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FISH, PCR and cytogenetic characterization in a girl with ambiguous genitalia and karyotype mos46,X,iso(Y)(qter→p11.3::p11.3→qter)[80]/45,X[17]/46,X,+mar[3]

S.R.F. Pereira[1], A.C.N. Pereira[2], M.T.V.L. Souza[1] and M.R.B.P. Ramos[2]

[1]Departamento de Biologia, Laboratório de Genética e Biologia Molecular, Universidade Federal do Maranhão, São Luís, MA, Brasil
[2]Hospital Materno Infantil, Universidade Federal do Maranhão, São Luís, MA, Brasil

Corresponding author: S.R.F. Pereira
E-mail: silmaregina@yahoo.com.br

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ABSTRACT. A cytogenetic study was carried out in a girl with virilized external genitalia, who showed a karyotype containing a Y isochromosome in mosaic form: mos46,X,iso(Y)(qter→p11.3::p11.3→qter)[80]/45,X[17]/46,X,+mar[3]. The chromosome aberrations were confirmed by fluorescence *in situ* hybridization analysis, with both whole chromosome paint Y probe and centromeric X chromosome probe. The molecular analyses by PCR detected the presence of the SRY, DAZ and AMGY genes, confirming the presence of the whole long arm and almost whole short arm of the Y chromosome. We suggest that the structural alteration of the Y chromosome was a new mutation, which occurred in the initial mitotic divisions of the embryo, originally 46,XY. The breakpoints occurred on the distal extremity of the short arm with later fusion of its extremities producing a Y isochromosome. The later numerical alteration occurred as a consequence of chromosomal instability. Although almost all cells (80%)

in peripheral blood belonged to the iso(Y) line with a duplicated SRY gene, this did not determine male sexual differentiation in the patient. The result of accurate evaluation provides correct sex assignment and the prevention of the neoplastic degeneration of a dysgenetic gonad.

Key words: Ambiguous genitalia; Y isochromosome; Mosaicism; SRY gene

