GENETICS AND GENODERMATOSES

MOTHER AND SON WITH TURNER'S AND NOONAN'S SYNDROME

Jaskanwal Kaur [1] - Sangita Jain [2]

Sdh Derabassi, Punjab Health Department, Mohali, India [1] - Sdh Dera Bassi, Punjab Health Department, Mohali, India [2]

Background: Noonan's syndrome is an autosomal dominant condition which phenotypically resembles Turner's syndrome. Children have intellectual impairment and communication difficulties. Dermatological manifestations are leukokeratosis of lips and gingivae, coarse hair and low posterior hairline. Ulerythema oophyryogenes may be cutaneous marker of NS. Turner's syndrome occurs due to defect of one of the X chromosomes with genotype 45 XO. Some case have 46 chromosomes with partial deletion of involved X chromosome. The patient has short stature, pterygium colli, shield shaped chest and high arched palate. Cutaneous features include redundant skin fold around the neck, low posterior hairline and hypoplastic nails. The intelligence is usually normal.

Observation: An eight year old male child presented in skin OPD with skin manifestations like pigmented lesions on the legs, low posterior hairline, webbed neck, short stature along with defect in vision and speech associated with learning defects and associated hyperactivity in the child. There was also visible pectus excavatum. The skin lesions were those of keratosis pilaris atrophicans. X ray chest showed no anomaly. Chromosomal analysis revealed translocation involving short arm of one of the chromosome 2 at region p23 and long arm of chromosome 12 at region q24.1 respectively. The mother of child also had short stature, webbed neck, broad chest . The female had hearing disability and subnormal intelligence. Skin changes consisted of Low hairline, Laxity of skin on the back and neck.

Key message: Fertility without treatment is rare in a patient with Turner's syndrome. In this case mother with Turner's syndrome bore a child with noonan's syndrome. Early diagnosis and intervention can improve the quality of life in such patients like treatment of cardiac anomalies, hearing prosthesis and ophthalmic treatment can improve vision in patients.