

Supplemental Table 1. Normal values for bile acids.

Serum (μ mol/L)		1y (N=5)	2y (N=4)	10-16y (N=7)	
CA		0.98 $\pm$ 0.84	0.98 $\pm$ 1.28	0.32 $\pm$ 0.16	
CDCA		3.69 $\pm$ 3.70	3.09 $\pm$ 1.80	1.24 $\pm$ 0.61	
UDCA		0.46 $\pm$ 0.35	1.32 $\pm$ 0.79	0.41 $\pm$ 0.45	
Allo-bile acids		0.06 $\pm$ 0.03	0.06 $\pm$ 0.03	0.02 $\pm$ 0.01	
3-Oxo- Δ 4 bile acids		0.01 $\pm$ 0.01	0.02 $\pm$ 0.01	0.01 $\pm$ 0.01	
Others		0.68 $\pm$ 0.45	0.59 $\pm$ 0.24	0.80 $\pm$ 0.34	
TBA		5.90 $\pm$ 5.18	6.05 $\pm$ 3.78	2.96 $\pm$ 1.56	
Urine (μ mol/mmol Cr)	8m (N=6)	1y (N=20)	2y (N=26)	9-14y (N=5)	
CA	0.26 $\pm$ 0.20	0.32 $\pm$ 0.29	0.19 $\pm$ 0.15	0.26 $\pm$ 0.47	
CDCA	0.69 $\pm$ 0.42	0.24 $\pm$ 0.21	0.36 $\pm$ 0.23	0.10 $\pm$ 0.08	
UDCA	0.03 $\pm$ 0.04	0.42 $\pm$ 0.87	0.35 $\pm$ 0.31	0.18 $\pm$ 0.17	
Allo-bile acids	0.01 $\pm$ 0.01	0.02 $\pm$ 0.04	0.02 $\pm$ 0.03	0.00 $\pm$ 0.00	
3-Oxo- Δ 4 bile acids	0.02 $\pm$ 0.03	0.05 $\pm$ 0.07	0.04 $\pm$ 0.04	0.01 $\pm$ 0.01	
Others	0.73 $\pm$ 0.82	0.34 $\pm$ 0.36	0.45 $\pm$ 0.37	0.13 $\pm$ 0.09	
TBA	1.71 $\pm$ 1.32	1.39 $\pm$ 1.26	1.41 $\pm$ 0.	0.65 $\pm$ 0.40	

CA, cholic acid; CDCA, chenodeoxycholic acid; UDCA, ursodeoxycholic acid; TBA, total bile acids; m, age in months; y, age in years.

Supplemental Table 2. Liver function tests and bile acid analyses in a patient with *AKR1D1* deficiency before and during CDCA treatment suspension at 2 years of age

	Before CDCA treatment suspension	During CDCA treatment suspension	
		After 2 weeks	After 4 weeks
Total bilirubin (mg/dL)	0.2	0.2	0.3
Direct bilirubin (mg/dL)	0.1	0.1	0.1
AST (U/L)	29	29	28
ALT (U/L)	13	12	12
Serum (μmol/L)			
Usual bile acids	23.8	5.2	1.5
Allo-bile acids	1.1 (4.2 %)	1.6 (18.9%)	1.4 (29.9 %)
3-Oxo Δ ⁴ bile acids	1.4 (5.2 %)	1.7 (19.7 %)	1.6 (34.3 %)
Other bile acids	0.1	0.1	0.1
Total bile acids	26.4	8.6	4.7
Urine (μmol/mmol Cr)			
Usual bile acids	1.7	0.5	0.2
Allo-bile acids	0.3	0.2	0.2
3-Oxo Δ ⁴ bile acids	37.1 (93.6 %)	30.6 (96.4)	33.7 (97.4%)
Other bile acids	0.5	0.5	0.4
Total bile acids	39.6	31.7	34.6

AST, aspartate aminotransferase; ALT, alanine aminotransferase. Usual bile acids: cholic acid, chenodeoxycholic acid, ursodeoxycholic acid, deoxycholic acid, and lithocholic acid. Allo-bile acids: allo-cholic acid and allo-chenodeoxycholic acid. 3-Oxo Δ⁴ bile acids: 7α,12α-dihydroxy-3-oxo-4-cholen-24-oic acid and 7α-hydroxy-3-oxo-4-cholen-24-oic acid.