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**Abstract Topic:-** Genetic counselling

**Abstract Title:-** Cytogenetic analysis of 714 cases of Down syndrome and the importance of genetic counseling.

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**Aims:-**To analyze the different variations found in Down syndrome cases confirmed by conventional karyotyping.

**Methods:-** Heparinized peripheral blood of suspected cases of Down syndrome Pan-India was received for karyotyping. A 72 hours phytohemagglutinin M (PHA-M) stimulated culture was set up and harvested using conventional karyotyping protocol. The slides were G-banded and analyzed using Zeiss photomicroscopes and Metasystems Ikaros software

**Results:-** Trisomy 21 was found in 714 cases over the past 8 years. Of these 401 (56.16%) were males and 309 (43.28%) were females, 2 (0.28%) had a disorder of sex development (DSD) and 2 (0.28%) had sex chromosome anomalies. Free trisomy was observed in 651 (91.17%) cases. Robertsonian translocation trisomy 21 was seen in 58 (8.12%) cases while 5 (0.7%) cases had trisomy 21 with an extra chromosome. Among the Robertsonian translocation cases, t(14;21) was seen in 30 and t(21;21) in 22 cases. In 5 other cases, the karyotypes were 48,XXY,+21, 48,XXX,+21, 48,XY,+21,+mar, 47,XY,+21,add(2)(p21) and 48,XX,+14,+21. Mosaic trisomy 21 ranging from 50-80% was observed in 5 cases.

**Conclusions:-** Our observational study showed that males with trisomy 21 (56.16%) were more frequent than females (43.28%). Also, 58 (8.12%) cases had a Robertsonian translocation which may have been inherited from one of the parents. In such cases, parental karyotyping is very important to check for carriers, as they have a high risk of trisomy 21 in future pregnancies, or early miscarriage due to aneuploidy. Carriers of t(21;21) need to be counseled that all their future children will have Down syndrome, so they could consider a donor gamete or adoption. In carriers of other translocations, prenatal diagnosis is mandatory. Awareness of preimplantation genetic testing for aneuploidy (PGT-A) to select euploid embryos could also be spread where affordable. 5 other cases showed presence of additional chromosomes with trisomy 21. Their parents will benefit with the use of PGT-A for a future pregnancy. Thus, proper parental genetic counseling and conventional karyotyping of parents and sibs of translocation cases is important to check for carriers in the extended family. Awareness of the importance of early prenatal diagnosis of trisomy 21 in rural India needs to be spread.

**Keywords:-** Down syndrome, Trisomy 21, Robertsonian translocation, PGT, free trisomy 21