A case of Rubinstein-Taybi Syndrome: Clinical Diagnosis and Multi-disciplinary Follow-up

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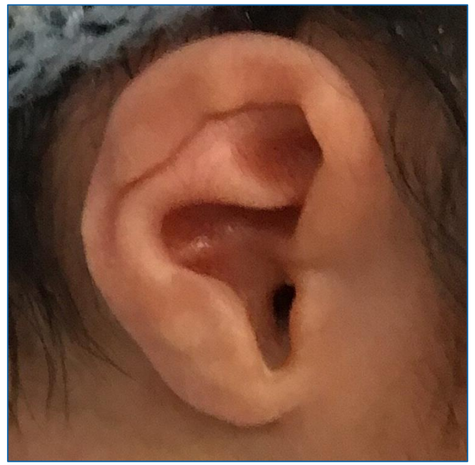
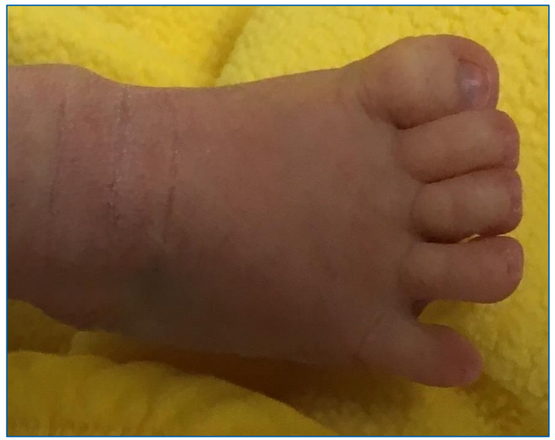
Clinical case

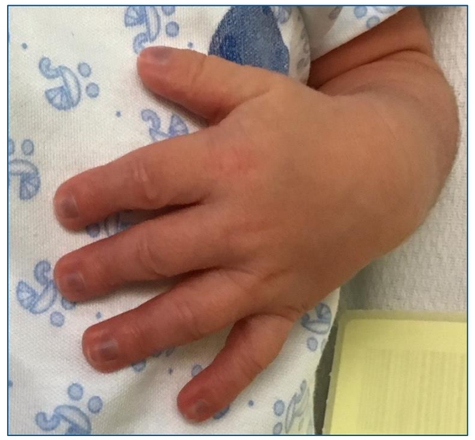
- **Birth**: 37+3/7 GW, vaginal birth induced for severe IUGR since GW 32 and polyhydramnios since GW 29; APGAR 8/9/10; PN 2250 G (P.<3 °), L. 45 cm (P. <3°), CC. 31 cm (P. <3°).

- **Dysmorphic features**: large nose with convex profile, short philtrum, micrognathia, small ears, large auditory canal, small tragus, large fontanelles, large and deviated toes and halluces, singular palmar creases.

- **Organic defects**: bicuspid aortic valve, slight ectasia of lateral ventricles, borderline medullar cone, double renal district, right cryptorchidism.







Diagnostic confirmation

Trio Exome Sequencing: 276kb *de novo* deletion on chromosome 16p13.3, comprehensive of 9 genes including CREBBP, DNASE1, SLX4.

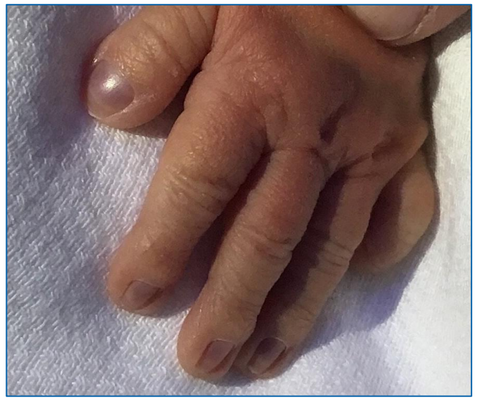
Discussion

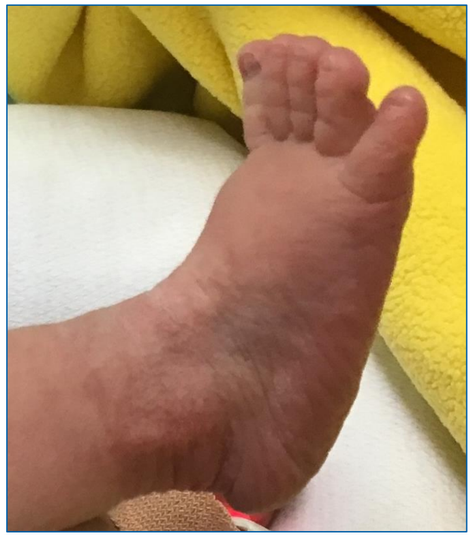
- Diagnosis: careful clinical examination and genetic confirmation.
- Transmission: autosomal dominant.
- There is no correlation between genotype and phenotype.

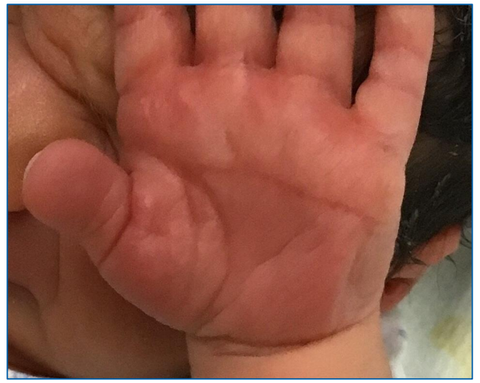
Complications

Early mortality derives from possible life-threatening complications:

- effects of a severe congenital heart disease;
- dysphagia and GERD, potentially leading to recurrent “ab ingestis” pneumonia;
- obstructive sleep apnea, potentially leading to PH with consequent RVH and HF;
- neural crest tumors.







Conclusions

- Our patient: bicuspid aortic valve with no impact on cardiac function. A multidisciplinary follow-up is planned.
- Early diagnosis results in better management, giving parents an opportunity to provide adequate follow up.
- Swiss support organizations do not exist yet.

Thank you for your attention.

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