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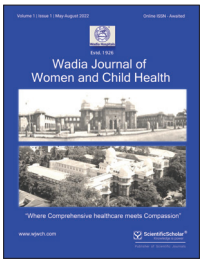

Clinical Image

A neonate with Alagille syndrome

Astha Amrit[1], Alpana Utture[1], Vaidehi Dande[1]

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

***Corresponding author:**
Alpana Utture,
Department of Neonatology,
B.J. Wadia Hospital
for Children, Mumbai,
Maharashtra, India.

utture.alpana@gmail.com

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A full term, 2.5 kg, small for gestation neonate born of a non-consanguineous marriage presented with cholestasis since the 3rd day of life. Baby was admitted in the neonatal intensive care unit for respiratory distress. Physical examination showed facial dysmorphism as broad forehead, depressed nasal bridge, low set ears, deep-set eyes, flattened nose tip with a prominent, and pointed chin making the face to appear as an inverted triangle [Figure 1]. Echocardiography showed perimembranous ventricular septal defect with large patent ductus arteriosus. Abdominal sonography showed bilateral small kidneys. Neonatal sepsis, intrauterine infections, and surgical causes of neonatal cholestasis were ruled out. Ophthalmological examination showed the presence of posterior embryotoxon. Clinical exome evaluation showed the presence of JAG1(-) mutation, variant c.2173dup (p.Asp725GlyfsTer4) suggestive of Alagille syndrome.

Alagille syndrome as a cause of neonatal cholestasis is infrequently seen in the neonatal intensive care unit.[1] Facial features often go unnoticed in neonatal period due to facial fat. However, a strong clinical suspicion can help in achieving the diagnosis aided by evaluation for cardiac, vertebral, ophthalmic, and renal anomalies.

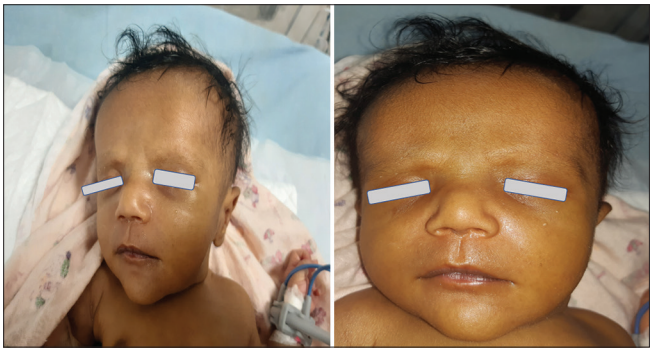

Figure 1: Image showing icterus and facial dysmorphisms as broad forehead, depressed nasal bridge, low set ears, deep-set eyes, flattened nose tip with a prominent, and pointed chin making facial shape to appear as inverted triangle.

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Declaration of patient consent

Patient's consent not required as patient's identity is not disclosed or compromised.

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Conflicts of interest

There are no conflicts of interest.

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