Supplementary

Table S1 positive gene analysis

Number of cases	Diagnosis	Genetic results
1	Alagille syndrome	JAG1, NM_000214: exon12: c.1436A>G (p. Y479C) [MIM: 118450]
1	Alagille syndrome	JAG1, NM_000214: exon15: c.1932C>A (p. C644X) [MIM: 118450]
2	pigment incontinence	IKBKG, NM_003639.4: exon4~10: deletion [MIM:300248]
1	Coproporphyria	CPOX, NM_000097: exon4: c.877G>A (p. A293T) (Maternal) + NM_000097: exon7: c.1285_1287dupTAC (Paternal) [MIM: 121300]
1	Williams-Beuren syndrome	arr [GRCh37] 7q11.23 (72677225_74172862) *1 [MIM: 612545]
1	Hyper-IgD syndrome	MVK, NM_000431: exon3: c.118C>T (p. R40W) (Maternal)+ NM_000431: exon5: c.439G>A (p. A147T) [MIM:260920]
1	Niemann-Pick disease, type C1	NPC1, NM_000271: exon1: c.10del (p. R4AfsTer55) + NM_000271: exon8: c.1211G>A (p. R404Q) [MIM:257220]
1	Trisomy 21	chromosome analysis: 47, XY, +21

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