TWO RARE VARIANTS OF TURNER SYNDROME WITH ISOCHROMOSOME STRUCTURAL ABNORMALITIES

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Background: Turner'ssyndrome (TS) is the most common cause of short stature and delayed puberty of female sex. Approximately half of the patients have its classic form of 45 XO, one fourth of patients are different mosaic forms and the remaining cases are structural abnormalities on X chromosome, among them most common structural abnormality is isochromosomeXq. These variant Turner's can present with delayed menarche, amenorrhoea and infertility rather than classic manifestations of TS.Here we describetwo uncommon variants of TS, one is structural abnormality on X chromosome as 46X, iso(Xq) and another one is mosaic variety of TS including Isochromosome X as form of 45XO/46X, iso(Xq).Both of them presented with short stature and secondary amenorrhea without classic manifestations of TS. In TS with or without mosaicism, the frequency of isochromosome is reportedto be about 15-18%. Due to lack of classical manifestations of TS, diagnosis may be delayed and/or missed. So, female of short stature with secondary amenorrhoea should be searched for rare variants of TS by chromosomal analysis.

Key Words: Turner syndrome, Isochromosome X, Amenorrhoea, short stature

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