

Case	Disease Category	Original technique	CNV results (GRCh37) aCGH /MLPA size + gene of interest	CNV coordinates from NGS (GRCh37)
Case A	NDD	180k aCGH	22q13.33(51123491-51178264) x1 55kb – including <i>SHANK3</i>	chr22:51123014-51220722 x1 >97kb – including <i>SHANK3</i>
Case B	NG	MLPA PMP22	14q11.2(20002226-20404761) x1 106bp – <i>PMP22</i> (exon 5)	chr14:20002226-20404761 x1 106bp – <i>PMP22</i> (exon 5)
Case C	NDD	180k aCGH	16q11.2(28824794-29042118) x1 217kb – including <i>SH2B1</i>	chr16:28834582-29064047 x1 >229kb – including <i>SH2B1</i>
Case D	NDD	180k aCGH	Xp11.22(52954520-53742476) x3 >788kb – including <i>HUWE1</i>	chrX:53105805-53681051 x3 >575kb - including <i>HUWE1</i>
Case E	NDD	180k aCGH	22q11.21(18894835-21801661) x3 >2.9Mb	chr22:18893889-21481925 x3 >2.58Mb

Kb: Kilobase pair; Mb: Megabase pair; NDD: Neurodevelopmental disorder; NG: Neurodegeneration.

Table S2: List of patients from Phase 1, to parametrize the NGS-based CNV pipeline.