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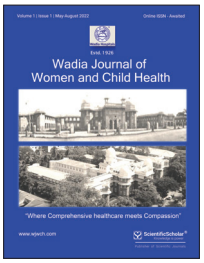

Clinical Image

A case of oro-cardio-digital syndrome

Komal Murli Shivnani[1], Bhavya Shah[1]

[1]Departmernt of Neonatology, Bai Jerbai Wadia Hospital for Children, Mumbai, Maharashtra, India.

***Corresponding author:**
Bhavya Shah,
Department of Neonatology,
Bai Jerbai Wadia Hospital
for Children, Mumbai,
Maharashtra, India.

bhavya87@gmail.com

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We report a case of a 42-day-old term female born out of 3rd degree consanguinity, delivered by cesarian section who presented with respiratory distress, skeletal abnormalities, and seizures. On examination, the baby had a broad forehead, low set ears, natal teeth, cleft of the upper lip, short stature with a short trunk, micromelic shortening of limbs [Figure 1a], bilateral dystrophic and hypoplastic nails of fingers and toes, and post-axial polydactyly of both hands [Figure 1b].

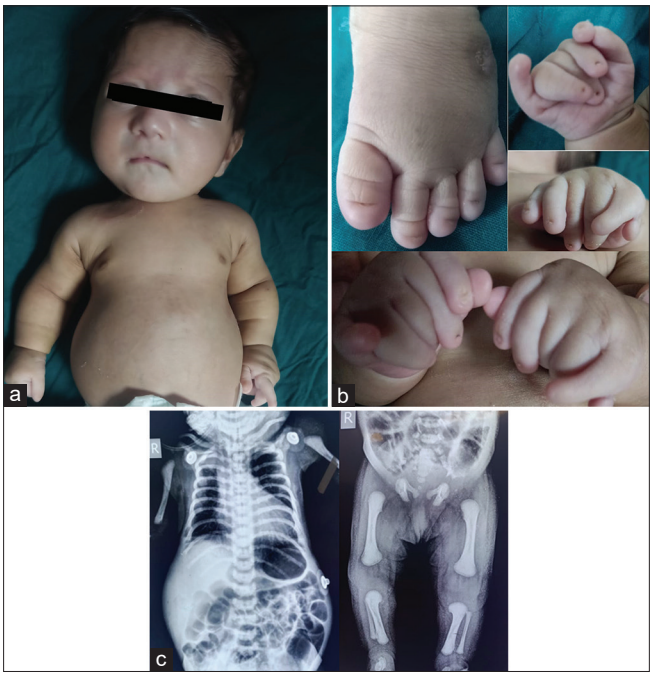

Figure 1: (a) Micromelic shortening of limbs, (b) Dystrophic nails, post-axial polydactyly, (c) Infantogram.

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Infantogram [Figure 1c] showed a narrow thorax with short ribs, cardiomegaly, all the long bones appearing thick and short with bulbous metaphysis. Echocardiogram revealed a partial atrioventricular canal defect with a large primum atrial septal defect amounting to a single atrium.

Ellis-van Creveld (EVC) syndrome was suspected in view of the above features and genetic studies for mutations in EVC and EVC2 genes have been advised for confirmation of this condition which is inherited in an autosomal recessive manner.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

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Conflicts of interest

There are no conflicts of interest.