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## MOSAICISM WITH TWO X CHROMOSOME DIFFERENT REARRANGEMENTS AND A TURNER-*LIKE* PHENOTYPE: CASE REPORT

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The frequency of Turner Syndrome (TS) has been reported as 1/5,000 live female births. This pathology is most commonly associated with a 45,X karyotype but in approximately 25% of the patients the karyotype shows both a normal X and a structurally abnormal X chromosome. These abnormalities, which include deletions, duplications, inversions, translocations and ring chromosomes, imply chromosomal breaks and significant imbalance of gene content; they are generally benign because of the preferential inactivation of the abnormal X. Six to 15% of patients with TS are mosaics for an X ring chromosome [r(X)] line; however, in these cases the incidence of mental disability and other congenital abnormalities may be significantly higher. Some authors report that severe r(X) phenotypes can be seen in patients with active r(X) chromosomes lacking the X-inactive specific transcript gene (XIST gene). The authors present a female patient aged 3 with clinical features of Turner syndrome. Cytogenetic studies revealed a novel mosaicism with two different abnormal cell lines: 1- the major line with a normal X chromosome and another X with a rearrangement corresponding to a deletion of the distal region of the short arm (Xp) and duplication of the long arm (Xq13->qter); 2- the other with a normal X chromosome and a r(X)(p22.3q13). FISH studies confirmed: in the line containing a rearranged X chromosome a deletion of the Xp subtelomeric region (Xp22.3) and a duplication of the Xq subtelomeric region (Xq28); in the line with r(X) a deletion of both subtelomeric regions. The presence of the XIST gene was demonstrated both in normal and abnormal X chromosomes, in the two cell lines. The authors will present a complete cytogenetic characterization of the patient and discuss all the factors that play an important role in determining the phenotypic outcome.

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## DISTAL XQ27->Q28 DUPLICATION AND FUNCTIONAL DISOMY: CLINICAL AND CYTOGENETIC CHARACTERIZATION

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Distal Xq duplications are intrachromosomal disorders that constitute the main cause of functional disomy in males. Most cases are inherited from heterozygote mothers. These duplications vary in size, location and gene content of the correspondent segment. Hemizygous male descendents have a functional partial Xq disomy and are phenotipically abnormal. A male proband aged 7 months was referred for cytogenetic studies due to psychomotor delay, coarse features and cardiopathy. Both the child and the mother's karyotypes were obtained from peripheral blood lymphocyte cultures using standard techniques and chromosomes were analysed with GTG banding. The child's mother was studied using fluorescence in situ hybridization (FISH) with a whole chromosome painting probe for the X chromosome (wcpX, Cytocell), to exclude the involvement of any other chromosome and X inactivation pattern techniques. Karyotypes were requested for the mother's parents. The cytogenetic analysis revealed extra material on the long arm of the X chromosome both in the proband and in the mother. Maternal grandparents had normal karyotypes. X inactivation studies in the mother showed that the abnormal X was always late replicating and therefore inactive. No further testing was possible in the child, since he deceased of pneumonia at the age of 8 months. The extra material observed in the distal segment of the long arm of the X chromosome in this family was interpreted as a duplication of chromosome X terminal region (q27.3->q28). Child's final karyotype: 46,Y,dup(X) (q27.3-q28)mat.Large cytogenetic visible duplications of Xq are rare, the most common being the Xq27->q28 region and there are only about 40 cases described in the literature. The prevalence of Xq duplications is still unknown but the clinical outcome is a well recognized phenotype. The proness to infections in individuals with this condition is almost invariably the cause of death in childhood. The clinical history and cytogenetic findings of this case are in agreement with similar cases previously reported. Parents were given appropriate genetic counselling and offered the possibility of prenatal genetic diagnosis in future pregnancies. Since then, two healthy 46,XX daughters were born.