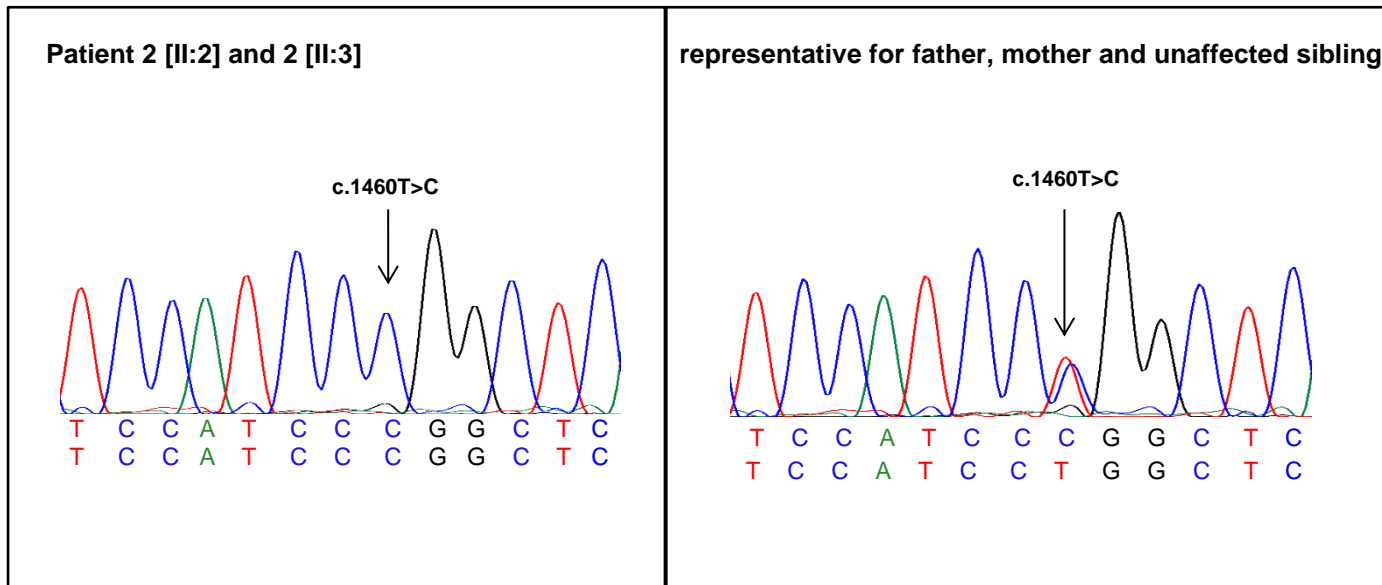
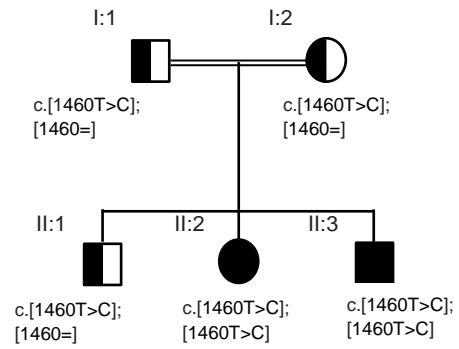


**Figure S1** Sanger sequencing electropherograms of family 1

Family 2

**CYP1B1: NM\_000104.3**



**Figure S2** Sanger sequencing electropherograms of family 2



Family 4

*CYP1B1*: NM\_000104.3

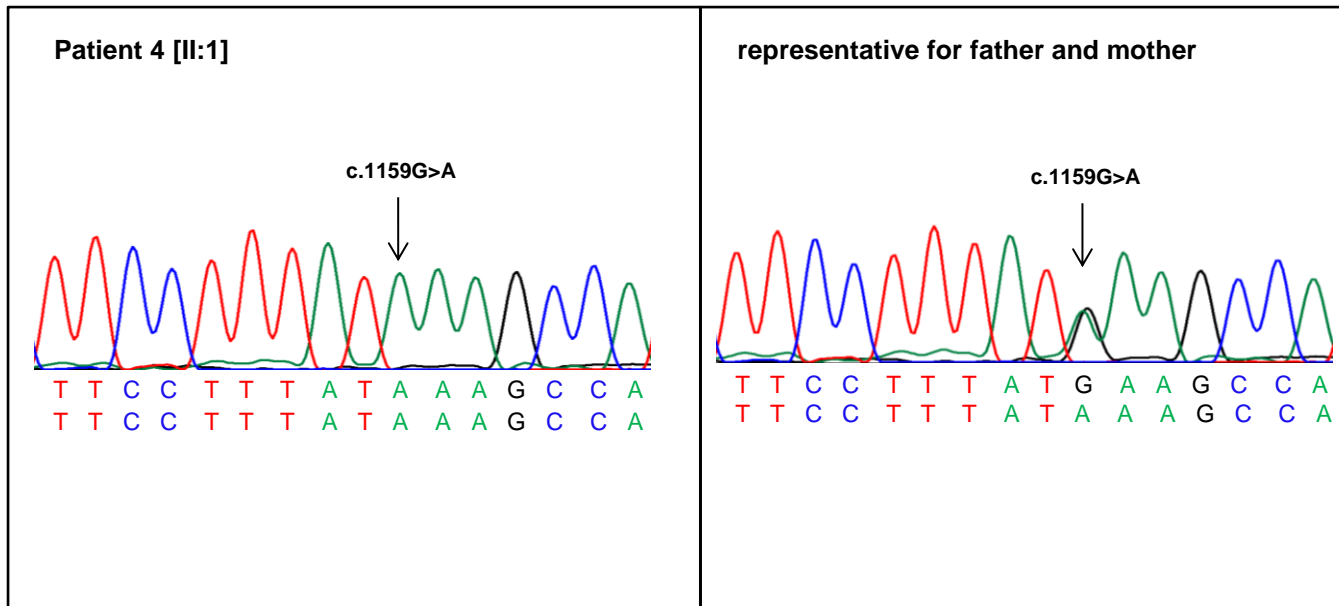
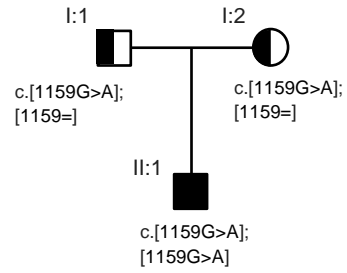
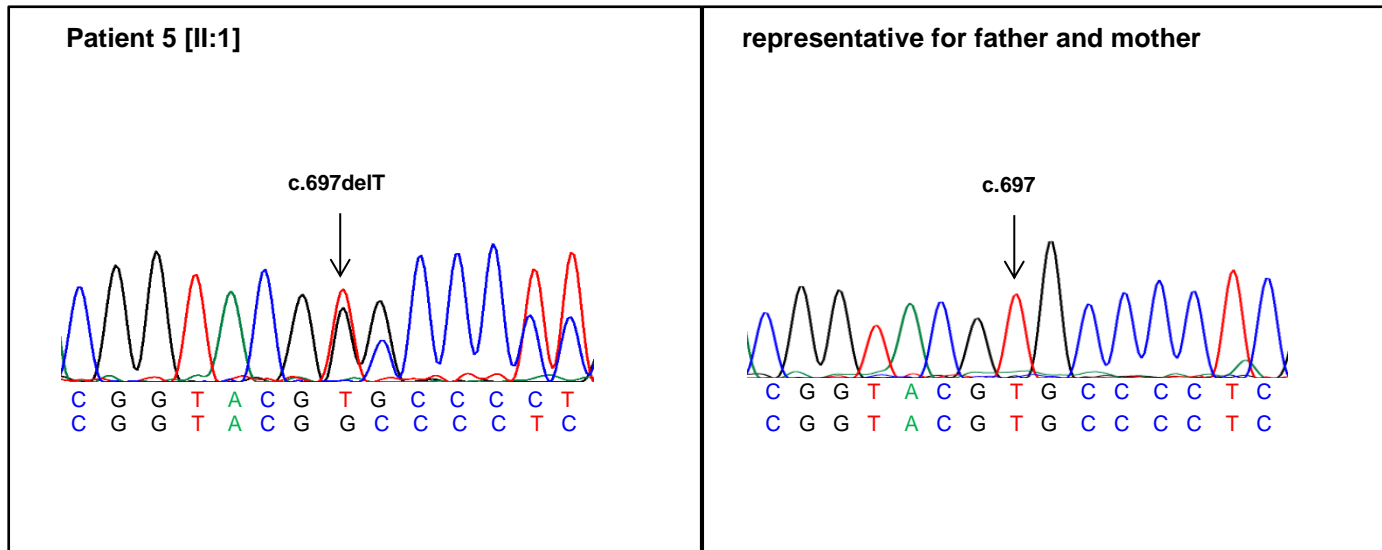
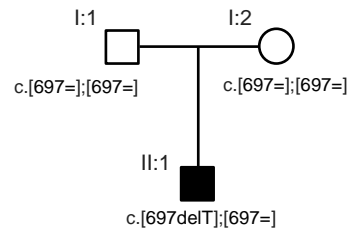


Figure S4 Sanger sequencing electropherograms of family 4

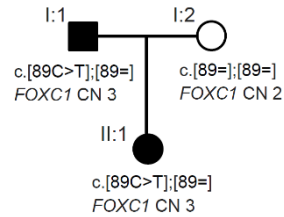
**Family 5**  
**FOXC1: NM\_001453.2**



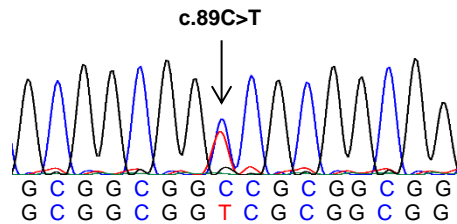
**Figure S5** Sanger sequencing electropherograms of family 5

Family 6

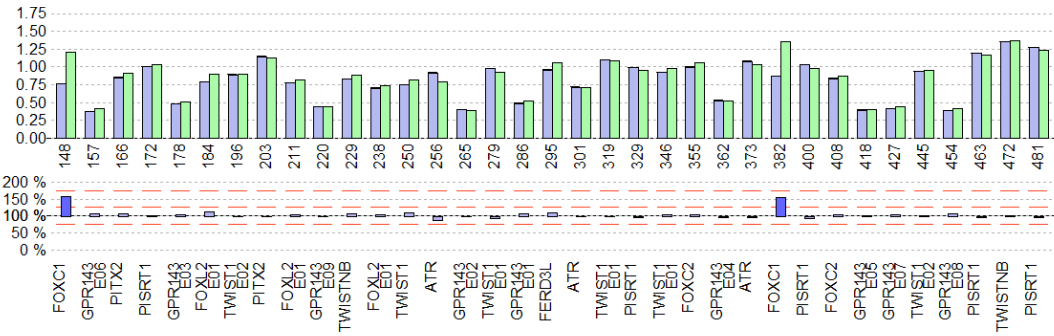
**FOXC1: NM\_001453.2**



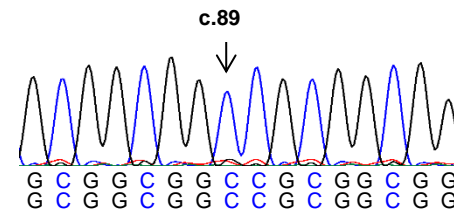
Representative for patient 6 [II:1] and 6 [I:1]



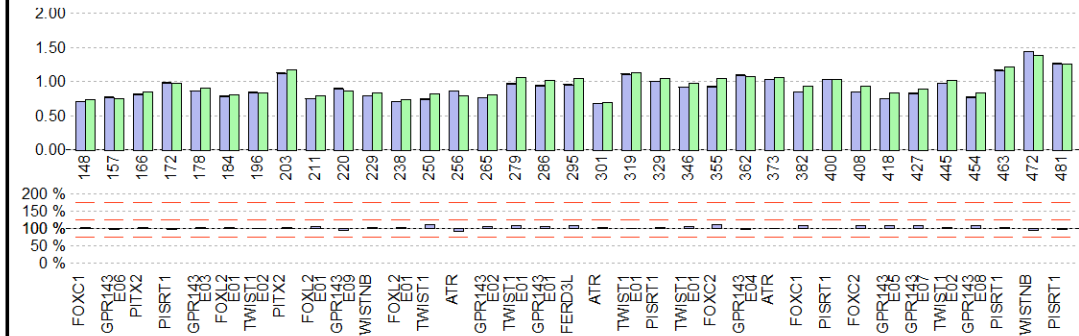
**FOXC1 copy number: 3 (MLPA)**



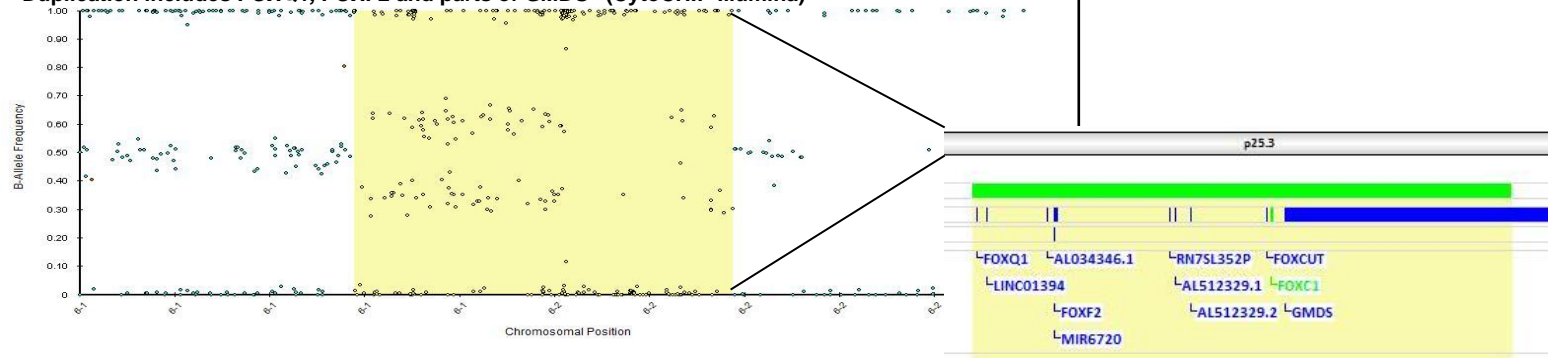
mother



**FOXC1 copy number: 2 (MLPA)**

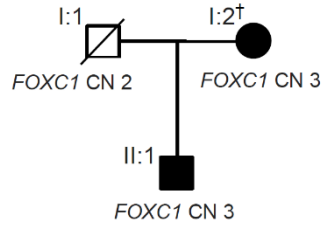


**Duplication includes FOXC1, FOXF2 and parts of GMDS (CytoCHIP Illumina)**



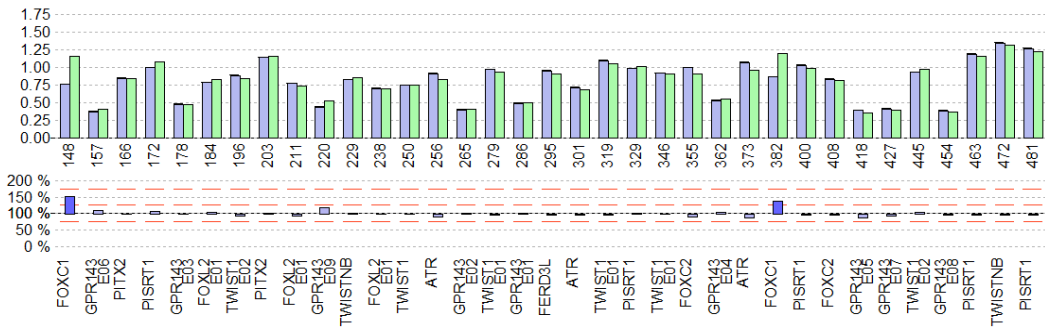
**Figure S6** Sanger sequencing electropherograms, multiplex ligation-dependent probe amplification (MLPA) results and microarray data of family 6

**Family 7**  
**FOXC1: NM\_001453.2**



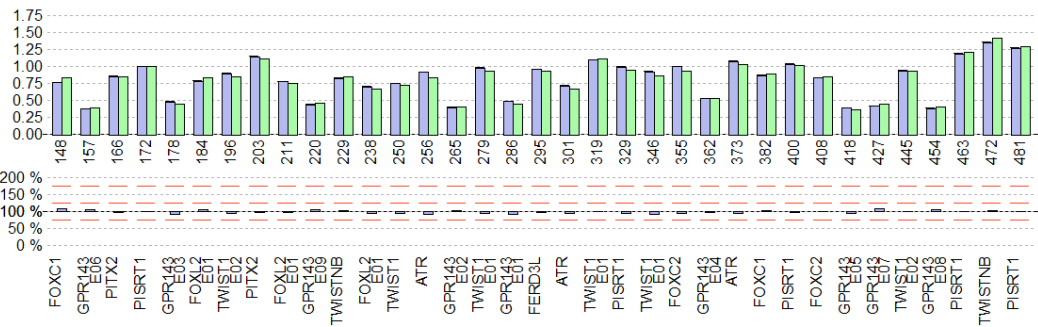
**Representative for patient 7 [II:1] and mother (patient 7 [I:2])**

**FOXC1 copy number: 3 (MLPA)**

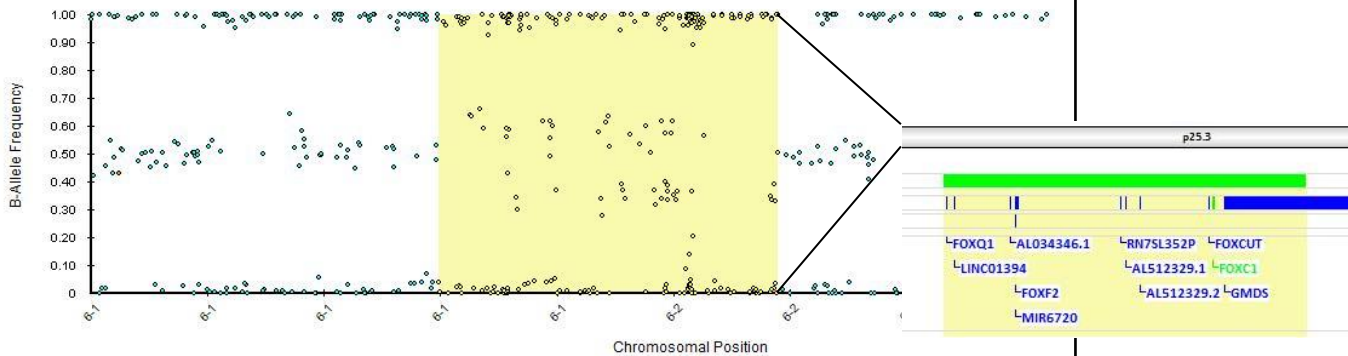


**father**

**FOXC1 copy number: 2 (MLPA)**



**Duplication includes FOXC1, FOXF2 and parts of GMDS (CytoCHIP Illumina)**



**Figure S7** Multiplex ligation-dependent probe amplification (MLPA) results and microarray data of family 7