

Table S4. Whole chromosomal duplications detected by ExomeDepth in two patients referred for WES.

Patient	Karyotype	Disease	ExomeDepth predicted coordinates (Grch37/hg19)	CNV size	Reads Ratio
279	47,XX,+21	Down syndrome (Trisomy 21)	DUPLICATION: chr21:10952913_48084286	37,1 Mb	1.31
35	47,XXY	Klinefelter syndrome	DUPLICATION: chrX:200855_155240074	155 Mb	1.8

Mb: Megabase pair