A 10 months old boy had bilateral web over popliteal region since birth leading to flexed knees. He also had cleft lip and palate, inguinal hernia.

What is the diagnosis?

Popliteal pterygium syndrome. It is an autosomal dominant disorder due to mutation of the IRF6 gene affecting the face, limbs and genitalia. (1,2) It is also known as popliteal web syndrome and facio-genito-popliteal syndrome. Males and females are equally affected.

The characteristic feature of this syndrome is a web extending from the heel to the ischial tuberosity which contains a palpable cord of connective tissue and occasionally the popliteal artery and peroneal nerve.

Facial defects include cleft palate with or without cleft lip and fibrous band in mouth. Limb findings include web behind the knee, webbing of toes and toenail malformation while genital defects occur in the form of hypoplasia of labia majora, scrotum or cryptorchidism. These children do not have growth or intelligence disturbance. (1,3) Prenatal sonography might detect this syndrome by detecting a cleft lip or palate along with inability of the fetus to stretch the knee. The overall prognosis is good.

References


A six months female child came with swelling over bridge of nose since birth. The swelling had increased slightly since then. On examination, it was soft and cystic.

SWELLING AT BRIDGE OF NOSE
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A six months female child came with swelling over bridge of nose since birth. The swelling had increased slightly since then. On examination, it was soft and cystic.
What is the diagnosis?
Nasoethmoidal encephalocele. A cranial meningocele consists of a cerebrospinal fluid (CSF) filled meningeal sac only, and a cranial encephalocele contains the sac plus cerebral cortex, cerebellum, or portions of the brainstem. Infants with a cranial encephalocele are at increased risk for developing hydrocephalus due to aqueductal stenosis, Chiari malformation, or the Dandy-Walker syndrome. Examination might show a small sac with a pedunculated stalk or a large cyst like structure that can exceed the size of the cranium. The lesion may be completely covered with skin, but areas of denuded lesion can occur and require urgent surgical management. Ultrasonography is most helpful in determining the contents of the sac. MRI or CT further helps define the spectrum of the lesion. Children with a cranial meningocele generally have a good prognosis, whereas patients with an encephalocele are at risk for vision problems, microcephaly, mental retardation, and seizures. Generally, children with neural tissue within the sac and associated hydrocephalus have the poorest prognosis.

References