Supplementary Table 1 Genomic coordinates and patient phenotypes of rare overlapping inherited CNVs

Internal	Type	Chr	Cytoband	Chromosome region HG19	Length	Genes affected	Inheritance	Classification
SKZ_1112	Gain	3	p14.1	chr3:64992802-65051455	58653	*ADAMTS9-AS2, BC040632*	no parental DNA	Uncertain
SKZ_0999	Gain	3	p14.1	chr3:64993928-65024494	30566	*ADAMTS9-AS2*	maternal	Uncertain : likely benign
DE82OSOUKBD100013	Loss	8	p23.2	chr8:3619348-3650542	31195	*CSMD1*	undetermined	Benign
DE27OSOUKBD100324	Loss	8	p23.2	chr8:4323965-4359306	35342	*CSMD1*	undetermined	Benign
DE37OSOUKBD100144	Loss	9	p24.1	chr9:8802640-8836439	33800	*PTPRD*	undetermined	Uncertain- likely benign
DE60OSOUKBD100312	Loss	9	p23	chr9:9919638-9990269	70632	*PTPRD*	undetermined	Uncertain- likely benign
DE63OSOUKBD100161	Gain	9	q34.3	chr9:138147997-138238503	90507	*C9orf62*	undetermined	Benign
DE63OSOUKBD100161	Gain	9	q34.3	chr9:138147997-138238503	90507	*C9orf62*	undetermined	Benign
DE14OSOUKBD100461	Loss	10	p12.33	chr10:18550958-18592873	41916	*CACNB2*	undetermined	Uncertain
DE37OSOUKBD100047	Loss	10	p12.33	chr10:18550958-18583866	32909	*CACNB2*	undetermined	Uncertain
SKZ_0773	Loss	11	q22.1	chr11:99497530-99583304	85774	*CNTN5*	undetermined	Benign
DE43OSOUKBD100133	Loss	11	q22.1	chr11:99519242-99568648	49407	*CNTN5*	undetermined	Benign
DE76OSOUKBD100315	Loss	11	q22.1	chr11:99519242-99568648	49407	*CNTN5*	undetermined	Benign
SKZ_1432	Loss	11	q22.1	chr11:99519242-99568648	49406	*CNTN5*	no parental DNA	Benign
DE35OSOUKBD100471	Gain	12	q24.12 - q24.13	chr12:112183225-112327166	143942	*ACAD10- MAPKAPK5*	undetermined	Benign
SKZ_1516	Gain	12	q24.12	chr12:112183225-112327166	143941	*ACAD10- MAPKAPK5*	maternal	Benign
148-04-01	Gain	12	q24.12	chr12:112183921-112318246	134325	*ACAD10- MAPKAPK5*	maternal	Benign
SKZ_1976	Gain	13	q31.3	chr13:92958981-93035128	76147	*GPC5*	maternal	Benign
SKZ_1746	Gain	13	q31.3	chr13:92979291-93049171	69880	*GPC5*	maternal	Benign
DE44OSOUKBD100309	Gain	15	q26.3	chr15:102325461-102373183	47723	*OR4F6, OR4F15*	undetermined	Benign
DE76OSOUKBD100315	Gain	15	q26.3	chr15:102332446-102368280	35835	*OR4F6, OR4F15*	undetermined	Benign
SKZ_2111	Gain	16	p13.11	chr16:14985615-17000304	2014689	*NOMO1-NPIP*	*de novo*	Uncertain : likely pathogenic
SKZ_1988	Gain	16	p13.11	chr16:15034035-15998820	964785	*NPIP-FOPNL*	paternal	Uncertain
SKZ_1150	Gain	16	p13.11	chr16:15539023-16291541	752518	*C16ORF45-ABCC6*	paternal	Uncertain
DE45OSOUKBD100291	Loss	16	q24.1	chr16:84429584-84490643	61060	*ATP2C2*	undetermined	Benign
SKZ_1500	Loss	16	q24.1	chr16:84435058-84470476	35418	*ATP2C2, KIAA0703*	maternal	Benign
DE94OSOUKBD100282	Gain	X	p22.33	chrX:370599-612414	241816	*SHOX*	undetermined	Uncertain- likely benign
SKZ_1508	Gain	X	p22.33	chrX:405941-596245	218468	*SHOX*	maternal	Uncertain- likely benign
SKZ_0374	Gain	X	p22.33	chrX:407015-1008051	788690	*SHOX*	maternal	Uncertain- likely benign

Casenr	Gender	Major anomalies
SKZ_1112	M	EA
SKZ_0999	M	EA/TEF
DE82OSOUKBD100013	F	EA, developmental delay
DE27OSOUKBD100324	F	EA, Atrial septum defect I, tracheomalacia
DE37OSOUKBD100144	M	EA
DE60OSOUKBD100312	F	EA, alopecia, Cafe-au-lait spots, eye anomalies
DE63OSOUKBD100161	F	EA, vesicourethral reflux, PUV
DE14OSOUKBD100461	F	EA
DE37OSOUKBD100047	M	EA, tracheomalacia
SKZ_0773	F	EA/TEF, vertebral anomalies, anal , genital and ear anomalies, renal anomalies, upper limb anomalies, cleft lip+jaw+palate, duodenal atresia
DE43OSOUKBD100133	F	EA, brochial anomalies
DE76OSOUKBD100315	F	EA, lung anomalies
SKZ_1432	M	EA/TEF, anal anomalies, renal anomalies, urethral fistula and atresia, genital anomalies
DE35OSOUKBD100471	M	Esophageal stenosis, achalasia
SKZ_1516	F	EA/TEF, ventricular septal defect, right sided aortic arch
148-04-01	F	TEF, ventricular septal defect, anal atresia and vertebral anomalies
SKZ_1976	M	EA/TEF, tracheo-laryngomalacia
SKZ_1746	M	EA/TEF
DE44OSOUKBD100309	F	EA, tracheomalacia
DE76OSOUKBD100315	F	EA, lung anomalies
SKZ_2111	M	TEF, anal anomaly, multiple VSDs, hypospadia
SKZ_1988	F	EA/TEF, anal anomalies,
SKZ_1150	F	EA/TEF + Atrio-ventricular septal defect
DE45OSOUKBD100291	F	EA, hypoplastic thumb, Atrial septum defect II, kidney anomalies, tracheomalacia
SKZ_1500	F	EA/TEF, anorectal anomaly,
DE94OSOUKBD100282	M	EA, tracheomalacia, scoliosis, undescended testicle
SKZ_1508	M	EA/TEF, upper limb anomalies Atrial septum defect, dysmorphisms