Emanuel Syndrome

Case of the month series 0808

Case

A 4 year old male child, pediatric referral to GeneTech showed dysmorphology and delayed development. The child is a product of non-consanguineous marriage. Clinical presentation and evaluation of medical reports showed prominent forehead, down slanting palpebral fissures, flat nasal bridge, microretroganthia, cleft-palate, club foot, renal hypoplasia, cryptorchidism, micropenis, diaphragmatic hernia, hypotonia, delayed development and poor weight gain. Pedigree analysis showed incidence of two neonatal deaths to maternal grandmother.

Genetic databases and possible syndromes:

Emanuel Syndrome + derivative 22 with t(11;22)
Fryns Syndrome autosomal recessive inheritance, unknown gene
Smith-Lemli Opitz autosomal recessive inheritance, *DHCR7* gene
Pallister Killan Syndrome iso 12p chromosome
Kabuki Syndrome sub microscopic deletion of 8p22-23
Wolf-Hirschhorn Syndrome microscopic deletion of 4p16 band

Features in Emanuel Syndrome

Diagnosis

Chromosomal analysis of the child recommended by genetic counselor revealed a supernumerary 22 chromosome caused due to a translocation between chromosome 11 and 22. The karyotype is designated as **47,XY,+der(22)t(11;22)(q23;q11)** confirming Emanuel syndrome and ruling out possibility of other disorders.

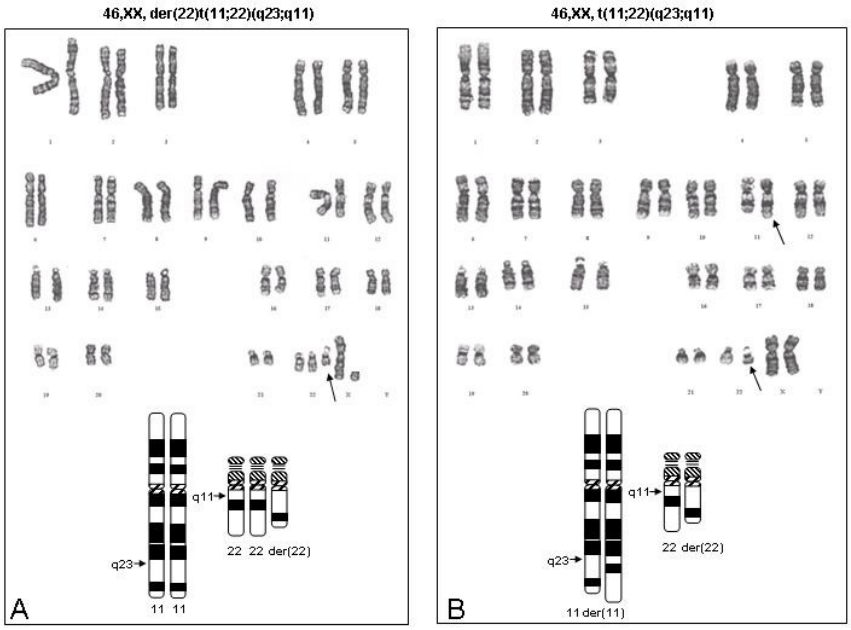
Emanuel syndrome

It is a rare disorder and is characterized by severe mental retardation, microcephaly, failure to thrive, ear anomalies, micrognathia, kidney abnormalities, cardiac and genital abnormalities. This clinical phenotype arises from duplication of 22q10-22q11 and duplication of 11q23-qter on the supernumerary der(22). Depending on the age and extent of systematic involvement of the individual with ES, evaluations and care involving healthcare providers from multiple specialties are necessary.

Recurrence risk and Genetic Counseling

Parental karyotyping was subsequently performed and mother was diagnosed with a balanced translocation of 11;22 All carriers of balanced translocations are unaffected and carry recurrence risk for Emanuel syndrome or a spontaneous abortion due to gametic chromosomal rearrangements. A conceptus with the same balanced translocation as mother will have a normal phenotype.

Availability of prenatal diagnostic options was discussed with family in the counseling session.



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