

What is the Diagnosis?

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Our patient was born at full term to a non-consanguineous couple and his birth weight was 3.045 kg. His perinatal history was uneventful. Newborn hearing screening detected that he had bilateral hearing impairment. The family history was unremarkable. He had poor weight gain and he was followed up at Paediatrics clinic for failure to thrive. He also had heart murmur. Echocardiogram showed that he had supravulvular pulmonary stenosis.

He was seen at Medical Genetics Clinic at 5 months old and his body weight and body length were both at 3rd centile and his head circumference was at 10th

centile. Physical examination showed that he had hypertelorism, depressed nasal bridge, brachycephaly, low set and mildly posterior rotated ears, pectus carinatum, wide spaced nipple (inter nipple distance 11 cm at 97th centile) and grade 3/6 ejection systolic murmur over left upper sternal border. The external genitalia was normal and there was no naevus. Clinical photos of the patient were shown in Figure 1.

In summary, this patient presented with failure to thrive, dysmorphism, pectus carinatum, pulmonary stenosis and bilateral hearing impairment.



Figure 1 Clinical photographs of our patient at 8 months of age (with consent for publication by parents).

The clinical quiz was prepared by:

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Answer to "Clinical Quiz" on Pages 146-147

N.B. The Editors invite contributions of illustrative clinical cases or materials to this section of the journal.