## Supplementary Table 2. Background information, subjects with genetically undiagnosed cholestasis

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| --- | --- | --- | --- | --- |
| **Patient** | **S** | **Sampling** | **UDCA** | **Status at last** |
| **UC** |  | **age** | **in mg** | **follow-up, age** |
| P1 | F | 0.3 y | N/A | Revovered, 3 y |
| P2 | F | 0.9 y | N/A | Recovered, 3 y |
| P3 | M | 0.8 y | 50 mg bid | Recovered, 7 y |
| N4 | F | 1.3 m | N/A | Recovered, 4 y |
| P5 | M | 0.4 y | stopped 5 d | Recovered, 2 y |
| P6 | M | 0.6 y | N/A | Liver-transplanted, 1 y |
| P7 | M | 0.5 y | no UDCA | Lost follow-up  |
| P8 | M | 0.9 y | 80 mg qd | Liver-transplanted, 3 y |
| P9 | F | 0.3 y | no UDCA | N/a |
| N10 | M | 0.7 y | 83.3 mg qd | Died, 2 y |
| P11 | F | 0.7 y | N/A | Persistent cholestasis, 3 y |
| P12 | N/A | N/A | N/A | N/a |

Days or months following “stopped indicate UDCA-free interval before sampling. UC\_12 indicates raw data lost due to label contamination. P1-12 were reported in a previous study, except for P4 and P10 who were excluded from the current study after being diagnosed with *MYO5B* deficiency. 28 The N4 and N10 in the current study were two new patients. P1&P2, P12&P13 were siblings. UC, genetically undiagnosed cholestasis; S, sex; M/F, male/female; N/A, not available or lost to follow-up.