

## Prenatal diagnosis of common fetal aneuploidies: Scenario in India

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In the present issue of the Journal Fauzdar *et al.*<sup>[1]</sup> described detection of fetal aneuploidies in 163 pregnancies using a combination of techniques. Different samples like amniotic fluid, chorionic villus, and fetal blood obtained through amniocentesis were used for detection of these aneuploidies. Investigators used classical Giemsa banded cytogenetic technique supplemented with fluorescent *in situ* hybridization (FISH) for chromosomes 13,14,15,16,18,21,22, and X for their aneuploidies. In several cases, where FISH study was normal, classical cytogenetics showed inversion of chromosome 9 as it is not associated with clinical abnormality and considered as polymorphism. It is not clear how these patients were counseled. Moreover, authors used fluorescence microscope for Giemsa Banding studies which points to lack of knowledge of the group for prenatal diagnosis. If we carefully examine Table 2 of the result where 15/163 (~9%) cases showed abnormal results, aneuploidies of only chromosome 21(4 Cases) and 1 patient with aneuploidy with chromosome 13 were seen. Two patients had monosomy X. Though authors could not correlate these findings with part delivery samples, in 10/163 cases (9.6%) classical cytogenetics failed but no aneuploidies were also found

in any of these cases using FISH based studies. Four cases were suspected of mosaicism and after counseling three opted for abortion; however, abortions were not checked for dysmorphology and cytogenetics. In the 4th case, the mother delivered the baby, but we do not know whether the baby was born with birth defect or have abnormal cytogenetics. These are serious flaws in the study. Authors agreed that the failure of classical cytogenetics present in 10% cases were too high; hence, their techniques need to be improved. Authors looked for aneuploidies, but Down's syndrome due to translocation is not uncommon in India. Surprisingly, in the present series new translocation Down case was detected using classical cytogenetics. Authors have quoted several papers from India<sup>[2-4]</sup> describing fetal chromosomal defect. However, several studies in this area from southern and western part of India, particularly on prenatal diagnosis of Down's syndrome have been overlooked. Translocation Down in India comprises 10-15% of Down's syndrome patients. The authors have used 8 FISH probes but agreed aneuploidic of 13,18,21,X, and Y contribute 95% of chromosomal aberrations; hence, the use of eight probes might have increased the cost of the test unnecessarily for the patient. In our country, cost is always an important component of diagnostic algorithm to that effect Halder *et al.*<sup>[5]</sup> in the same issue of the Journal have published their short technical report on PRINS (Primed in situ Labeling/Synthesis) technique for detecting aneuploidies of chromosomes X,Y, 13,18, and 21. It may be possible to use the technique more often in many laboratories in this country. Increasingly, non-invasive prenatal

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diagnosis using DNA from mother's blood for using fetal aneuploidies are being increasingly used on high throughput platform.<sup>[6]</sup> Present report presages a new era in the scenario of prenatal diagnosis of aneuploidies in India. We can only hope that more user friendly fast and cheaper techniques will finally be evolved.

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