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Ngubane, Neliswa

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CONGENITAL ABNORMALITIES DIAGNOSED ANTENATALLY AT CHBAH: A RETROSPECTIVE DESCRIPTIVE STUDY 2016-2018

Neliswa Ngubane (MBCHB, Natal)
Jayshree Jeebodh (FCOG, MBCHB)

ABSTRACT

Background

Information on trends and prevalence of congenital abnormalities (CAs) in South Africa remains limited despite adverse associated perinatal and lifelong outcomes. Multiple factors have been shown to contribute to development of CAs. An understanding of our at-risk population will assist us in appropriately and timeously managing our patients.

Objectives

To describe the patterns and trends of CA diagnosed antenatally in our setting, in terms of: chromosomal and structural abnormalities; maternal age; gestational age (GA) at diagnosis; potential management of the diagnosed CA; and frequency of termination of pregnancy (TOP) after diagnosis.

Methods

This was a retrospective study conducted over a 14 month period on pregnant women with a diagnosed CA. Data were obtained from the fetal medicine unit's record books and electronic database. SPSS software was used for data analysis.

Setting

The study was done at Chris Hani Baragwanath Academic Hospital, a tertiary level hospital in Johannesburg, South Africa. It is the third-largest hospital in the world. Participants were

attendees at the hospital's Fetal Medicine Unit (FMU), led by a team of three fetal medicine specialists.

Results

Four hundred and fifty-one records were extracted, with 404 participants enrolled into the study. The mean GA at diagnosis of a CA was 27 weeks and 4 days; 21/404 (5.1%) were confirmed chromosomal abnormalities. Central nervous system (CNS) was the leading system in structural CAs, followed by musculoskeletal and soft tissue (MSS), renal and cardiac systems. Trisomies 21 and 18 had similar occurrences. After sonographic diagnosis of a CA, 94/404 women (23.3%) proceeded to invasive testing; 79 (19.6%) women opted for TOP after CA diagnosis, and 32.9% of these were fetocides. Teenage mothers had no diagnosis of a chromosomal CA during the study but had a 30.8% occurrence of gastrointestinal birth defects. A majority (52.4%) of chromosomal abnormalities were diagnosed in the advanced maternal age group; 71% of the chromosomal abnormalities had an MSS defect detected on ultrasound scanning.

Conclusion

This study has shown that we are diagnosing CAs in advanced pregnancies as a result of delayed referral and presentation, with likely consequent psychosocial and socioeconomic difficulties for the families involved. The overwhelmed system with an understaffed FMU is also of concern. Attempts should be made to reduce this delay, to prevent not only family distress, but also to decrease the rate of fetocide, which poses an ethical dilemma to the fetal medicine specialists. Our findings on CAs are similar to those in the developed world, and we must strive to have our patients managed with similar care. Fetal surgery capability is still limited in South Africa, and greater availability of such skills can improve outcomes in affected patients.