# Pattern of Congenital Anomalies in Aborted Fetuses Reporting to Tertiary Care Centre in Rishikesh: A Pilot Study

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**Abstract**

*Background:* Congenital anomalies are a major cause of fetal loss, neonatal morbidity, and mortality worldwide. Prenatal ultrasonography, particularly the level-II anomaly scan, enables early detection of many structural abnormalities; however, it may not always identify the underlying genetic etiology. Molecular testing of fetal tissue and products of conception has significantly improved the detection of chromosomal abnormalities and genetic defects associated with congenital malformations. Combining molecular diagnostics with fetal autopsy can enhance diagnostic accuracy and help determine recurrence risk in future pregnancies.

*Aim:* To evaluate congenital abnormalities in aborted fetuses using molecular genetic testing and fetal autopsy at a tertiary care hospital.

*Materials nd Methods:* This descriptive cross-sectional observational study will include aborted fetuses with gestational age ≥12 weeks received during the study period, following informed parental consent. Maternal demographic and obstetric data, including maternal age, consanguinity, infections, systemic illnesses, teratogenic exposures, folic acid intake, and previous reproductive history, will be recorded. Prenatal ultrasonography findings will be correlated with fetal examination. Each fetus will undergo detailed external examination, morphometric measurements, and systematic fetal autopsy with photographic documentation of anomalies. Fetal tissue or products of conception will be subjected to molecular genetic testing, including karyotyping, chromosomal microarray analysis, or targeted molecular testing where indicated.

*Expected Outcome:* The study will determine the spectrum of congenital anomalies and the proportion associated with genetic abnormalities. Correlation of molecular findings with fetal autopsy and prenatal imaging will improve diagnostic accuracy and facilitate appropriate genetic counselling for future pregnancies.

**Index Terms**

Congenital Anomalies; Molecular Genetic Testing; Fetal Autopsy; Chromosomal Abnormalities; Products of Conception; Prenatal Ultrasonography