

## SPOT DIAGNOSIS (IMAGE GALLERY)



### **POPLITEAL WEBS**

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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**

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