## Supplementary Table 1. Background information, subjects with *ABCB11* mutations

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| **Patient** | **S** | **Sampling** | **UDCA** | **ABCB11 mutations;** | **Status and age** |
|  |  | **age** | **in mg** | **BSEP consequences** | **at last follow-up** |
| ABCB11(NM\_003742) | | |  |  |  |
| P1 | F | 2.7 m | No | c.145C>T (p.Gln49Ter); | Liver-transplanted |
|  |  |  | UDCA | c.908+5G>A | 3.3 y |
| P2 | M | 3.3 m | No | c.145C>T (p.Gln49Ter); | Died, |
|  |  |  | UDCA | c.651G>A (p.Met217Ile) | 2 y |
| P3 | F | 2.0 y | Stopped | c.2815-8A>G; | Died, |
|  |  |  | 1 m | c.3458G>A (p.Arg1153His) | 4.8 y |
| P4 | M | 12.3 m | 30 mgbid | c.G634A (p.Ala212Thr), | Died, |
|  |  |  |  | c.A849C (p.Glu283Asp); | 1.5 y |
|  |  |  |  | c.G1638T (p.Gln546His) |  |
| P5 | M | 1.5 y | 250 mgqd | c.612- (2\_4)CTA>TT;  - | Liver-transplanted  9 y |
| P6 | M | 1.5 y | Stopped | c.1760C>G (p.Ser587Ter); | Died, |
|  |  |  | 2 m | c.3677G>C (p.Ser1226Pro) | 2 y |
| P7 | F | 3 m | 50 mgbid | c.145C>T (p.Gln49Ter); | Liver-transplanted, |
|  |  |  |  | c.1809G>C (p.Lys603Asn) | 4.5 y |
| P8 | M | 5 m | 50 mgqd | c.3457C>T (p.Arg1153Cys); | Died, |
|  |  |  |  | c.3623A>G (p.Tyr1208Cys) | 1.5 y |
| P9 | F | 4.6 y | 62.5 mg | c.1460G>A (p.Arg487His); | Recurrent cholestasis, |
|  |  |  | bid | c.3169C>T (p.Arg1057Ter) | 10 y |
| P10 | M | 3.7 m | 60 mgbid | c.409G>A (p.Glu137Lys); | Recurrent cholestasis, |
|  |  |  |  | c.2216delC (p.Pro740Glnfs\*6) | 6 y |
| P11 | F | 5.1 m | 50 mgqd | c.1493T>C (p.Ile498Thr); | Recovered, |
|  |  |  |  | c.1493T>C (p.Ile498Thr) | 4 y |
| P12 | F | 5.0 y | No  UDCA | c.2944G>A (p.Gly982Arg);  - | Persistent cholestasis,  8 y |
| P13 | M | 4.9 m | No | c.212T>A (p.Leu71His); | N/a |
|  |  |  | UDCA | c.677C>T (p.Ser226Leu) |  |
| P14 | F | 9 m | N/A | c.2104C>T (p.Gln702Ter);  - | Died,  1 y |
| P15 | F | 1.2 y | N/A | c.1496G>A (p.Gly499Glu); | Persistent cholestasis, |
|  |  |  |  | c.2606A>C (p.Gln869Pro) | 6 y |
| P16 | M | 1.7 y | 83.3 bid | c.1243C>T (p.Arg415Ter); | Persistent cholestasis, |
|  |  |  |  | c.1493T>C (p.Ile498Thr) | 3.5 y |
| P17 | F | 2.9 y | No | c.1489C>T (p.Gln497Ter); | Persistent cholestasis, |
|  |  |  | UDCA | c.2197C>T (p.Gln733Ter) | 4.2 y |

Days or months following “stopped” indicate UDCA-free interval before sampling. P1-17 have been reported in a previous study. 28 The follow-up information for P1, P5, P15, P16, and P17 has been updated. S, sex; M/F, male/female; N/A, not available or lost to follow-up; ND, not done.