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46,X,DER(X)T(X;8)(P11.2;P11.4)MAT IN A TURNER SYNDROME PATIENT

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Turner syndrome is a sex chromosome disorder occurring in 1 in 2500 female births and is characterized by retarded growth and gonadal failure. While the vast majority of cases are related to a full X chromosome loss, some are consequence of structural rearrangements leading to functional inactivation of parts of the chromosome.
We report a case of an 11-year-old girl with phenotypic expression of Turner syndrome, namely short stature, hypogonadism hypergonadotropic and typical dysmorphic features. Cytogenetic analysis revealed an unbalanced translocation between the X and 8 chromosomes: 46,X,der(X),t(X;8)(p11.2;p11.4)mat, thus indicating a monosomy of Xp and an 8p trisomy. Correlation between the cytogenetic findings and the phenotype are discussed