GENETICS AND GENODERMATOSES

MISSING ISLAND OF HAIR AND PAWS – A RARE SYNDROME

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Background: Adams-Oliver syndrome is a rare, variably expressed, multisystem congenital disorder characterized by aplasia cutis congenita of scalp and transverse limb reduction defects.20 % cases had congenital heart defects at birth which include ASD, VSD, tetralogy of fallot and left sided obstructive lesions. Under frieden's classification , it is classified as type 2 aplasia cutis congenita. Only 130 cases have been reported throughout the world till now.

Observation: A 5-year-old female born out of third-degree consanguineous marriage presented with an atrophic bald patch on the vertex of the scalp with the absence of toes since birth. The patient's mother reported that at birth there was complete absence of skin over the vertex of the scalp that later healed by scar. Mother denied any h/o drug intake, infection or radiation during pregnancy. Echocardiography done at the time of birth revealed 6mm ostium secundum type of ASD with a left to right shunt and dilated right atrium and ventricle. No signs & symptoms were suggestive of neurological deficit. No family h/o Adams-Oliver syndrome, mental retardation or CNS abnormalities. X-ray of the feet revealed absent phalanges. Echocardiography, chest x-ray, ECG, abdominopelvic ultrasonography, echocardiography, CT brain were all normal.

Key message: Presented this case due to its rarity in literature