Turner's Syndrome: A Case Report

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Turner's syndrome is the most common sex chromosomal abnormality in females. This occurs due to complete/ partial absence of the second X chromosome or presence of a functionally defective second X chromosome in XX genotype due to fragmentation, deletion, ring formation, isochromosome or expression problems. In placental mammals, one functional X chromosome is maintained and any additional X chromosome is heterochromatinised randomly (Barr Body). Therefore, conventional XO genotype in Turner's will be reflected as absent Barr Bodies. The two features that best characterise this condition are gonadal dysgenesis and short stature. However, there may be features like neck webbing and cubitaus valgus deformity which are not constantly seen in all diagnosed Turner's cases even though similar chromosomal defect is detected. Here, we present two sisters with proportionate short stature who were referred for sex chromatin analysis.

The two sisters were 6 and 4 years of age and both of them were well below the 3rd centile for age and sex specific height. The younger sister had neck webbing and cubitus valgus deformity where as the elder sister did not have any dysmorphic feature. Younger sister also showed some features of nutritional deficiencies and markedly delayed skeletal age with a global development delay. Ultrasonically, their uteri were normal but ovaries were poorly visualised. Barr body analysis revealed abnormally large heterochromatinised X chromosomes with a high count in both sisters.

A diagnosis of Turner's syndrome may be suspected when there are a number of typical physical features observed. The Noonan's syndrome too could be considered in the differential diagnosis as it has a similar phenotype. In the present study, sex chromosomal monosomy and mosaicism can be excluded with the findings in Barr body analysis and an abnormally large sex chromatin may suggest an isochromosome. The delay in the bone age too possesses a diagnostic dilemma in the younger child. Short stature in children may occur due to different reasons. However, it is important to do sex chromatin studies even though dysmorphic features are not apparent. Similarly in a Turner's syndrome patient, it is important to investigate for other contributory causes for the clinical findings which may be treatable.