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Supplemental Data

CRB2 Mutations Produce a Phenotype Resembling Congenital Nephrosis, Finnish type, with Cerebral Ventriculomegaly and Raised Alpha-Fetoprotein

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Figure S1. Pedigree and Chromatograms for Families 1-3 with Mutations in *CRB2*.

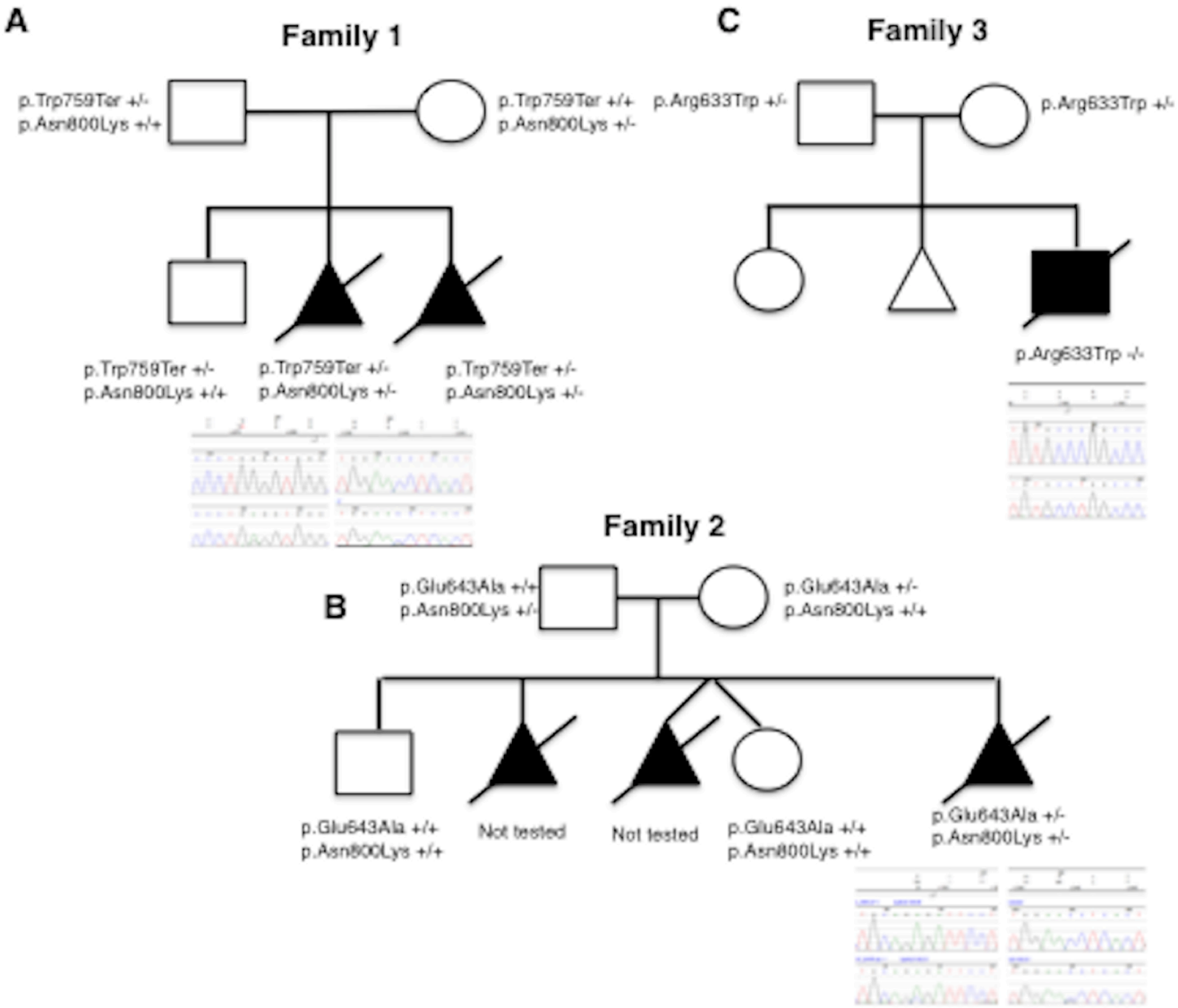

Figure Legend for Supplemental Figure 1.

Figure S1. Pedigree and Chromatograms for Families 1-3 with Mutations in *CRB2*.

Figure S1A. Pedigree for Family 1, showing c.2277G>A, predicting p.(Trp759Ter) in *CRB2* that was paternally inherited, and c.2400C>G, predicting p.(Asn800Lys) in *CRB2* that was maternally inherited in both affected children. The unaffected son is a carrier of p.(Trp759Ter) in *CRB2*.

Figure S1B. Pedigree for Family 2, showing c.2400C>G, predicting p.(Asn800Lys) in *CRB2* that was paternally inherited and c.1928A>C, predicting p.(Glu643Ala) in *CRB2* that was maternally inherited. Two healthy children tested negative for both mutations.

Figure S1C. Pedigree for Family 3, showing homozygosity for c.1897C>T, predicting p.(Arg633Trp) in *CRB2*. Both parents were carriers of the same mutation.

Table S1. Clinical Features Associated with Cerebral Ventriculomegaly and Congenital Nephrosis in Published Patients

	Jolly et al., 2003[37] (Fam. A Sib 1)	Jolly et al., 2003[37] (Fam. A Sib 2)	Jolly et al., 2003[37] (Fam. A Sib 3)	Jolly et al., 2003[37] (Fam. B Sib 1)
Age at Diagnosis	20w[e] gestation	14w gestation	12w gestation	15w gestation
Central Nervous System Ventriculomegaly Grey matter heterotopias	+; 'mild'	+; 'mild'		+
Cardiac Ventricular septal defect Pericardial effusion	+			+
Renal Hydronephrosis Microcysts Hyperechogenicity	+	+		+
Outcome	TOP[f]	TOP	TOP	TOP
Amniotic Fluid Testing MSAFP[a] AFAFP[b] AF Acetylcholinesterase[c]	27/29 MoM[g]	17/19 MoM 13 MoM	8 MoM	21 MoM
Investigations Karyotype Array CGH[d] *NPHS1* Sequencing		46,XX Negative		46,XX

MSAFP[a] = Maternal serum alpha-fetoprotein; AFAFP[b] = Amniotic fluid alpha-fetoprotein; AF Acetylcholinesterase[c] = Amniotic fluid acetylcholinesterase; Array CGH[d] = array comparative genomic hybridization; w[e] = weeks; TOP[f] = Termination of Pregnancy; MoM[g] = Multiple of Median.