Gene	Exon	Nucleotide mutation	Allele state in the affected boys	Allele state in the mother	Allele state in the father	Protein alteration	Frequency	Prediction and Conservation
LRP1B (NM_018557) [MIM 608766]	20	c.3019T>C	heterozygous	wild-type	heterozygous	p.(Phe1007Leu)		Polyphen-2: predicted to be benign with a score of 0.000 (sensitivity: 1.00; specificity: 0.00) SIFT: Tolerated (score: 1, median: 4.32) MutationTaster: polymorphism (p-value: 0.987) UCSC: Leu found in some species including mouse, rat and dog
	31	c.5131G>A	heterozygous	heterozygous	wild-type	p.(Asp1711Asn)	rs567734617 ExAC: ALL:0.0035% - SAS:0.019% - NFE:0.0016%	Polyphen-2: predicted to be probably damaging with a score of 1.000 (sensitivity: 0.00; specificity: 1.00) SIFT: Deleterious (score: 0, median: 4.32) MutationTaster: disease causing (p-value: 1) UCSC: highly conserved
MUC22 (NM_001198815) [MIM 613917]	3	c.1162G>A	heterozygous	wild-type	heterozygous	p.(Val388Ile)	rs138073256 dbSNP: MAF/MinorAlleleCount: A=0.001/0 ExAC: ALL:0.04% - NFE:0.16%	SIFT: Tolerated (score: 0.53, median: 2.94) MutationTaster: polymorphism (p-value: 1) UCSC: Ile found in some species including rabbit, dog and elephant
	3	c.3292_3294del	heterozygous	heterozygous	wild-type	p.(Thr1098del)	rs550190545 ExAC: ALL:5.28% - AFR:1.04% - AMR:9.78% - EAS:29.10% - SAS:5.91% - NFE:2.49% - OTH:6.67%	
UTRN (NM_007124) [MIM 128240]	20	c.2654G>A	heterozygous	wild-type	heterozygous	p.(Arg885His)	rs34643516 dbSNP: MAF/MinorAlleleCount: A=0.002/0 EVS: Eur.Am:0.07% - Afr.Am:0.02% ExAC: ALL:0.15% - AFR:0.029% - AMR:0.029% - AMR:0.096% - EAS:0.88% - SAS:0.055% - NFE:0.12% - FIN:0.03% - OTH:0.44%	Polyphen-2: predicted to be benign with a score of 0.011 (sensitivity: 0.96; specificity: 0.78) SIFT: Tolerated (score: 0.55, median: 3.40) MutationTaster: polymorphism (p-value: 1) UCSC: His found in several species including mouse, rat and rabbit
	32	c.4445A>G	heterozygous	heterozygous	wild-type	p.(Tyr1482Cys)	ExAC: ALL:0.0017% - NFE:0.003%	Polyphen-2: predicted to be probably damaging with a score of 1.000 (sensitivity: 0.00; specificity: 1.00) SIFT: Deleterious (score: 0, median: 3.40) MutationTaster: disease causing (p-value: 1) UCSC: highly conserved
PDCD11 (NM_014976) [MIM 612333]	32	c.4829G>T	hemizygous	heterozygous	wild-type	p.(Arg1610Leu)	rs146747336 dbSNP: MAF/MinorAlleleCount: T=0.001/0 EVS: Eur.Am:0.12% - Afr.Am:0.02% ExAC: ALL:0.18% - AFR:0.019% - AMR:0.23% - SAS:0.19% - NFE:0.22% - FIN:0.03% - OTH:0.11%	Polyphen-2: predicted to be probably damaging with a score of 1.000 (sensitivity: 0.00; specificity: 1.00) SIFT: Deleterious (score: 0, median: 2.76) MutationTaster: disease causing (p-value: 1) UCSC: highly conserved
PABPC5 (NM_080832) [MIM 300407]	2	c.355A>T	hemizygous	heterozygous	wild-type	p.(Asn119Tyr)		Polyphen-2: predicted to be probably damaging with a score of 1.000 (sensitivity: 0.00; specificity: 1.00) SIFT: Deleterious (score: 0, median: 3.46) MutationTaster: disease causing (p-value: 1) UCSC: highly conserved
ZMAT1 (NM_001011657)	7	c.833G>A	hemizygous	heterozygous	wild-type	p.(Arg278Gln)	rs147524708 EVS: Efr.Am:0.03% ExAC: ALL:0.0091% - AFR:0.012% - AMR:0.011% - NFE:0.01% - OTH: 0.16%	Polyphen-2: predicted to be benign with a score of 0.003 (sensitivity: 0.98; specificity: 0.44) SIFT: Tolerated (score: 0.51, median: 3.02) MutationTaster: polymorphism (p-value: 1)

								UCSC: Gln found in several species including baboon, squirrel monkey and bushbaby
AMOT (NM_001113490) [MIM 300410]	10	c.2738_2764del	hemizygous	heterozygous	wild-type	p.(Val913_Pro921del)	EVS: Eur.Am:2.7% - Afr.Am:1.83% ExAC: ALL:2.56% - AFR:1.64% - AMR:3.41% - SAS:0.29% - NFE:4.41% - FIN:3.49% - OTH:0.63%	