Figure S1. Genomic distribution of CNVs detected in the present study, among all the individuals analysed. Chromosomal regions for CNVs detected in more one affected family member are marked. Blue indicates copy number gain; pink indicates copy number loss. CNVs, copy number variants; chr, chromosome.

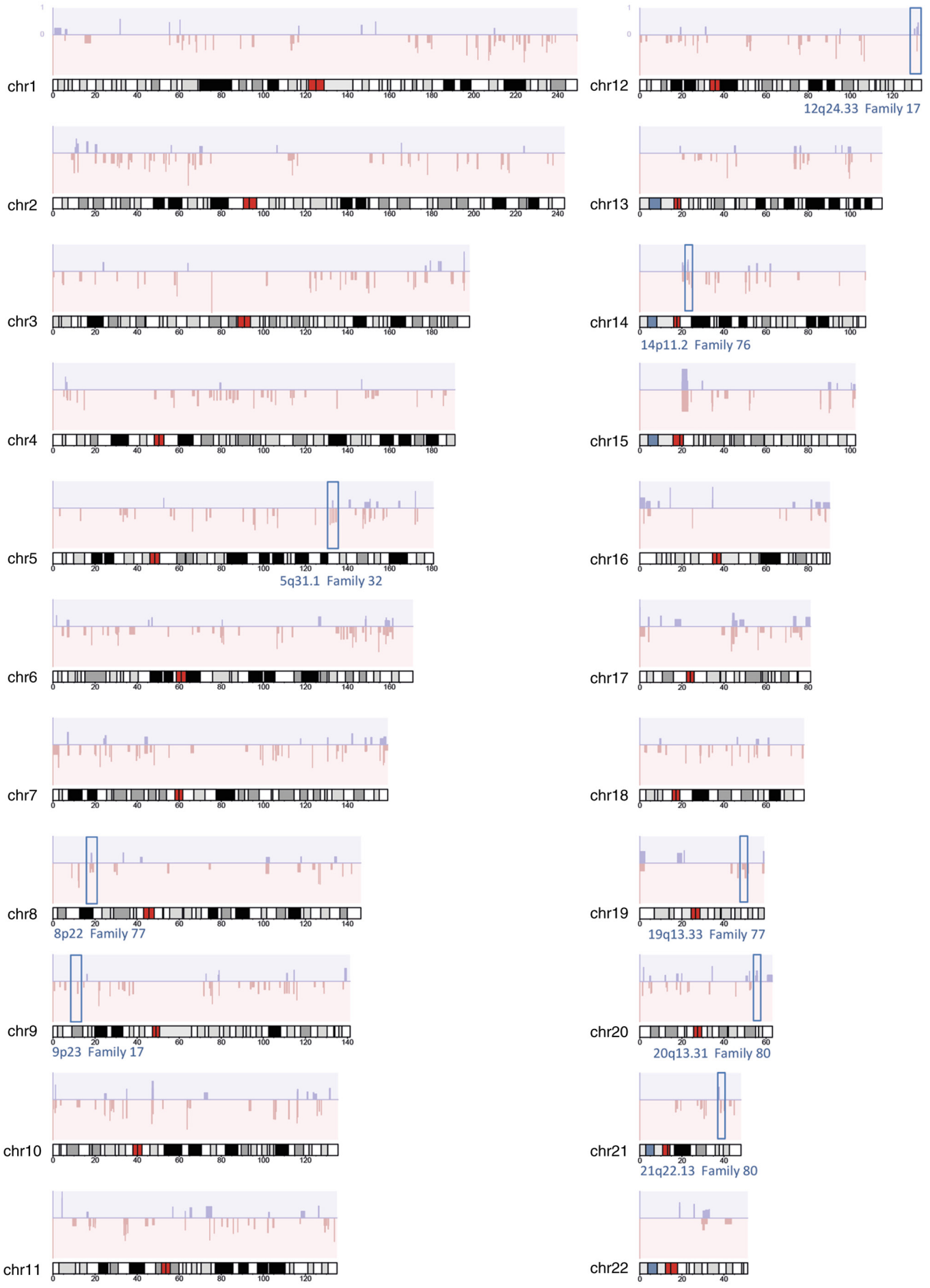

Table SI. Additional malformations of the UT and other organ systems.

Family-individual	Anomaly UT	Extra-renal anomaly
17-351		Scoliosis, MI, JIA, suspected Marfan syndrome
19-357		CP, diplegia, autism, mild intellectual disability
32-667		VSD
32-395	Bladder diverticula diverticula	
49-391		Epilepsy
49-604	Bilateral-duplex kidney	
76-648	Bladder diverticula	
78-646		VSD, cerebral-, skeletal-, tracheal malformations
79-715	Unilateral megaureter	
80-682		Epilepsy, ADHD, dyslexia
82-705	Bladder diverticula	
83-657		Ulcerative proctitis

CP, cerebral palsy; JIA, juvenile idiopathic arthritis; MI, mitral insufficiency; UT, urinary tract; VSD, ventricular septal defect.

Table SII. CNV shared by two or more affected individuals within the families, detailing CNV locus and characteristics.

A, Family 32

	CNV position					
Individual	Genomic location	Cytogenetic band	Duplication/ deletion, log2	Size of region, Mb	CNV region shared in family, %	Population frequency, %
656[a]	Chr5:132581687-132690433	5q31.1	-0.518	0.108	87	0.02
395[a]	Chr5:132596102-132690433		-0.554	0.094	100	
236[a]	Chr5:132596102-132690433		-0.513	0.094	100	

B, Family 77

	CNV position					
Individual	Genomic location	Cytogenetic band	Duplication/ deletion, log2	Size of region, Mb	CNV region shared in family, %	Population frequency, %
645[a]	Chr8:18100755-18457679	8p22	0.368	0.356	70	0.02-0.15
690[a]	Chr8:18188774-18437512		0.288	0.248	100	

C, Family 17

	CNV position					
Individual	Genomic location	Cytogenetic band	Duplication/ deletion, log2	Size of region, Mb	CNV region shared in family, %	Population frequency, %
347[a]	Chr9:11647401-11913220	9p23	-0.449	0.27	96	0.02-0.24
351[a]	Chr9:11647401-11901376		-0.45	0.25	100	

D, Family 17

	CNV position					
Individual	Genomic location	Cytogenetic band	Duplication/ deletion, log2	Size of region, Mb	CNV region shared in family, %	Population frequency, %
347[a]	Chr12:131734725-131813804	12q24.33	-0.59	0.079	100	0.61-1.0
351[a]	Chr12:131734725-131813804		-0.605	0.079	100	

E, Family 76

	CNV position					
Individual	Genomic location	Cytogenetic band	Duplication/ deletion, log2	Size of region, Mb	CNV region shared in family, %	Population frequency, %
644	Chr14:22653758-23029819	14q11.2	0.247	0.376	39	0.02-0.59
653	Chr14:22737977-23029875		0.359	0.291	50	
660[a]	Chr14:22855145-23000062		0.439	0.144	100	
650[a]	Chr14: 22487698-23029819		0.171	0.54	27	

F, Family 77, affected members

	CNV position					
Individual	Genomic location	Cytogenetic band	Duplication/ deletion, log2	Size of region, Mb	CNV region shared in family, %	Population frequency, %
645[a]	Chr19:48715269-50281369	19q13.33	-0.221	1.566	18	1

Table SII. Continued.

F, Family 77, affected members

Individual	CNV position: Genomic location	Cytogenetic band	Duplication/ deletion, log2	Size of region, Mb	CNV region shared in family, %	Population frequency, %
690[a]	Chr19:49998299-50506366		-0.55	0.508	56	

G, Family 80

Individual	CNV position: Genomic location	Cytogenetic band	Duplication/ deletion, log2	Size of region, Mb	CNV region shared in family, %	Population frequency, %
652[a]	Chr20:54967565-55918939	20q13.31	0.214	0.951	18	0.02
695	Chr20:55402690-55919277		0.398	0.508	56	

H, Family 80

Individual	CNV position: Genomic location	Cytogenetic band	Duplication/ deletion, log2	Size of region, Mb	CNV region shared in family, %	Population frequency, %
658[a]	Chr21:38260090-38712209	21q22.13	-471	0.452	70	0.37-0.48
682[a]	Chr21:38399356-38784624		-312	0.385	81	
652[a]	Chr21:37803667-41148845		0.104	3.345	10	0.02

[a]Affected family members. CNV, copy number variants. CNVs with different genomic locations in the same family are presented separately.