Challenges and Potentials in Prenatal Diagnosis of Genetic Disorders
Chinmayee R, Goundan A
Navodaya Hospital, Hyderabad, India

Objective
To study the incidence of genetic disorders in our population and analyse the challenges and potentials in prenatal diagnosis.

Methods
All cases that presented in Fetal Medicine clinic between Feb- Dec 2016 for possible prenatal diagnosis of genetic disorders were studied for the type of genetic abnormality, primary condition confirmation by molecular diagnosis and whether appropriate prenatal testing was possible. The cases were followed through pregnancy and till delivery.

Results
Out of a total of 3183 patients seen, 52 cases(1.63%) had history of confirmed or features strongly suggestive of genetic abnormalities. In 2 cases (3.08%), carrier screening of parents was done and then prenatal diagnosis was possible. Only 12 cases (23.08%) had a proven primary molecular diagnosis. In the other cases interim surveillance with ultrasound scans or empirical approach was carried out based on the known features in previous cases. In 2 cases (3.08%) there was a repeat of the previous problem and no diagnosis could be established due to limitations of testing. Of cases who did not have a primary problem, 1 case of a rare genetic syndrome was incidentally diagnosed postnatally.

Conclusion
In suspected genetic disorders, a primary molecular diagnosis must be obtained ideally pre conceptionally so as to plan effective prenatal diagnosis Establishing primary molecular diagnosis in the present pregnancy is a formidable challenge and often clinically not feasible.