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Cytogenetic and molecular studies of patients with disorders of sex development

Mona Kamal Mekkawy
National Research Center, Cairo, Egypt

Abstract

Disorders of sex development (DSD) are defined as congenital conditions in which development of chromosomal, gonadal or anatomical sex is atypical. The incidence of ambiguous genitalia in Egypt is one per 5000 live births.

Sex determination depends on the sex-chromosome complement of the embryo and is established by multiple molecular events that direct the development of germ cells, their migration to the urogenital ridge, and the formation of either a testis, or an ovary. The type of gonad determines the hormonal environment, which directs the internal duct differentiation and the formation of the external genitalia.

This study is conducted through a project funded by NRC, aimed at long term evaluation of the cytogenetic spectrum and molecular abnormalities associated with DSD among Egyptian patients. The study included 232 patients presented to the clinical genetics department over a period of 2½ years (2010-2013).

The patients showed a wide spectrum of presenting features associated with different arrays of chromosomal abnormalities.
Variable numerical and structural sex chromosomal abnormalities were found in 125 patients. Autosomal abnormalities were detected in 6 patients in association with congenital anomalies. 46, XX karyotype was found in 4 male patients with testicular DSD. Molecular studies were carried out for 45 patients with 46, XY DSD and revealed different mutations in *HSD17B3, SF1*, *SRD5A2* and *AR* genes, including two novel mutations.

The comprehensive study of DSD patients with careful genotype- phenotype correlation can provide a great help in better understanding of the genetic basis, which will have a strong influence on management strategy.

Biography

Mona Mekkawy has completed her Ph.D in Human Genetics at the age of 44 years from The National Research Institute, Alexandria University, Egypt, January, 2010.

Experienced in the field of cytogenetics and molecular techniques (1995- 2013).

Participated as a speaker in many International genetics conferences and in cytogenetic training courses and workshops.

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