Noha H Issa et al., J Clin Case Rep 2016, 6:2 (Suppl)
http://dx.doi.org/10.4172/2165-7920.C1.003

International Meeting on
Clinical Case Reports
April 18-20, 2016 Dubai, UAE

Prenatal and postnatal findings in new cases of Fraser syndrome in Egypt

Noha H Issa, Ebtesam M Abdalla, Louay H Zayed and **Asmaa K Amin**
Alexandria University, Egypt

Fraser syndrome (FS) is a rare autosomal recessive disorder characterized by cryptophthalmos, cutaneous syndactyly, laryngeal malformations and urogenital defects. It may be also associated with ear, nose and skeletal abnormalities. Almost half of affected infants are stillborn or die in infancy and mental retardation is common. There is marked interfamilial clinical variability. However, there is strong phenotypic similarity and concordance of the degree of severity of the disease within a family. The pathogenesis evidently involves abnormal epithelial integrity during prenatal life. FS is a genetically heterogeneous condition. Thus far, mutations in FRAS1, FREM2 and GRIP1 have been identified as cause of FS. The three genes encode components of a protein complex that are essential for the adhesion between epidermal basement membrane and the underlying dermal connective tissues during embryonic development. Here, we report on 10 new cases of Fraser syndrome from six Egyptian families with inconsistent compatibility with life and variable expression in prenatal sonographic findings and postnatal clinical manifestations.

Biography

Noha H Issa is a Lecturer of Human Genetics at the Medical Research Institute, Alexandria University, Egypt. She has received her MBBCh from Alexandria Faculty of Medicine and her MSc and PhD in Human Genetics from Medical Research Institute, Alexandria University. Her main scope of work is the diagnosis of genetic diseases, performing genetic tests and provides genetic counseling to affected families.

issanoha@yahoo.com

Notes: