A full term normal female new born without any antenatal, intranatal and postnatal issues, on day 3 of life was noticed to have developed unilateral erythema with contralateral pallor with a striking demarcation at midline (Figure 1). This unequality of skin color was noticed after the baby was put on left side with dependent side becoming erythematous. This phenomenon lasted for 20 minutes and resolved spontaneously without any other symptoms.

What is the diagnosis?

Harlequin Color Change. It was first described by Neligan and Strang in 1952. It is defined as a transient erythema involving one half of the infant’s body with simultaneous blanching of the other side with a demarcation on the midline. It is an unusual vascular phenomenon seen transiently in approximately 10 percent of healthy newborns. (1) It occurs when the newborn is placed on one side, an erythematous color change occurs on the dependent site while the upper side becomes pale and there is a pronounced demarcation line between them. The exact mechanism of this phenomenon is not known, but is thought to be due to immaturity of the hypothalamic center that controls dilation of peripheral blood vessels. (2) The Harlequin color change usually occurs on the 2nd to 5th day of life, sometimes it can be seen up to 21 days of life. (3) It resolves with crying or movement and is believed to be benign. The head and genitalia are usually spared. If the color change is persistent, a large capillary malformation may be suspected. The differential diagnosis includes port-wine stain and nascent hemangioma of infancy. (4) It does not require any treatment.

References


A 3 years old girl presented with knock knees and polydactyly. She was first in birth order and single child born to non-consanguineous married parents and had normal milestones. She also had brittle nails. On examination, she had thin hair, slightly elongated face, serrated incisor margins, absent incisor teeth, pegged teeth and transposition of teeth apart from genu valgum and polydactyly. There was no organomegaly. X-ray wrist and hand showing postaxial polydactyly, polymetacarpalism and syncarpalism with shortening of middle phalanges. X-rays of lower limbs shows ischial spurs at acetabulum level, mesical angulation
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: