Supplementary table 1: Rare, homozygous, predicted pathogenic exonic variants identified in patient 1 DNA.

chr	bp	ref	mut	gene	AA change
1	175129955	G	T	KIAA0040	N-65-K
2	48808095	C	A	STON1	S-108-*
3	136240051	T	C	STAG1	splice site
3	142987754	G	A	SLC9A9	P-558-L
4	120177605	C	T	USP53	R-166-C
11	12901385	C	G	TEAD1	P-154-R
11	20066981	G	A	NAV2	A-1159-T
12	14947485	T	C	WBP11	Y-236-C
15	65342421	T	-	SLC51B	F-27-F
16	175076	A	-	NPRL3	splice site
19	8564466	C	A	PRAM1	V-76-F
X	38013815	G	A	SRPX	L-351-F
X	50213149	G	A	DGKK	P-177-S
X	106070571	A	G	TBC1D8B	splice site
X	134707883	A	G	DDX26B	Y-509-C

Supplementary table 2: All *SLC51A* (OSTα) and *SLC51B* (OSTβ) variants identified in patient 1 DNA.

The sequences was compared to the following Ensembl Identifers for *SLC51A* (gene identifier: ENSG00000163959; transcript identifier: ENST00000296327; protein identifier: ENSP00000296327) and *SLC51B* (gene identifier: ENSG00000186198; transcript identifier: ENST00000334287; protein identifier: ENSP00000335292).

									Location				
Chromo-some	**Position**	**Gene**	**Reference**	**Allele**	**Zygosity**	**dbsnp**	**Effect**	**1kg_MAF**	**Exon**	**Intron**	**cDNA Sequence**	**Coding Sequence**	**Protein Sequence**
3	195941216	**SLC51A**	C	G	Hom	rs7642243	Upstream gene variant	0.3264 (C)					
3	195943481	**SLC51A**	C	T	Hom	rs56030157	5' prime UTR variant	0.2319 (T)	1 (of 9)		107		
3	195950608	**SLC51A**	T	A	Het	.	Intron variant			2 (of 8)			
15	65342420	**SLC51B**	GT	-	Hom	.	Frameshift variant truncation		2 (of 4)		400	79	27
15	65344035	**SLC51B**	G	A	Hom	rs2946676	Intron variant	0.0937 (G)		3 (of 3)			