and shortening of tibia and fibula. X-ray chest showed borderline cardiomegaly with narrow chest and echocardiography revealed large atrial septal defect (ASD).

What is the diagnosis?

Ellis Van Crevald Syndrome (EVC). It is a rare mesenchymal ectodermal dysplasia first described in 1940 by Richard WB Ellis of Edinburg and Simon Van Crevald of Amsterdam now known as EVC syndrome. (1) It is autosomal recessively inherited. (2) EVC is thought to be due to mutation in EVC and EVC – 2 genes located on chromosome 4p16. (3) Child usually has constant finding of polydactyly which is usually bilateral, post-axial and on ulnar side. Polydactyly of feet is present only in 10 percent of patients. (3) Mesomelic shortness of limbs affecting distal segment of limbs is present. Nails are hypoplastic, friable and sometimes absent. Disproportionate dwarfism is there which becomes apparent with subsequent growth. Ischial spurs, genu valgum, narrow chest with poorly developed ribs are present. Oral manifestations are varied including pegged teeth or hypoplastic teeth, accessory labio-gingival frenulum, dental transpositions. Cardiac abnormalities occur in 50-60 percent of cases including AV canal defect, ASD (large) single atrium. (4) One-third of these patients die at early age or infancy from cardio-respiratory problem and those who survive require multidisciplinary approach for treatment i.e. orthopaedic correction of genu valgum, amputation of extra digit, surgical repair of cardiac malformation and dental interventions. Those who survive have normal life span and intelligence in the normal range but final adult height is between 43-60 inches. (5)

References:

SPOT DIAGNOSIS (IMAGE GALLERY)

YELLOW DEPOSITS BELOW EYELIDS
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An 8-year-old girl presented with small yellow deposits on both eyelids and periorbital area lasted for one year (Figure 1). These lesions were first seen on the lower lids and then progressively enlarged and spread to the upper lids and periorbital area. She had no similar lesions elsewhere on her skin. Routine laboratory investigations; complete blood cell count, urinalysis, serum chemistry, including blood glucose, liver function tests, and lipid profile were all normal.

What is the diagnosis?
She was diagnosed as xanthelasma. Xanthelasma is the most common cutaneous xanthoma and presents as asymptomatic, generally bilaterally symmetrical yellow plaques, most commonly near the inner canthus of the eyelid (1). One half of these lesions are associated with elevated plasma lipid levels. They frequently occur in patients with type II hyperlipidemia. These asymptomatic minor growths have a tendency to progress, coalesce, and become permanent and may be disfiguring. Therapy is usually undertaken only for cosmetic reasons (2). It can be removed with trichloroacetic acid peel, surgery, lasers or cryotherapy (3, 4).
A five-month-old male child presented with an umbilical mass. It had appeared on the 8th day, 2 days after the umbilical stump has fallen off. There was a history of serous and sometimes bloody discharge from the mass. On examination, a red polypoidal, firm, non-tender umbilical mass with a nodular and glistening surface, and 2cm x 2cm in size was found. Hemogram and ultrasonography (USG) of abdomen were normal. Surgical excision of the mass was done. Histopathological examination of the excised mass showed presence of intestinal tract mucosa in it.

What is the diagnosis?

Umbilical polyp. It is a rare anomaly, resulting from persistence of all or part of the omphalomesenteric duct (OMD) or the urachus. (1) Usually umbilical polyp represents only the distal remnant of OMD or the urachus. In such cases, it is present in absence of other OMD or urachal anomalies. (2) Umbilical polyp must be differentiated from the much common umbilical granuloma. The latter appear as 1mm to 1cm pink friable lesions. Unlike umbilical granuloma, umbilical polyp does not respond to silver nitrate cauterization. (2) Most infants with umbilical polyps will have small amount of discharge, but significant and persistent discharge, particularly if it resembles intestinal content or urine, should suggest the possibility of co-existent vitelline or urachal remnants. (1,2) USG should be obtained and a fistulogram, sinogram may be occasionally useful in this setting. Histologically, the polyp consists of intestinal or urinary tract mucosa. Treatment is surgical excision of the polyp and the OMD or urachal anomalies, if associated. (1,2)

References: