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Birth of a healthy child by a woman with inherited Xq duplications who had experienced stillbirths

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ABSTRACT. A 23-year-old woman who had experienced repeated stillbirths, was found to carry an additional segment on the long arm of the X chromosome. Array comparative genomic hybridization (aCGH) confirmed the origin of the 2 duplications (about 17.11 Mb). Thus, her karyotype was 46, X, dup (X) (q13.2-q21.1), dup(X) (q21.32-q22.1). We demonstrate that aCGH is a useful complementary tool to cytogenetic analysis for accurately determining banding. To our knowledge, this is the first case with normal apparently phenotype who inherited 2 duplications on Xq. Notably, after 2 stillbirths, she bore a healthy, normal female infant via natural pregnancy. Thus, a carrier of this karyotype can birth a phenotypically normal child.

Key words: Array comparative genomic hybridization; Duplication X; Inherited; Stillbirth

