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Genetic Study of Abnormal Sexual Differentiation in Alexandria, Egypt

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In Egypt, abnormal sexual differentiation (ASD) constitutes a considerable sector among cases with genetic disorders. The study was conducted with the aim of investigating the different types of ASD and identifying the role of heredity in the development of familial sex disorders. The study included 133 patients.
Disorders of gonadal differentiation were found in 69 patients (51.9%). True hermaphroditism was diagnosed in 6 patients (4.5%). Female pseudohermaphroditism was present in 32 patients (24%). Male pseudohermaphroditism was diagnosed in 26 patients (19.5%); 13 had complete androgen insensitivity, 7 had partial androgen insensitivity, 5 had 5(X reductase deficiency and one patient had agonadism. Overall; 42 (31.6%) patients had chromosomal abnormalities and 85 (63.9%) patients had monogenic disorders.
The frequency of consanguinity was high (51.9%). The average inbreeding coefficient was higher (0.030.06) than that reported for the Egyptian population in general (0.01). In conclusion, the study shows that monogenic disorders have an important role in the aetiology of ASD in Egypt. Therefore screening program may be beneficial for early detection and management.