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Prenatal detection of congenital abnormalities

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Objective

To describe the patterns and trends of congenital abnormalities (CA), chromosomal and structural, diagnosed in the antenatal period in CHBAH fetal medicine unit. Maternal age, gestational age (GA) at diagnosis, correctability of the diagnosed structural abnormality and option to terminate the pregnancy post diagnosis were factors evaluated.

Methods

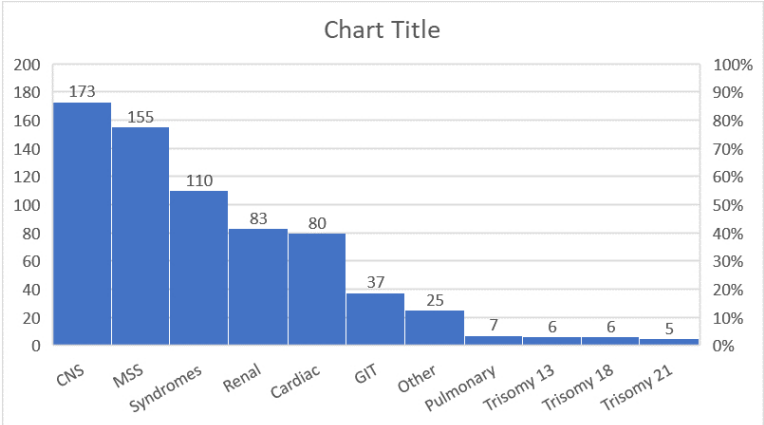
Retrospective study of all women with a fetal CA on ultrasound and/or chromosomal abnormality during the period January 2016 to March 2018 in the fetal medicine unit. Data was obtained from the unit's database and record books, which consisted of patient's history, findings and management options. SPSS was used for data analysis, with respect to maternal age, abnormality diagnosed and GA at diagnosis, whether invasive testing/procedure done or not as well as decision to continue or terminate the pregnancy.

Results

During the study, 404 patients were included. 21/404 (5.1%) were confirmed chromosomal abnormalities on invasive testing. CNS was the leading CA under the structural abnormalities, followed by musculoskeletal (MSS) with renal and cardiac systems paralleling each other. Trisomy 21 (6/21) and Trisomy 18 (6/21) had the same frequencies during the study. Post diagnosis sonographically of a CA, 94/404 (23.3%) of patients proceeded to invasive testing. The mean GA at diagnosis was 27w4d. 79 (19.6%) patients opted to have a TOP after CA was diagnosed, with 26 (32.9%) being fetocides. From the structural abnormalities, 180 (44.6%) were classified as correctable. Leading CA in teenage pregnancy was CNS (50%) followed by GIT (30.8%), with no chromosomal abnormalities diagnosed during study period. MSS birth defects were the leading CA in AMA pregnancies, followed by CNS. 52.4% of the chromosomal abnormalities were diagnosed in the AMA group. 71% of the chromosomal abnormalities had an MSS defect detected on ultrasound.

Conclusion

CNS and MSS are the leading birth defects (BD) in our setting, with cardiac and renal defects having the same frequencies overall. Multiple congenital abnormalities ranked 3rd in the study. There is a higher rate of GIT defects in teenage pregnancies compared to the general population overall. The mean GA at diagnosis is high, resulting mostly from late antenal booking by patients but also the congested system contributing to a delay in the time level 3 scans are done. More than 50% of chromosomal abnormalities were associated with AMA. A functional national as well as provincial surveillance system for BD would be a beneficial tool moving forward to assist with future plans to lower the mean GA at diagnosis and the rising burden of BD in AMA as reproductive years are also increasing.



Trends and frequencies of diagnosed congenital abnormalities. CNS leading birth defect, followed by MSS, noted is similar occurrence between renal and the cardiac systems. Pulmonary abnormalities are the least diagnosed. The 3 trisomies had a similar incidence rate during the study.

Table. Recorded successful procedures during study period. Amniocentesis is the commonest diagnostic procedure with CVS done the least, owing to the mean GA at which CA are diagnosed sonographically in our setting.

INVASIVE PROCEDURES PERFORMED	no	%
Amniocentesis	101	63.5
Cordocentesis	29	18.2
Fetocides	26	16.3
CVS	2	1.2
Pericardiocentesis	1	0.62