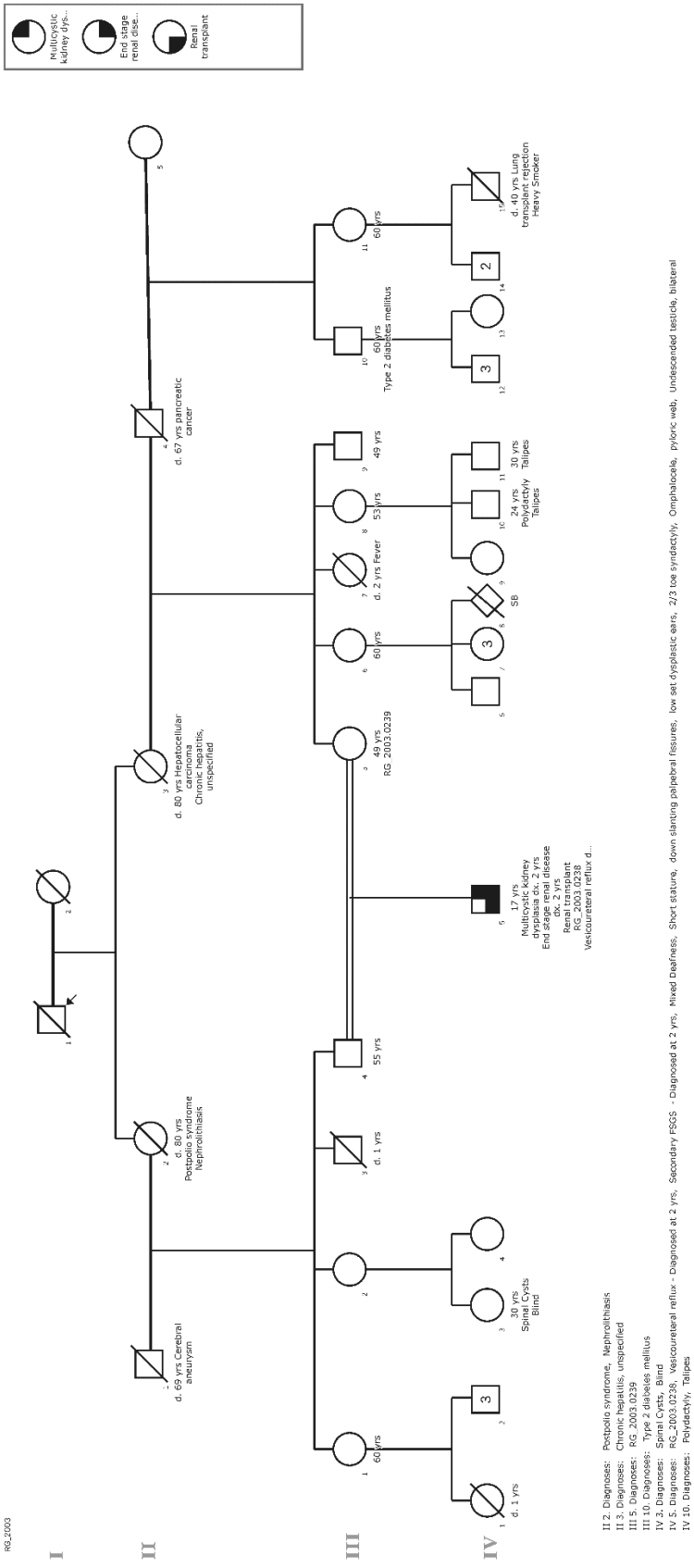

Figure S1 – Pedigree of the affected proband

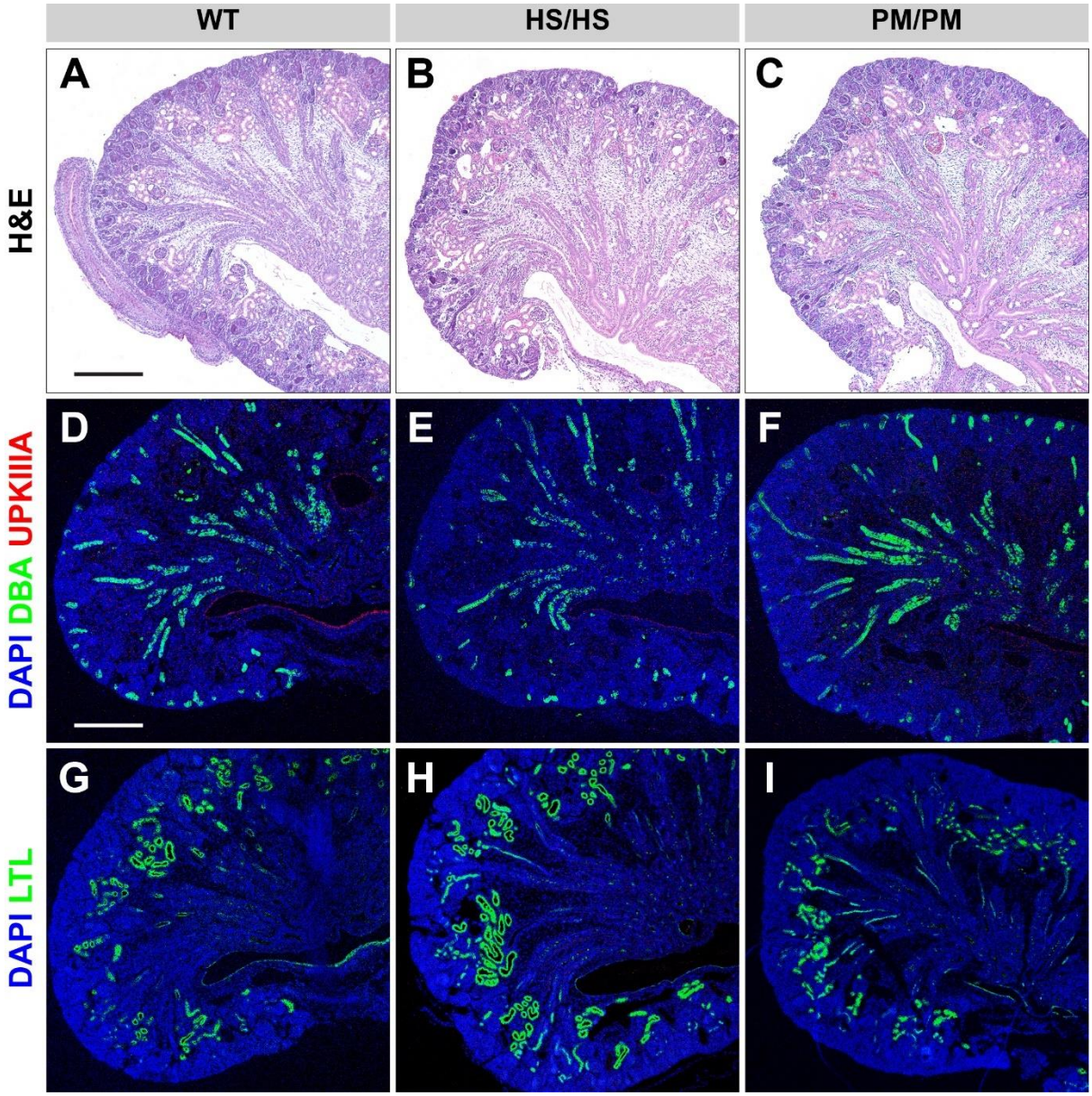

Figure S2

Patterning of the collecting duct and nephron segments in mice of different genotypes indicate that fetal kidney development is unaffected by the introduction of CRISPR mediated changes in *Lama5*.

Table S1: Homozygous candidate variants

CHROM:POS:REF:ALT	HGVS	Symbol	Impact	CADD[1]	GnomAD	
					Popmax AF[2]	Hom Alt count[3]
4:71394470:G:A	NM_212557.4:c.325G>A (p.Ala109Thr)	AMTN	Missense	22.7	0.00609089	3
6:126236629:G:A	NM_181782.5:c.2244+3G>A	NCOA7	Splice Region	14.2	0.00137387	0
9:71836037:C:T	NM_001170416.2:c.670C>T (p.Arg224Trp)	TJP2	Missense	23.0	0.000225225	0
9:79322098:T:C	NM_015225.3:c.5092A>G (p.Ile1698Val)	PRUNE2	Missense	0.0	3.27E-05	0
9:84205855:C:T	NM_005077.5:c.1694G>A (p.Arg565His)	TLE1	Missense	31.0	8.79E-06	0
12:25261759:T:TAAAAAAAAAAAAAA	NM_018272.5:c.1894-16_1894-3dupTTTTTTTTTTTTTT	CASC1	Splice Region	12.6	n.f.	n.f.
12:108988326:C:CAAAAAAAAAA	NM_181724.3:c.-14-2163_-14-2154dupTTTTTTTTT	TMEM119	Splice Region	0.6	n.f.	n.f.
13:32367033:C:G	NM_130806.5:c.1594C>G (p.Arg532Gly)	RXFP2	Missense	23.0	0.00155521	0
13:45150143:G:C	NM_183422.4:c.68C>G (p.Ala23Gly)	TSC22D1	Missense	26.0	n.f.	n.f.
13:46124056:T:C	NM_182542.3:c.1618A>G (p.Thr540Ala)	ERICH6B	Missense	25.1	0.00530845	8
14:73996946:C:T	NM_203309.2:c.-72-7018G>A	HEATR4	Splice Region	0.4	0.00673563	0
16:47005573:T:TAAAAAAAA	NM_005880.4:c.139-97_139-90dupTTTTTTTT	DNAJA2	Splice Region	3.9	n.f.	n.f.
19:9075370:T:C	NM_024690.2:c.12076A>G (p.Thr4026Ala)	MUC16	Missense	0.0	0.00911944	8
19:12991919:G:C	NM_001375.3:c.134C>G (p.Ala45Gly)	DNASE2	Missense	9.1	0.00789852	5
19:17445470:C:T	NM_020959.3:c.10G>A (p.Ala4Thr)	ANO8	Missense	11.6	0.00865209	9
20:5528409:G:C	NM_019593.5:c.1917C>G (p.Ser639Arg)	GPCPD1	Missense	22.2	n.f.	n.f.
20:18295980:C:T	NM_001083330.4:c.482C>T (p.Thr161Ile)	ZNF133	Missense	11.8	0.00466805	0
20:60926966:C:A	NM_005560.6:c.857G>T (p.Arg286Leu)	LAMA5	Missense	32.0	n.f.	n.f.
22:47164079:G:A	NM_014346.5:c.62+5334G>A	TBC1D22A	Splice Region	8.7	0.00331169	0

1) CADD *In silico* damage prediction score. Scores > 20 are predicted to be damaging. CADD v1.6 (Rentzsch P et al, NAR, 2019).
2) Maximum population allele frequency observed in gnomAD v2.1.1 database.
3) Number of individuals in gnomAD v2.1.1 database homozygous for the variant.

Table contains all variants homozygous variants identified in the proband that meet all of the following criteria:

- PASS GATK VQSR quality filters.
- Homozygous in the proband and heterozygous in the probands mother.
- Maximum population Allele frequency < 0.01 (gnomAD v2.1.1).
- Fewer than 20 homozygous individuals observed in gnomAD v2.1.1.
- Predicted to alter the protein coding sequence of one or more gencode tsl1 transcripts.

Reference
Rentzsch P, Witten D, Cooper GM, Shendure J, Kircher M. CADD: predicting the deleteriousness of variants throughout the human genome. Nucleic Acids Res. 2019 Jan 8;47(D1):D886-D894.