same condition. The moment at the time of diagnosis also had an impact on these women were viewed in society. By taking the historical context into consideration as a researcher one could understand why the mothers the participants felt a sense of guilt or angst when they heard that their daughters where infertile for an example.

I would like to specifically refer to the stories of Erna and Liezel who were told to either keep quiet about their infertility or just continue with their lives as if nothing was wrong and almost deny their diagnosis. My interpretation of this reaction would be based on the fact that women had a very specific place and role within society at that time which

predominantly centred around being a wife and mother and now being diagnosed with Turners Syndrome meant that they would have difficulty fulfilling that role.

This discussion likes directly to the generational aspect I alluded to in the previous section when I compared the different reactions to the initial diagnoses. What I can also comment on here is the reaction of parents, in particular the reactions of mothers. For the older participants their mothers had very negative reactions to their daughters being diagnosed with Turners Syndrome, which can be attributed to the fact that women had a very specific place in society at that time. Their daughters did not necessarily adhere to the very stringent ideas of femininity during the 1960's and 1970's. If one were to discuss this point further the mother's reaction reflect changing roles and expectations around *gender* in society.

The second aspect of the participant's narrative that I chose to focus on was their childhood and background. As I have mentioned previously each of the participants come from white middle-class families living in various parts of Johannesburg and Pretoria. This also meant that they had access to decent healthcare and medication meaning that their narratives where predominantly very optimistic and hopeful. This is of particular importance especially in a country such as South Africa that has one of the highest rates of inequality. For example, if I had interviewed someone from an underprivileged background and living in a township their experiences of Turners Syndrome would be significantly different. Each participant was in a position to access information about Turners Syndrome, and access the available hormone treatment.

Essentially what I wanted to extract what specific events or situations played a role in leading up to their diagnosis. Furthermore, I wanted to analyse what life was like for each participants before and after their diagnosis. What I found was that the participants each had very stable and 'normal' childhoods and grew up in happy homes. Growing up in a household with both parents and siblings meant that they had a strong foundation that helped in their development. This was particularly important for Britt and Carla who were both diagnosed at birth which meant that they needed strong sense of family, and a stable support structure to development as normally as possible.

Thus, here I can raise a more reflective question pertaining to how their experiences might have been different without this stability given by family and well as the, financial security.

This is what has attributed to each participant being able to accept and manage their condition.

At this point I also need to mention that all five of the participants insisted that they were 'normal' thus the Turners Syndrome diagnosis didn't have much of an impact on how they viewed themselves. Three participants recalled how there were days that they compared themselves to their friends, while others mentioned that they did struggle with their confidence and self -esteem. Apart from the issue of confidence there weren't any major problems with the development of their identities that were stable. One could argue from an analytical point of view that the reason why there was such a strong emphasis placed on being 'normal' that could potentially reveal that they did in fact experience some anxiety from feeling different There was actually a very strong sense of needing to blend in to society.

As I have mentioned previously all of the participants were raised with a strong feminine identity. What I found during the interviews was that there was a definite continuity in their identities even after their diagnosis. None of the participants ever doubted or questioned their feminine identity. This can be attributed to the fact that they had a strong foundation and support structure at home. From a personal perspective I never doubted my own identity because I just told myself that the Turners Syndrome doesn't change who I am.

From the perspective of the researcher the insight into the participants' childhood helped me understand how they were able to negotiate their role within their community after diagnosis. I questioned how the participants were able to maintain a level of continuity while living with Turners Syndrome. This continuity can be largely attributed to each participant engaging with extramural activities and extended members of their community.

For example, involving yourself in something like a sport or even acting classes will teach you vital social skills and also give a certain level of confidence. For example, Erna takes her dogs to hospitals where they are used as part of a therapeutic programme. Cayleigh plays three musical instruments and regularly performs in front of crowds. As for myself I played hockey at school and was involved in acting classes which helped me make friends and prove to myself that I could do exactly what everyone else can do. This largely has to do with being integrated into a community that had helped build each participant develop a strong sense of self.

Another important point of discussion is the process of diagnosis and what each of my participants experienced before they found out that they had Turners Syndrome. The aspect that I focused on with regards to diagnosis was on the differences between an early diagnosis in comparison to being diagnosed much later when girls generally start puberty. The two participants that were diagnosed in high school mentioned that medication and treatment play a vital role when one has a discussion on the time of diagnosis.

Since the participants are middle class South African citizens I can draw a link to the discussion on class and access to private healthcare. Having financial and social stability will have a massive impact with regards to how one experiences having this condition. The earlier one is diagnosed the earlier one can start treatment and get examined for other health complications such as the heart and thyroid problems associated with Turners Syndrome. Early detection is also dependent on availability and access to information and medical specialists such as endocrinologists and general awareness about Turners Syndrome which at the time of their diagnosis was a huge problem.

I have already established that the two older participants were only diagnosed later in life because no one knew what Turners Syndrome was at that stage, the only information they received was from their doctor that explained their diagnosis from a textbook. The participants that were diagnosed at birth were gradually introduced to their diagnosis by their parents and slowly started treatment. The participants diagnosed in high school had to absorb a substantial amount of information and still adjust to the fact that they had Turners Syndrome in a shorter period of time. However, I do feel that as much as one needs time to accept the diagnosis there is a personal element attached to that. Your reaction to a diagnosis is also dependent on you as an individual and how intense the diagnostic process was. What I can infer from the interviews and from my own personal experience is that the process of diagnosis is never easy or definitive no matter when you are diagnosed.

For example, Erna and Liezel were tested for other conditions before the doctors decided to test for Turners Syndrome. Cayleigh had to start by going to an A.D.D specialist and numerous other doctors before going to an endocrinologist that finally diagnosed her with Turners Syndrome. What really surprised me was that not one of the doctors would diagnose Turners Syndrome immediately, which just reinforces the notion that we need more

information and awareness about this condition. I argue this from the perspective that doctors should diagnose their patients much earlier so that these women can receive the necessary support, guidance and treatment. This is where the narratives of these participants play a role because these narratives can actually be a useful point of reference for other patients that are recently diagnosed, as well as doctors. I argued this because one can actually learn from the narratives of women who have already been diagnosed, one can learn from their experiences as well as what support is available to them.

Support is a very brief but also a very important aspect of Turners Syndrome that should be expanded on. Each participant described how, after their diagnosis, the support that the doctor was able to offer them or was very limited. The participants mentioned how doctors need support because they need to feel understood. From the participants perspective they need to feel like there is someone listening to them. A support groups helps in so far as it provides the patient with a platform to share their experiences and to feel connected to others. A diagnosis can be very daunting and very isolating especially with a genetic condition. I got the distinct impression from these participants that the type of support needed for women with Turners Syndrome has to be holistic in nature. It has to focus on the personal and psychological aspects and should also include families and partners.

My argument is as that Turners Syndrome is seen as a chromosomal anomaly that affects the second x chromosome. As a result, one cannot be biologically defined as female because these women do not share the same genetic structures or hormones as other women. Despite not being biologically classified as female women with Turners Syndrome are still raised as females and socialised into the female role; this was exemplified by the participants who have all been socially accepted as women. What I can also mention here is that these participants do not ‘hide’ their condition from anyone especially from their intimate others. What I have in fact learned from the participants is that your identity as a woman is not necessarily determined by how many children you have. Just because these women cannot have children that are biologically theirs also doesn’t devalue their feminine identity or position as a female in society.

I argue that instead of seeing the feminine identity as a hegemonic concept that excludes exceptions from the accepted definition of what is considered feminine or female; we should rather see the feminine identity as forming part of spectrum. I argue this especially in relation to the notion of infertility where women have used alternative methods to fall pregnant. What

I observed was that while speaking about infertility was that the participants have separated their infertility from their feminine identity thus; they did not equate the fact that they could not have children to their identity as women. This was largely based on the fact that there are alternative methods available such as adoption, or make use of IVF should one choose to have a baby in future. I noted that the participants had a very positive attitude towards their diagnosis which they attributed to having a strong support structure and loved ones. Here I would like to reiterate the argument made in the section on *gender* were I state that fertility and the ability to have children is but one aspect of one's feminine identity, thus infertility doesn't play a significant role in how the participants viewed themselves. They had never questioned or doubted their femininity despite the fact that they were infertile.

Conclusion

This research project began by firstly outlining the more conceptual elements such as the aims and objectives of this project as well as the rationale. The core aim of this project is to raise awareness about Turner's syndrome, as well as to expand on current research with emphasis being placed on the importance of narratives and how narratives contribute particularly to our understanding of experiences and interpretations of living with Turners syndrome. My hope was to provide future researchers and other patients diagnosed with Turners Syndrome with balanced information so that we can expand our analyses and deepen our understanding of the condition. Thus, I interviewed and collected the narratives of five women living with Turners syndrome.

The rationale of this research project was based on the fact that I couldn't find any tangible research being done from the patient's perspective. By identifying a gap in the literature I realised that patient narratives have the potential to be used in the discussion of issues such as the how these women developed their identities despite their diagnosis. Essentially what I saw was a more practical way of using narratives as a means of viewing how biology and society interact. I argue that we must develop new perspectives and go beyond the restrictions of medical and physiological knowledge which limits research and analyses.

I refer to patient narratives in the report based on the fact that it is essential that one gains a deeper understanding of what it is like living with a genetic condition from multiple perspectives including the perspective of each participant as a patient , and how this impacts various aspects of one's life which is discussed in the findings sections. When one considers hormone replacement therapy and specialists this relates directly to the experience of the participant as a patient.

Chapter two focused on the practical and methodological parts of this project. I identified my project as being qualitative in nature considering that I was going to be using the narratives and experiences of the participants as my primary data. More specifically I did an auto-ethnography or applying the auto-ethnographic research method because I have integrated my own experiences of living with the Turners Syndrome into the study. As mentioned in my objective I collected my data by means of an interview with each individual participant and developed an interview schedule that covered a range of topics which included the participant's childhood to their diagnosis, interpersonal relationships as well as addressing

issues of infertility. In findings by expanding on the notion of narratives and context. Essentially I argue that a narrative, or in the instance of this project the narrative of the participants, is of great value to us as researchers, particularly when it comes to understanding the experience of being ill or diagnosed with a genetic condition such as Turners Syndrome. I argue that there is real value in understanding these narratives because it gives a unique perspective to research and analyses for researchers. This research done on Turners Syndrome in particular is of value because the narratives of the participants can actually be of great value for people diagnosed with the same condition. Instead of relying on doctors reports, people diagnosed with Turners Syndrome now have access to the narratives and lived experiences of the participants of this study to give them a new perspective and understanding of their diagnosis and how they can approach living with a rare genetic condition and how this impacts on their lives and their sense of self.

My literature review covered literature from three major areas namely, literature on Turners Syndrome as well as *sex* and *gender* and finally narratives. I tried to frame Turners Syndrome as a condition within the arguments that related to broader discursive social issues. What I noted however was that there was very little literature from a South African perspective such as availability of information and access to treatment and general awareness about Turner's syndrome as a condition.

The following two chapters presented my findings which I summarised into five major sections namely early childhood and development, diagnosis and treatment, living with Turners syndrome, infertility and finally support which were the sections and themes I covered in my interviews. Based on the narratives collected in the interviews I can conclude that Turners Syndrome did have an impact on the lives of the participants in some way. The one participant mentioned how very mundane tasks such as cleaning .driving or finding clothes became a challenge. None of the participants struggled at school besides finding maths a challenge. Socially I noted how the participants had no problems making friends but decided to keep their group of friends very small.

In terms of finding love and being in relationships all of the participants are in stable marriages and relationships were their partners support them. There was some anxiety expressed around disclosing their diagnosis to their partners as many felt that their partners would not see them as female anymore or reject them because they cannot have children. However, none of the participants reported any negative experiences. The biggest challenge

for most of the participants was with family as many of the participants spoke about family members having a sense of guilt of feeling responsible for the diagnosis.

All of the participants spoke about the challenges pertaining to the process of diagnosis and the hormone treatment.Thos links to the portion of the report were I speak about a patient narrative.Being diagnosed with this condition coupled with speaking to doctors is what informs the participants narrative as a patient. Furthermore, speaking about the process of diagnosis links to the point made pertaining to access to specialists such as Endocrinologists and the expensive medication which is a financial burden and within the context of South Africa only available to a small group of individuals to have access to private healthcare and medical aid.

In terms of the conversation around *gender* and identity it can be noted that none of the participants actually ever doubted or questioned their role as females based on the fact there was no other factor, besides genetics, that could have bought their feminine identity into question. The discussion around infertility was a minor and participants seemed to have separated their identity as females from their ability to bare children. This is largely based on the participants having a very positive and open attitude towards having children and almost all of the participants noted the alternatives available to women who want to fall pregnant. Linking this to the argument around *gender* I can conclude that one's ability to have children is but one aspect of one's feminine identity and thus is not as significant in this argument.

In chapter four I developed and expanded the discussion into an analysis of what it was like living with Turners Syndrome. In this chapter I considered variables such as the historical or biographical moment in which the participant was diagnosed. I looked at whether participants were diagnosed at birth or at the onset of puberty. The two elder participants, including myself, were diagnosed at puberty largely based on the fact there wasn't much awareness around Turner Syndrome and because the symptoms are only visible when one reaches puberty. The other participants were diagnosed earlier either at birth or at six years old. The age of diagnosis is important to this project because it connects to other important facets such as when to commence with treatment.

The second part of the chapter centred around an argument pertaining to how Turners Syndrome problematises notions of *sex* and *gender*. As mentioned throughout the report the women diagnosed with Turners Syndrome do not share the same genetic makeup as other

women however, I argue that despite not being ‘genetically female’ these women are still raised as females and consider themselves to be female. Each participant was raised as female regardless of when they were diagnosed, thus it can be argued that there no other factors or obvious reasons that could have changed the fact that they were female and have been raised as female.

Having a chromosomal anomaly does not mean that you are a dysfunctional human making you unworthy of your place within society.. The fact they are living with this rare genetic condition didn’t hinder them in their pursuit of success and happiness which is critical to emphasise. Thus to conclude I approached this research project with the hope that it would help raise awareness around Turners Syndrome as a genetic condition and promote further research. What I can comment on is how Turners Syndrome is a condition that is completely manageable , however. your external circumstances such as your economic position, access to information and medication , your family structure all play a role in how your manage your condition and how you experience living with Turners Syndrome.

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