

An infant cholestasis patient with a novel missense mutation in the aldo-keto reductase family 1 member D1 gene successfully treated by early adequate supplementation with chenodeoxycholic acid: A case report and review of literature

Wang HH *et al.* A case with the AKR1D1 mutation

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Conclusions: **Analysis** of blood samples for SRD5B1 **mutations** can be used to ... **Patients** with **genetic** 5 β -reductase deficiency may respond well to **treatment** with ... Keywords: bile acid synthesis, inborn error, giant cell hepatitis, **cholestasis**, bile ... This **infant** had failed to improve on **ursodeoxycholic acid** therapy but liver ...

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Mutations in the **novel gene** FOPV are associated with familial autosomal dominant ... Cholic **Acid** to **Treat** HSD3B7 and **AKR1D1** Deficiencies **sufficient** serum vitamin E level in the majority of children with **cholestasis**, single-center, **case-cohort study** including 16 **patients** with PSIS diagnosed before one year of age.

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Methods: This was a single-center, **case-control pilot study** conducted at ... Among SUBA, **chenodeoxycholic acid** (CDCA) was significantly elevated in A **novel** homozygous frameshift germline **mutation** (c.587delG) in the **AKR1D1 gene**; which ... management of the youngest **patient** led to **successful treatment** of the liver ...

Chapter 165. Disorders of Bile Acid Synthesis | Rudolph's Pediatrics ...

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