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Supplemental Data

Integrin Alpha 8 Recessive Mutations Are Responsible for Bilateral Renal Agenesis in Humans

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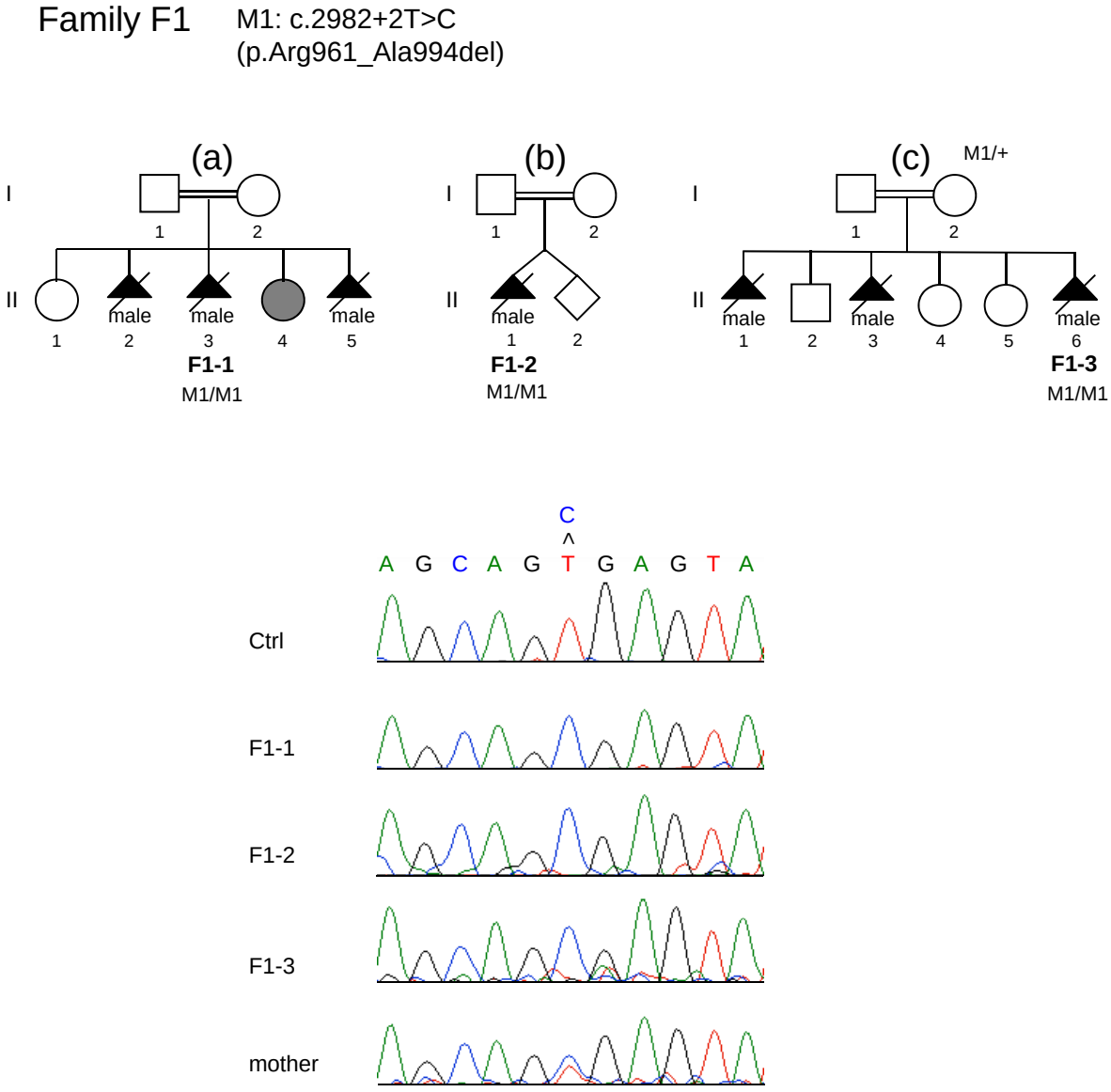

Figure S1. Pedigrees of the three branches of family F1 (a, b and c) showing the studied samples: F1-1 (II-3 in pedigree a), F1-2 (II-1 in pedigree b), F1-3 (II-6 in pedigree c) and the mother of fetus F1-3 (I-2 in pedigree c). Presence of the mutation in the three fetuses (homozygous) and the mother (heterozygous) is visualized on Sanger sequencing chromatograms.

Gene	Name	Variant position	Sequence	Nucleotidic change	Class of mutation	Protein change	Polyphen2/Sift	ho/he
Family F1/case F1-1(WES)								
ITGA8	Integrin α8	10:15570047	NM_003638.1	c.2982+2T>C	5'-splice	p.Arg961_Ala994del	-	ho
FFAR4	free fatty acid receptor 4	10:95347195	NM_001195755.1	c.915G>T	missense	p.Trp305Cys	0.982/0.02	ho
ASB11	ankyrin repeat and SOCS box containing 11	X:15301701	NM_001012428.2	c.835G>A	missense	p.Gly279Ser	0.999/0	Hem
Family F2/cases F2-1 and F2-2 (WES)								
ZSCAN20	zinc finger and SCAN domain containing 20	1:33944890	NM_145238.3	c.1A>G	start loss	p.Met1Val	-	he
		1:33957219		c.1361G>A	missense	p.Cys454Tyr	0.992/0	he
ITGA8	Integrin α8	10:15649814	NM_003638.1	c.1622_1626del AGGTG	frameshift	p.Glu541Alafs*12	-	he
		10:15686209		c.1219 G>A	missense	p.Gly407Arg	1/0,01	he
CCDC153	coiled-coil domain containing 153	11:119061052	NM_001145018.1	c.590T>C	missense	p.Leu197Pro	0.853/0	he
		11:119063873		c.337C>T	missense	p.Arg113Trp	0.892/0	he
NACAD	NAC alpha domain containing	7:45121237	NM_001146334.1	c.4220A>G	missense	p.Lys1407Arg	0.499/0	he
		7:45125615		c.164C>T	missense	p.Thr55Met	0.518/0.02	he
Simplex case BRA-1 (Sanger sequencing)								
ITGA8	Integrin α8	10:15714661	NM_003638.1	c.764 C>T	missense	p.Thr255Met	0.999/0.00	he

ho: homozygous, he: heterozygous, Hem: hemizygous

Table S1. Mutations identified by Whole Exome Sequencing and Sanger sequencing