

Supplement 10

Table

Family	Individual (gender)	BEEC phenotype	Nucleotide Change (Transcript) Chr. Pos. (GRCh37/hg19)	Amino Acid Change (conserved down to) <i>Protein Domain</i>	ACMG (Classification of variants)	rsNumber gnomAD Allele Frequencies (hom/het/wildtype) [ethnicity, gender]
Family 1*	I.1 Female	cloacal exstrophy	c.709G>A (ENST00000272542.7) Chr2: 113414749	p.Gly237Arg (danio rerio) TMD 7	Pathogenic	-
Family 2	I.1 female	classic bladder exstrophy	c.893T>C (ENST00000272542.7) Chr2: 113416516	p.Val298Ala (mus musculus) Intracellular loop between TMD 7 and TMD 8	Pathogenic	- 0.000003979 (0/1/251332) [1 Latino female]
Family 3	I.1 male	classic bladder exstrophy	c.1321A>C (ENST00000272542.7) Chr2: 113417053	p.Lys441Gln (mus musculus) Intracellular loop between TMD 7 and TMD 8	Likely pathogenic	-
	II.2 female	division of the symphysis				

S10 Table: Heterozygous Variants of SLC20A1 in BEEC families

Family 1 reported in Reutter et al. 2016. Bladder exstrophy epispadias complex (BEEC), Chromosome Position (Chr.Pos.), homozygous (hom), heterozygous (het), Genome Aggregation Database (gnomAD), transmembrane domain (TMD), American College of Medical Genetics (ACMG)