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Clinical Image

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# Alagille Syndrome: Not Always Hepatic Involvement

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## 1. Clinical Image

We present a 22-month-old girl affected by surgical palliative corrected Tetralogy of Fallot and peculiar facial features characterized by prominent forehead, deep-set eyes, pointed chin and bulbous tip of nose (Figure 1) typical of Alagille Syndrome (AS).

Hypoplasia of the distal phalanges of both feet and two butterfly vertebrae at D10-D11 on spine radiography (Figure 2) were found. Liver, kidney and eye structure and function were normal.

Genetic analysis discovered a novel missense heterozygote mutation in exon 5 (c.753G>A) of JAG1 gene, confirming AS according to the revised diagnostic criteria [1,2]. proposed by Kamath. In fact one of the classical clinical features (cholestasis, cardiac defect, skeletal abnormalities, ocular abnormalities and characteristic facial features) is enough to make the diagnosis in the presence of a positive genetic test. These new diagnostic criteria reflect the important role of Notch signalling pathway in angiogenesis and AS needs to be considered primarily as a vasculopathy where hepatic involvement not necessarily might be detected.

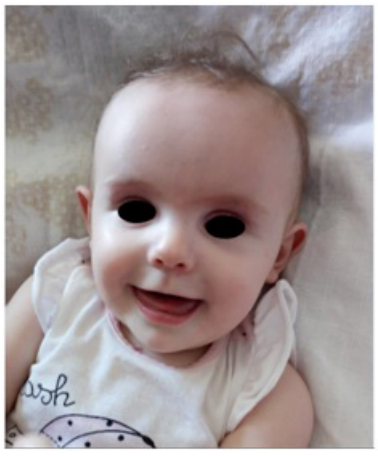

**Figure 1**

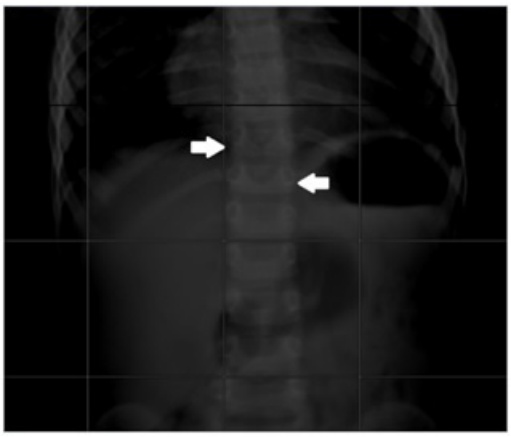

**Figure 2**

## References

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2. Kamath BM, Loomes KM, Oakey RJ, Emerick K EM, Conversano T, Spinner NB et al. Facial features in Alagille syndrome: specific or cholestasis facies? American Journal of Medical Genetics. 2002; 112:163-170.