Table S3. List of recessive cases with known disease-causing genes identified by reanalysis in other articles.

ID	Gene	HGVS_g (GRCh37)	Variants	Segregation	Variant identification	ACMG/AMP Codes	Pathogenicity	Diagnosis (#OMIM)	PMID
1	*PHGDH*	*NC_000001.10:g.120284440G>A*	NM_006623.4:c.1129G>A, (p.Gly377Ser)	Unknown	Reanalysis	PM1, PP5, PM2, PP3	P	Neu-Laxova syndrome 1 [#256520]; Phosphoglycerate dehydrogenase deficiency	31216405
2	*SKIC2*	*NC_000006.11:g.31931511G>A*	NM_006929.5:c.1647+1G>A	Maternal	Initial finding	PVS1, PM2	LP	Trichohepatoenteric syndrome 2 (#614602)	32963807
		NC_000006.11:g.31931188A>G	NM_006929.5:c.1404-2 A>G	Paternal	Reanalysis	PVS1, PM2	LP		
3	*VARS1*	*NC_000006.11:g.31747238G>A*	NM_006295.3:c.3364C>T, (p.Arg1122Trp)	Maternal	Initial finding	PP3, PM2, PM3	VUS	Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy (#617802)	34560057
		NC_000006.11:g.31762906_31762925del	NM_006295.3:c.71_90del, (p.Tyr24SerfsTer18)	Paternal	Reanalysis	PVS1, PM2, PP3	P		

LP: likely pathogenic; P: pathogenic; VUS: variant of uncertain significance.