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Cytogenetic Analysis of 1400 Referral Cases: Manipal Experience

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ABSTRACT Karyotypes were examined in 1400 cases, suspected of having chromosomal abnormalities. A total of 343 (24.5 per cent) had abnormal karyotypes including 43 (3.07%) polymorphic variants; 14.28% of children exhibited chromosome abnormalities including 12.07% of Down syndrome, 2.21% of congenital anomalies including global developmental delay, 0.5% with intersex disorders. Chromosomal abnormalities were observed in individuals with pubertal failures including short stature and amenorrhea in females (3.35%), and were recorded in 0.43% males. Cases of reproductive failures (3.64%) included recurrent miscarriages, bad obstetric history and infertility. Of these 2.28% were instances of polymorphic variants. Fifty seven patients who diagnosed of various symptoms of cancer were studied and found to have 56% structural variations including Philadelphia chromosome. Cytogenetic analysis is found to be useful in providing genetic counseling.