Supplemental Data Table S6. Validation rate of NewbornSeq using Sanger sequencing

Category	N of alleles screened (N of samples)	N of alleles validated (N of samples)	Validation rate
Association positive cases			
Heterozygote	53 (50)*	48 (45)	91%
Compound heterozygote	16 (8)	16 (8)	100%
Homozygote	2 (1)	2 (1)	100%
Sub total	71 (59)	66 (54)	93%
Positive cases with discrepancy			
Heterozygote	nt	nt	nt
Compound heterozygote	12 (6)	11 (6)	92%
Homozygote	16 (8)	14 (7)	88%
Sub total	28 (14)	25 (13)	89%
Total	99 (73)	91 (67)	92%

The same mutant alleles identified in different samples were counted as different alleles. Eight mutant alleles including one case with homozygote were not validated; four alleles were not amplified via PCR and four alleles were called falsely.
*One case (IMD_144) with concurrent *TSHR* and *DUOX2* mutations, another case (IMD_152) with concurrent *DUOX2* and *DUOXA2* mutations, and the other case (IMD_153) with concurrent *DUOXA2* and *PAX8* mutations.
Abbreviation: nt, not tested.