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Abstract Topic: - Clinical Genetics

Abstract Title: - The varied hues of X:autosomal translocations

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Aims: - To describe the clinical presentations and cytogenetic characteristics of 8 cases of X: autosomal translocation

Methods: - Retrospective data from January 2001 to December 2022 was reviewed to compile the cases. G-banded karyotyping had been performed on short term cultures of phytohemagglutinin stimulated peripheral blood.

Results: - Eight cases of X: autosomal translocation were identified during this period. Seven patients were female and one was male. Partner chromosomes differed. Chromosomes 4,6,7,9,13,14,16, and 17 were involved. In 5 cases, the breakpoint was on the long (q) arm of X extending from q13 to q23. In the remaining 3, the breakpoint was on the short (p) arm of X extending from p11.2 to p21.

Three of the female patients presented as children with global developmental delay. One of them was diagnosed with Menke's disease, an X-linked recessive disorder. The breakpoint in this patient was Xq21, which corresponds to the location of ATP7A, the causative gene of Menke's disease.

The adult females were investigated for primary amenorrhoea and premature ovarian failure but did not show Turner stigmata. The male patient presented with infertility and azoospermia.

Parental karyotype was available for only one child, which was normal in both parents.

Conclusions: - Our study demonstrates the clinical spectrum of balanced X: autosomal translocations, that may have a phenotypic impact in contrast to most other balanced cytogenetic translocations that affect two or more autosomes. Three of the eight ascertained patients showed developmental delay, including one female who manifested an X linked recessive disorder, pointing to skewed lyonization. The adult patients presented with reproductive consequences. It is pertinent to note that balanced translocations will not be detected on most molecular genomic assays which are replacing karyotype. We concede that our findings are likely to be influenced by ascertainment bias, which might explain the skewed male: female ratio, as males carrying the abnormality are usually phenotypically normal, though nearly always infertile.

Keywords: - Eight cases of X: autosomal translocation were identified during this period. Seven patients were female and one was male. Partner chromosomes differed. Chromosomes 4,6,7,9,13,14,16, and 17 were involved. In 5 cases, the breakpoint was on the long (q) arm of X

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