SUPPLEMENTARY MATERIALS

Figure S1: Visualization of HTS results for control samples C1 and C2 by The Integrative Genomics Viewer (IGV) (A) Sample C1 showing a no-stop mutation in *MAGEB4* (B) Sample C2 showing a stop-gain mutation in *TEX15*

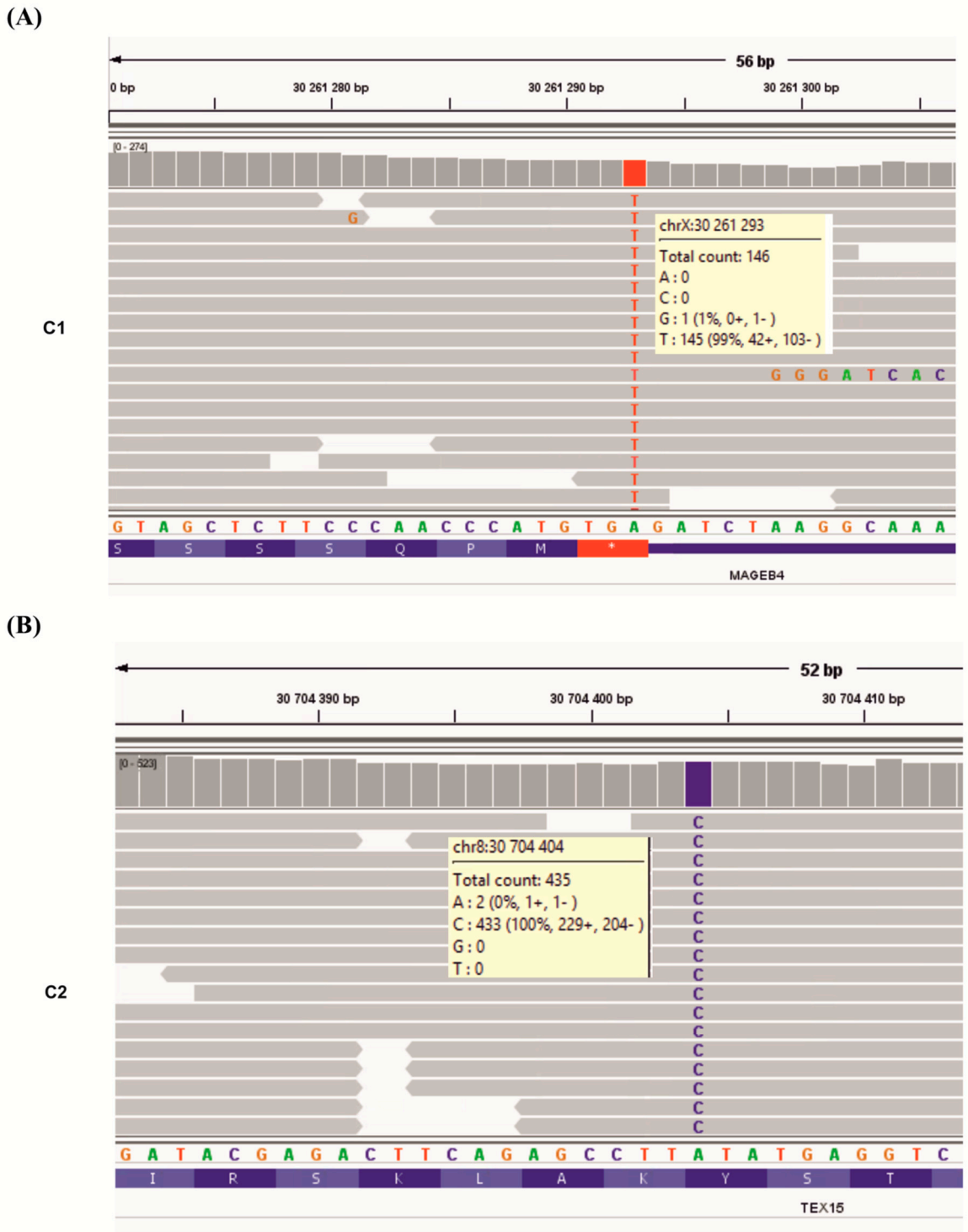
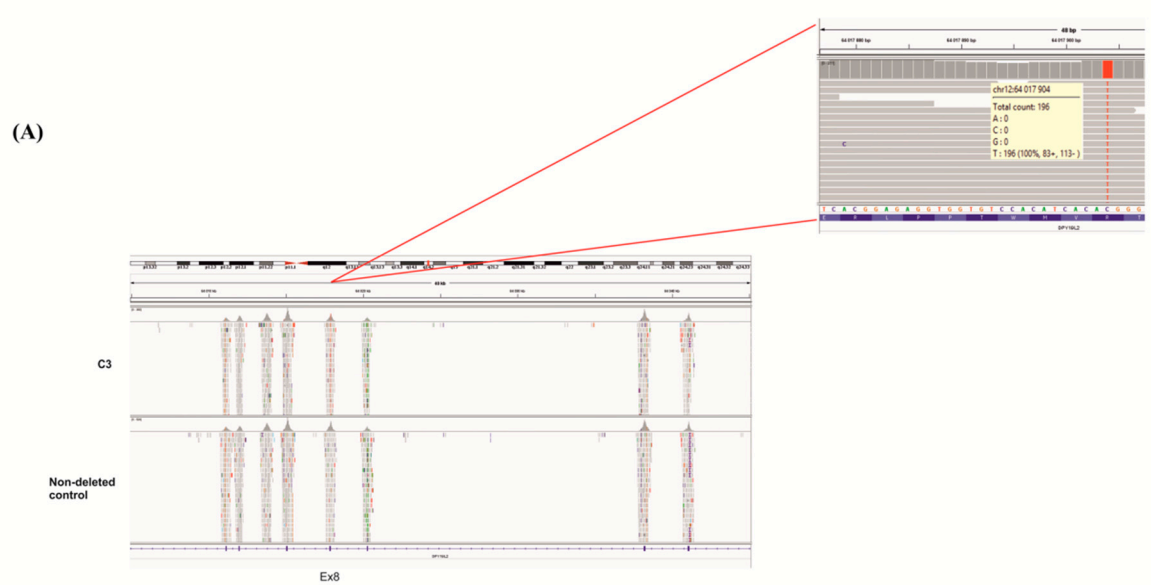


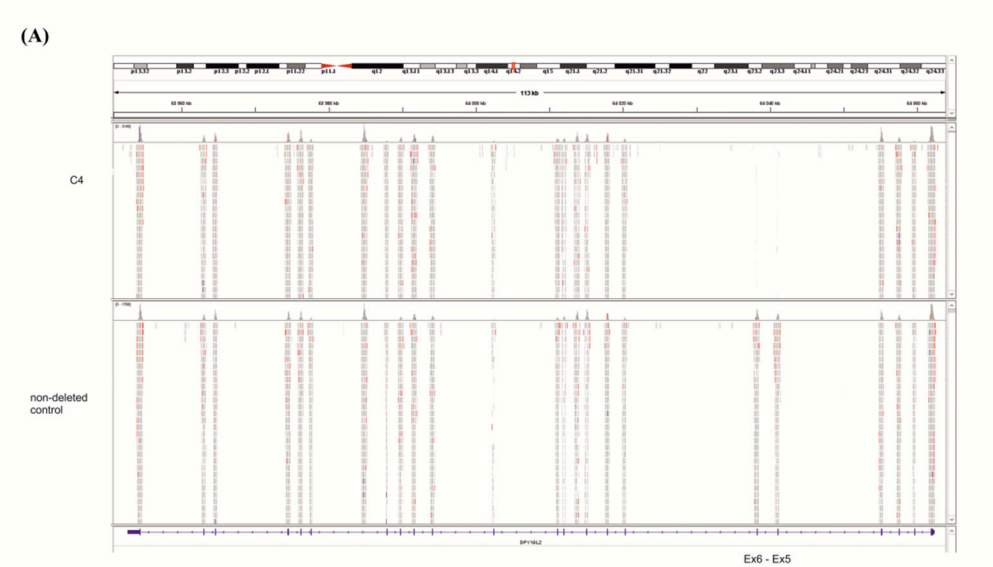
Figure S2: HTS data visualized with the IGV for control sample C3 showing heterozygous *DPY19L2* gene deletion with point mutation in exon 8 (A) *DPY19L2* gene sequencing data around exon 8 for sample C3 and a non-deleted sample for a control. There are 3149 reads in the non-deleted sample for the entire DPY19L2 gene in comparison to 1768 reads in sample C3, which is approximately half of the non-deleted samples indicating heterozygous *DPY19L2* gene deletion. Schematic representation of the mapped sequencing reads for the point mutation in exon 8 is shown in the enlarged area. (B) CANOES report which shows deletion in DPY19L2 for sample C3.



(B)

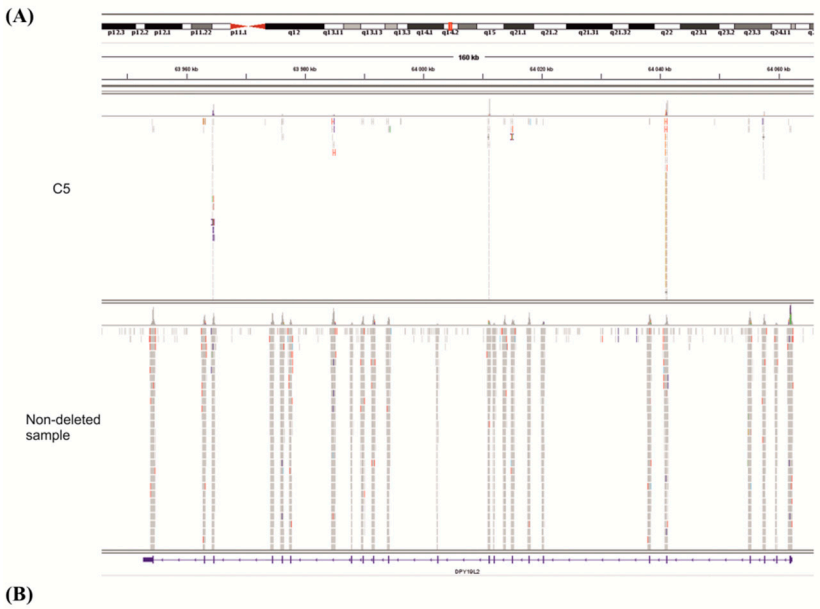
#Sex for sample C3 is: SEX_M									
#CNV chrom	CNV start	CNV end	CNV type	Sample ID	Gene name	NM	CDS length	tx lenth	location
12	63954266	64062198	DEL	C3	DPY19L2	NM_173812	2299	107933	exon1-exon22

Figure S3: Data visualization of HTS for control sample C4 (A) IGV plot showing the deletion of exon 5 and exon 6 of *DPY19L2* for sample C4, in comparison with non-deleted sample. (B) CANOES report also shows deletion of exon 5 and exon 6 in *DPY19L2* .



#Sex for sample C4 is: SEX_M									
#CNV chrom	CNV start	CNV end	CNV type	Sample ID	Gene name	NM	CDS length	tx lenth	location
12	64038157	64041170	DEL	C4	DPY19L2	NM_173812	217	3014	intron4-intron6

Figure S4: A whole *DPY19L2* deletion in control sample C5 (A) visualization of sequencing data of *DPY19L2* gene for sample C5 in comparison with non-deleted sample (B) CANOES report of C5 showing a whole *DPY19L2* gene deletion



#Sex for sample C5 is: SEX_M									
#CNV chrom	CNV start	CNV end	CNV type	Sample ID	Gene name	NM	CDS length	tx lenth	location
12	63954266	64062198	DEL	C5	DPY19L2	NM_173812	2299	107933	exon1-exon22

Table S1: Primers used for amplification of ten candidate variants selected in 8 genes after panel analysis. PCR reactions were carried out with Qiagen Multiplex PCR kit (QIAGEN GmbH, Hilden, Germany), 35 cycles and in total 25µl volume

Gene name	Exon	Forward primer	Reverse primer	Annealing Temperature (°C)	PCR size product (bp)
PATL2	6-7	tgtccatcagagctgacctg	tcccacctgaacagacatga	64	623
AURKC	3	gactttccctccgcctaccctac	gctgggctcagacgtcaaaga	60	301
CFAP43	28	caagtgggagcatctctggt	atgaggagggagggtattgg	65	489
HFM1	16	aaaatgttcctgaagcaacaaag	ttttctctgctatattacttttaagcc	58	327
GALNTL5	2	agggccatcaacgtgattgt	agctctctgaagcaaatatagcca	64	634
KLHL10	3	agcaattcaaaggcagatcg	ccggctttgcattactttc	60	906
DNAH1	38	cccacttacaggatgtgcag	tggattcctgcagggagtag	64	394
	60	cacccgaagtgggattagaa	ctcctaccaccctcttgctg	64	427
	62	aggccaacaaatggatcaag	tactgtggcctgagcatcac	64	361
TUBB8	4	gtgtgacgcttggctctttc	ttaaaacgcagcaggagatg	64	1264

Table S2: Sequencing quality for each gene from the Infertility Panel. Our diagnostic criteria require a sequencing depth of over 30 reads per base pair in at least 95% of all analyzed regions. 100% of coding exons were accurately sequenced in 40 genes; between 95 and 99.9% of coding exons were accurately sequenced in 11 genes

Gene name	RefSeq	Chromosome	#Coding Exon	%Coverage >30X
AKAP4	NM_003886.2	X	6	100
AURKC	NM_001015878.1	19	7	100
BMP15	NM_005448.2	X	2	100
BRDT	NM_001242806.2	1	18	100
CATSPER1	NM_053054.3	11	12	100
CCDC39	NM_181426.1	3	20	97.79
CFAP43	NM_025145.6	10	38	100
CFAP44	NM_001164496.1	3	34	97.73
DAX1(NROB1)	NM_000475.4	X	2	100
DNAH1	NM_015512.4	3	77	100
DNAH6	NM_001370.1	2	76	100
DPY19L2	NM_173812.4	12	22	100
FIGLA	NM_001004311.3	2	5	100
FMR1	NM_002024.5	X	17	100
FSHR	NM_000145.3	2	10	100
GALNTL5	NM_145292.3	7	8	100
GDF9	NM_005260.5	5	2	100
HFM1	NM_001017975.4	1	38	100
HIWI (PIWIL1)	NM_004764.4	12	20	100
HSF2	NM_004506.3	6	13	99.66
KLHL10	NM_152467.4	17	5	100
MAGEB4	NM_002367.3	X	1	100
MCM8	NM_001281521.1	20	18	99.13
MCM9	NM_017696.2	6	12	97.44
MEIOB	NM_001163560.2	16	13	95.57
MSH4	NM_002440.3	1	20	100
MTUS1	NM_001001924.2	8	14	95.59
NANOS1	NM_199461.3	10	1	100
NANOS3	NM_001098622.2	19	2	100
NOBOX	NM_001080413.3	7	10	100
NPAS2	NM_002518.3	2	20	100
NR5A1	NM_004959.4	9	6	100
PATL2	NM_001145112.1	15	15	100
PGRMC1	NM_006667.4	X	3	100
PICK1	NM_001039583.1	22	12	100
PLCZ1	NM_033123.3	12	14	100
SEPT12	NM_144605.4	16	9	100
SLC26A8	NM_001193476.1	6	19	100
SOHLH1	NM_001012415.2	9	7	97.92
SPAG17	NM_206996.3	1	48	100
SPATA16	NM_031955.5	3	10	100
SPINK2	NM_001271722.1	4	2	100
STAG3	NM_001282717.1	7	33	100
SYCE1	*NM_001143764.1*	10	13	100
TAF4B	NM_001293725.1	18	15	95.24
TEX11	NM_001003811.1	X	29	96.05
TEX14	NM_001201457.1	17	32	100
TEX15	NM_031271.3	8	4	100
TUBB8	NM_177987.2	10	4	100
Wt1	NM_024426.3	11	10	99.52
ZMYND15	NM_001267822.1	17	13	100

Table S3 : Sequencing quality and bioinformatics analysis information of the 94 patients. F :Female, M :Male, POI :Primary ovarian insufficiency, SPD:Sperm production defect, OOMD:oocyte maturation defect, OA: Oligoasthenozoospermia, AT:Asthenozoospermia, OT:Oligoteratozoospermia, OAT:Oligoasthenoteratozoospermia, CNV: copy number variations, DUP:Duplication, DEL: Deletions, Hom:homozygous, Het:heterozygous

Code	Sex	Clinical indication	Coverage (X)	30x(%)	Total #variants	# Filtered variations	# Variations related to phenotype	Mutation	Zygosity	Coding Effect	Gene name
Pt1	F	POI	652.41	99.9085	281	18	0	0			
Pt2	F	POI	585.05	99.9363	274	26	2	1	Hom	stop-gain	*HFM1*
Pt3	F	POI	652.45	99.9455	248	18	3	0			
Pt4	M	SPD	447.55	99.9663	330	20	5	0			
Pt5	M	SPD	652.1	99.993	293	20	7	0			
Pt6	M	SPD	493.2	99.9813	308	17	5	0			
Pt7	M	SPD	410.68	99.916	268	19	5	0			
Pt8	M	SPD	711.68	99.9101	314	25	9	0			
Pt9	M	SPD	695.67	99.9781	276	15	4	0			
Pt10	M	SPD	798.11	99.9791	300	24	5	0			
Pt11	M	SPD	685.53	99.9631	285	26	8	0			
Pt12	M	SPD	563.27	99.9524	315	22	10	1	Het	missense	*KLHL10*
Pt13	M	SPD	643.01	99.9936	227	20	3	0			
Pt14	M	SPD	623.99	99.9663	249	18	6	0			
Pt15	M	SPD	756.82	99.993	275	20	6	0			
Pt16	M	SPD	379.8	99.9048	297	16	5	0			
Pt17	M	SPD	633.17	99.992	303	21	6	0			
Pt18	M	SPD	772.57	99.9294	295	15	4	0			
Pt19	M	SPD	804.98	99.992	334	27	8	0			
Pt20	M	SPD	868.19	99.9807	320	25	6	0			
Pt21	M	SPD	905.33	99.9845	276	14	3	0			
Pt22	M	SPD	861.61	99.9631	314	23	7	0			

Pt23	M	SPD	689.16	99.9133	287	20	8	0			
Pt24	M	SPD	922.67	99.993	241	13	3	0			
Pt25	M	SPD	781.31	99.9914	282	20	5	0			
Pt26	M	SPD	852.86	99.9936	291	25	7	0			
Pt27	M	SPD	683.43	99.9679	259	24	7	0			
Pt28	M	SPD	782.95	100	283	30	8	0			
Pt29	M	SPD	799.33	99.9759	306	31	4	0			
Pt30	M	SPD	650.32	99.8936	297	26	9	0			
Pt31	F	POI	369.24	99.2966	290	13	1	0			
Pt32	F	POI	361.34	99.403	310	17	2	0			
Pt33	F	POI	283.58	99.7127	365	40	3	0			
Pt34	F	POI	264.95	99.6742	333	29	7	0			
Pt35	F	POI	308.89	99.8952	269	19	5	0			
Pt36	F	POI	393.42	99.9406	298	21	5	0			
Pt37	F	OOMD	280.39	99.5475	318	20	0	0			
Pt38	F	OOMD	258.23	99.2335	266	19	1	1	Hom	missense	*TUBB8*
Pt39	F	OOMD	346.39	99.9626	332	19	3	0			
Pt40	M	Teratozoospermia	305.08	99.6528	304	26	8	0			
Pt41	M	Teratozoospermia	402.61	99.9497	272	27	10	1	Hom	frameshift	*AURKC*
Pt42	M	SPD	264.27	99.7165	311	26	15	0			
Pt43	M	SPD	218.54	99.4506	272	22	9	0			
Pt44	M	SPD	252.03	99.6058	280	19	6	0			
Pt45	M	SPD	208.21	99.7577	319	15	9	0			
Pt46	M	SPD	295.59	99.877	272	26	12	0			
Pt47	M	SPD	294.12	99.8219	325	28	6	0			
Pt48	M	SPD	329.02	99.587	332	23	10	0			
Pt49	M	SPD	262.25	99.6058	270	20	8	0			

Pt50	M	SPD	323.52	99.8379	302	30	14	0			
Pt51	M	SPD	237.59	99.7058	329	30	12	0			
Pt52	M	Asthenozoospermia	183.91	98.6755	307	23	2	0			
Pt53	M	SPD	286.69	99.8235	329	40	18	0			
Pt54	M	SPD	324.74	99.8155	269	31	6	0			
Pt55	M	Teratozoospermia	300.88	99.8481	298	27	10	1	Hom	splice-site	*CFAP43*
Pt56	M	AT	323.38	99.8245	297	23	13	0			
Pt57	M	Teratozoospermia	350.41	99.8171	257	13	5	0			
Pt58	M	Teratozoospermia	358.86	99.8978	306	25	5	0			
Pt59	M	AT	420	99.9497	310	21	8	0			
Pt60	M	OAT	317.91	99.9315	312	20	14	0			
Pt61	F	POI	419.89	99.9187	287	16	1	0			
Pt62	M	SPD	324.78	99.4121	284	17	6	0			
Pt63	M	SPD	416.1	99.8064	255	12	7	0			
Pt64	M	SPD	275.2	99.6475	269	21	10	0			
Pt65	M	AT	378.91	99.9636	283	21	8	3	Het Het Het	stop-gain frameshift missense	*DNAH1*
Pt66	M	OAT	337.08	99.9321	286	28	26	0			
Pt67	F	POI	402.92	99.9733	269	18	3	0			
Pt68	M	Teratozoospermia	424.96	99.9797	289	24	7	0			
Pt69	M	OT	448.57	99.7368	277	29	22	0			
Pt70	M	SPD	439.7	99.985	264	20	11	0			
Pt71	F	OOMD	388.86	99.7641	310	18	3	1	Hom	stop-gain	*PATL2*
Pt72	M	OAT	330.95	99.7919	244	12	6	0			
Pt73	M	Teratozoospermia	379.2	99.9872	250	26	4	0			
Pt74	M	OAT	337.81	99.8278	279	19	14	0			

Pt75	M	OAT	341.86	99.839	346	27	14	0			
Pt76	M	OAT	301.18	99.7801	281	16	13	0			
Pt77	M	OAT	369.18	99.9716	267	26	19	1	Het	frameshift	*GALNTL5*
Pt78	M	OAT	473.53	99.9711	297	33	21	0			
Pt79	M	SPD	403.85	99.9449	292	14	8	0			
Pt80	M	SPD	531.58	99.9412	254	23	10	0			
Pt81	M	SPD	425.04	99.9438	291	28	15	0			
Pt82	M	SPD	325.35	99.8871	263	20	7	0			
Pt83	M	SPD	383.32	99.9551	276	22	12	0			
Pt84	M	OA	444.95	99.908	304	16	11	0			
Pt85	M	SPD	541.4	99.9106	320	31	11	0			
Pt86	M	SPD	451.98	99.8026	269	12	7	0			
Pt87	M	OAT	443.05	99.8754	268	25	18	0			
Pt88	M	OAT	420.11	99.9128	355	34	28	0			
Pt89	M	OAT	365.09	99.7892	275	19	15	0			
Pt90	M	OAT	502.81	99.9337	309	26	19	0			
Pt91	M	OAT	468.43	99.9604	299	18	12	0			
Pt92	M	OAT	487.56	99.9139	374	30	25	0			
Pt93	M	OAT	528.62	99.8973	361	40	28	0			
Pt94	M	OAT	455.93	99.9005	272	15	10	0			

Table S4: Previously identified mutations described in the literature for the 8 genes that we determined as mutated in this study, with corresponding zygosity and relevant references. Heterozygous mutations that exists with another heterozygous mutation in patients indicated by using asterisk(*)

AURKC : NM_001015878.1

	c.DNA change	Protein change	Zygosity	References
1	c.144delC	p.Leu49Trpfs*23	Homozygous Heterozygous*	Dieterich et al 2007, Harbuz et al 2009 Dieterich et al 2009, Ben Khelifa et al 2011
2	c.269G>A	p.Arg90Gln	Homozygous	Hua and Wan 2018
3	c.382C>T	p.Leu128Phe	Heterozygous*	Bai et al 2020
4	c.436-2A>G	p. ?	Heterozygous*	Ben Khelifa et al 2011
5	c.572C>T	p. Thr191Ile	Heterozygous*	Bai et al 2020
6	c.686G>A	p.Cys229Tyr	Heterozygous*	Harbuz et al 2009, Dieterich et al 2009
7	c.744C>G	p.Tyr248*	Homozygous	Ben Khelifa et al 2012, Ortega et al 2019

CFAP43 : NM_025145.6

	c.DNA change	Protein change	Zygosity	References
1	c.114_117delCACC	p.Asn38Lysfs*10	Heterozygous*	Coutton et al 2019
2	c.120_121delTT	p.Ile40Metfs*12	Heterozygous*	Coutton et al 2019
3	c.253C>T	p.Arg85Trp	Heterozygous*	Tang et al 2017
4	c.386 C>A	p.Ser129Tyr	Heterozygous*	Tang et al 2017
5	c.739A>T	p.Lys247*	Homozygous	Wu et al 2019
6	c.1040T>C	p.Val347Ala	Heterozygous*	Coutton et al 2019
7	c.1140_1143delTAAA	p.Asn380Lysfs*3	Homozygous	Wu et al 2019
8	c.1240_1241delGT	p.Val414Leufs*46	Homozygous	Coutton et al 2019
9	c.1302dupT	p.Leu435Serfs*26	Heterozygous*	Coutton et al 2019
10	c.1474G>C	p.Gln492Arg	Homozygous	Wu et al 2019
11	c.2141+5G>A	p. ?	Homozygous	Coutton et al 2019
12	c.2658G>A	p.Trp886*	Homozygous	Coutton et al 2019
13	c.2680C>T	p.Arg894*	Homozygous	Coutton et al 2019
14	c.2802T>A	p.Cys934*	Heterozygous**	Tang et al 2017
15	c.3352C>T	p.Arg1118*	Homozygous	Coutton et al 2019
16	c.3374delG	p.Gly1125Glufs*12	Homozygous	Coutton et al 2019
17	c.3541-2A>C	p.Ser1181Lysfs*4	Homozygous	Coutton et al 2019
18	c.3661-2delA	p.Glu1221_Lys1256del	Homozygous Homozygous	Li et al 2020 Sha et al 2019
19	c.3882delA	p.Glu1294Aspfs*47	Homozygous	Coutton et al 2019
20	c.3945_4431del	DEL ex32-34 (p.Ile1316Leufs*10)	Heterozygous*	Tang et al 2017
21	c.4132C>T	p.Arg1378*	Heterozygous*	Tang et al 2017
22	c.4600C>G	p.Leu1534Val	Homozygous	Wu et al 2019

GALNTL5 : NM_145292.3

	c.DNA change	Protein change	Zygosity	References
1	c.369-2A>G	p. ?	Heterozygous	Hagiuda et al 2019*
2	c.764delT	p.Leu255Argfs*2	Heterozygous	Takasaki et al 2014

*Identified in sperm

KLHL10 : NM_152467.4

	c.DNA change	Protein change	Zygosity	References
1	c.242A>T	p.Asn81Ile	Heterozygous Heterozygous	Araujo et al 2019 Rocco et al 2020
2	c.647A>C	p.Gln216Pro	Heterozygous	Yatsenko et al 2006
3	c.887T>C	p.Ile296Thr	Heterozygous Heterozygous	Araujo et al 2019 Rocca et al 2019
4	c.937G>A	p.Ala313Thr	Heterozygous	Yatsenko et al 2006
5	c.1302+123_1302+126delTCTT	p. ?	Heterozygous	Yatsenko et al 2006

DNAH1 : NM_015512.4

	c.DNA change	Protein change	Zygosity	References
1	c.2127dupC	p.Ile710Hisfs*4	Homozygous	Coutton et al 2018
2	c.2610G>A	p.Trp870*	Heterozygous *	Tang et al 2017, Sha et al 2017, Liu et al 2019
3	c.3108G>A	p.Trp1036*	Heterozygous*	Tang et al 2017, Sha et al 2017, Liu et al 2019
4	c.3593_3593delA	p.Asp1198Alafs*6	Heterozygous*	Tang et al 2017
5	c.3836A>G	p.Lys1279Arg	Heterozygous*	Sha et al 2017, Liu et al 2019
6	c.3846C>G	p.Tyr1282*	Homozygous	Liu et al 2019
7	c.3860T>G	p.Val1287Gly	Homozygous	Amiri-Yekta et al 2016
8	c.3877G>A	p.Asp1293Asn	Homozygous	Ben Khalifa et al 2014
9	c.3931C>T	p.Gln1311*	Homozygous	Coutton et al 2019
10	c.4115C>T	p.Thr1372Met	Heterozygous*	Tang et al 2017, Sha et al 2017, Liu et al 2019
11	c.4531G>A	p.Val1511Met	Heterozygous	Coutton et al 2018
12	c.4642C>G	p.Leu1548Val	Heterozygous	Coutton et al 2018
13	c.4744_4752delCCAGCTGGC	p.Pro1582_Gly1584del	Homozygous	Coutton et al 2018
14	c.4983G>A	p.Val1661=	Homozygous	Coutton et al 2019
15	c.5094+1G>A	NA	Homozygous	Ben Khalifa et al 2014
16	c.5104A>T	p.Arg1702*	Heterozygous*	Tang et al 2017
17	c.5105G>C	p.Arg1702Pro	Heterozygous *	Tu et al 2019
18	c.5094+1G>A	NA	Homozygous	Ben Khalifa et al 2014
19	c.5584G>A	p.Gly1862Arg	Heterozygous*	Tang et al 2017
20	c.5626G>C	p.Ala1876Pro	Heterozygous *	Yang et al 2018, Liu et al 2019
21	c.5766-2A>G	NA	Heterozygous*	Sha et al 2017, Liu et al 2019
22	c.5864G>A	p.Trp1955*	Heterozygous*	Tang et al 2017, Sha et al 2017, Liu et al 2019
23	c.6212T>G	p.Leu2071Arg	Heterozygous*	Tang et al 2017, Sha et al 2017, Liu et al 2019
24	c.6253_6254del	p.Arg2085fs	Heterozygous*	Sha et al 2017, Liu et al 2019
25	c.6257_6258delAG	p.Glu2086Glyfs*8	Heterozygous*	Tang et al 2017
26	c.6448G>A	p.Val2150Met	Heterozygous*	Tang et al 2017
27	c.6822C>G	p.Asp2274Glu	Heterozygous*	Sha et al 2017, Liu et al 2019
28	c.7066C>T	p.Arg2356Trp	Heterozygous	Sha et al 2017, Yang et al 2018
29	c.7153T>A	p.Trp2385Arg	Heterozygous *	Coutton et al 2018
30	c.7377+1G>C	NA	Homozygous	Sha et al 2017, Liu et al 2019
31	c.7378+1G>C	NA	Homozygous	Tang et al 2017
32	c.7396C>T	p.Arg2466*	Heterozygous*	Tang et al 2017
33	c.7471C>A	p.Gln2491Lys	Heterozygous *	Li et al 2019
34	c.7531delC	p.Gln2511Serfs*27	Homozygous	Coutton et al 2018
35	c.7864C>T	p.Arg2622*	Heterozygous*	Tang et al 2017
36	c.8151-1G>C	NA	Homozygous Heterozygous*	Liu et al 2019 Tu et al 2019
37	c.8322+3del	NA	Heterozygous*	Yang et al 2018
38	c.8626-1G>A	NA	Homozygous	Amiri-Yekta et al 2016, Coutton et al 2019

39	c.8811+1G>T	NA	Heterozygous*	Tang et al 2017
40	c.9505C>G	p.Arg3169Gly	Heterozygous*	Coutton et al 2018, Liu et al 2019
41	c.9567delC	p.Glu3190Serfs*4	Homozygous	Coutton et al 2019
42	c.9621+1G>C	NA	Homozygous	Coutton et al 2019
43	c.9661G>A	p.Gly3221Arg	Heterozygous*	Tang et al 2017
44	c.9685C>T	p.Arg3229Cys	Heterozygous*	Tang et al 2017
45	c.9850G>A	p.Glu3284Lys	Heterozygous*	Sha et al 2017, Liu et al 2019
46	c.10464_10467delAGAG	p.Arg3490Glnfs*4	Heterozygous*	Tang et al 2017
47	c.10630G>T	p.Glu3544*	Heterozygous*	Sha et al 2017, Liu et al 2019
48	c.11213G>A	p.Trp3738*	Heterozygous*	Tang et al 2017
49	c.11726_11727delCT	p.Pro3909Argfs*33	Homozygous Heterozygous *	Wang et al 2017 Yang et al 2018, Tang et al 2017, Sha et al 2017, Liu et al 2019, Tu et al 2019
50	c.11779G>A	p.Asp3927Asn	Heterozygous *	Liu et al 2019
51	c.11787_11787+1insT	NA	Heterozygous*	Tang et al 2017
52	c.11788-1G>A	NA	Homozygous	Ben Khalifa et al 2014
53	c.12200_12202del	p.4067_4068del	Heterozygous *	Sha et al 2017, Liu et al 2019
54	c.12200_12202delACA	p.Asn4068del	Heterozygous*	Tang et al 2017
55	c.12286C>T	p.Arg4096Cys	Heterozygous *	Tu et al 2019
56	c.12287G>T	p.Arg4096Leu	Heterozygous*	Tang et al 2017, Sha et al 2017, Liu et al 2019
57	c.12397C>T	p.Arg4133Cys	Heterozygous*	Tang et al 2017, Sha et al 2017, Liu et al 2019
58	c.12520G>A	p.Ala4174Thr	Homozygous	Tang et al 2017
59	c.12796T>C	p.Ter4266Glnext*21	Homozygous	Ben Khalifa et al 2014

PATL2 : NM_001145112.1

	c.DNA change	**Protein change**	**Zygosity**	**References**
1	c.223-14_223-2del13	p.Arg75Valfs*21	Heterozygous *	Chen et al 2017a, Huang 2018, Wu 2019
2	c.478C>T	p.Arg160*	Homozygous	Maddirevula et al 2017, Christou-Kent et al 2018
3	c.558T>A	p.Tyr186*	Heterozygous*	Chen et al 2017a
4	c.566T>G	p.Leu189Arg	Heterozygous*	Chen et al 2017a
5	c.649T>A	p.Tyr217Asn	Heterozygous*	Chen et al 2017a
6	c.716delA	p.Asn239Thrfs*9	Heterozygous*	Wu et al 2019
7	c.778G>A	p.Val260Met	Heterozygous*	Wu et al 2019
8	c.784C>T	p.Arg262*	Homozygous	Chen et al 2017a
9	c.839G>A	p.Arg280Gln	Heterozygous*	Chen et al 2017a
10	c.865delA	p.Thr289Leufs*6	Heterozygous*	Huang et al 2018
11	c.877G>T	p.Asp293Tyr	Heterozygous*	Wu et al 2019
12	c.898C>T	p.Gln300*	Heterozygous*	Wu et al 2019
13	c.953T>C	p.Ile318Thr	Heterozygous*	Chen et al 2017a
14	c.1108G>A	p.Gly370Arg	Homozygous	Maddirevula et al 2017
15	c.1127G>A	p.Arg376Gln	Homozygous	Huang et al 2018
16	c.1225-2A>G	p. ?	Heterozygous*	Huang et al 2018
17	c.1225+2T>C	p. ?	Heterozygous*	Chen et al 2017a
18	c.1273A>C	p.Thr425Pro	Heterozygous*	Wu et al 2019
19	c.1282G>T	p.Glu428*	Heterozygous*	Huang et al 2018
20	c.1300C>T	p.Gln434*	Heterozygous*	Huang et al 2018
21	c.1376C>A	p.Ser459Tyr	Homozygous	Liu et al 2020
22	c.1528C>A	p.Pro510Thr	Homozygous	Liu et al 2020

HFM1 :NM_001017975.4

	c.DNA change	Protein change	Zygosity	References
1	c.1006 + 1G > T	p. ?	Heterozygous*	Liu et al 2020
2	c.1241A>C	p.His414Pro	Heterozygous	Pu et al 2016
3	c.1686-1G>C	p. ?	Heterozygous* Heterozygous	Wang et al 2014 Pu et al 2016
4	c.2206G>A	p.Gly736Ser	Heterozygous*	Wang et al 2014
5	c.2325C>A	p.Phe775Leu	Heterozygous	Pu et al 2016
6	c.2651T>G	p.Ile884Ser	Heterozygous*	Wang et al 2014
7	c.3100G > A	p.Gly1034Ser	Heterozygous*	Liu et al 2020
8	c.3470G>A	p.Cys1157Tyr	Heterozygous	Zhe et al 2019
9	c.3580C>T	p.Arg1194Cys	Heterozygous	Pu et al 2016
10	c.3929_3930delinsG	p.Pro1310Argfs*41	Heterozygous*	Wang et al 2014
11	c.4061delC	p.Pro1354Leufs*71	No info	Yanus et al 2019

TUBB8 : NM_177987.2

	c.DNA change	Protein change	Zygosity	References
1	c.5G>A	p.Arg2Lys	Heterozygous	Feng et al 2016a
2	c.5G>T	p.Arg2Met	Heterozygous	Huang et al 2017,Chen et al 2017b
3	c.10A>C	p.Ile4Leu	Heterozygous	Chen et al 2017b, Chen et al 2019
4	c.12C>G	p.Ile4Met	Heterozygous	Zhao et al 2020
5	c.35G>A	P.Cys12Tyr	Homozygous	Chen et al 2017b
6	c.107G>A	p.Gly17Ser	Heterozygous	Zhao et al 2020
7	c.80_100del21	p.Glu27_Ala33del	Homozygous Heterozygous*	Feng et al 2016b Zhao et al 2020
8	c.140C>T	p.His28Tyr	Heterozygous*	Zhao et al 2020
9		p.Arg46Cys		Yang et al 2021
10	c.206T>C	p.Tyr50His	Heterozygous	Zhao et al 2020
11	c.148_154delins CACCACCACGAGGCC AGCGGTGCGACCCCC GTCCTTCCCCCACCCA ACGTGCACCACC	p.Tyr50_Asn52delins HHHESGATPVLPPP NVHHH	Heterozygous*	Zhao et al 2020
12	c.161C>T	p. A54V	Homozygous	Xing et al 2020
13	c.178G>C	p.Val60Leu	Heterozygous	Zhao et al 2020
14	c.181C>A	p.Pro61Thr	Heterozygous	Chen et al 2019
15	c.209C>T	p.Pro70Leu	Heterozygous	Chen et al 2017b
16	c.292G>A	p.Gly79Glu	Heterozygous	Zhao et al 2020
17	c.305C>T	p.Pro87Leu	Heterozygous	Zhao et al 2020
18	c.292G>A	p.Gly98Arg	Heterozygous	Wang et al 2018, Chen et al 2019
19	c.351G>A	p.Gly98Glu	Heterozygous	Zhao et al 2020
20	c.322G>A	p.Glu108Lys	Homozygous Heterozygous *	Yuan et al 2018 Chen et al 2019
21	c.411A>G	p.Asp118Gly	Heterozygous*	Zhao et al 2020
22	c.363_366delAAAG	p.Lys122Argfs*13	Heterozygous	Christou-Kent et al 2018
23	c.367G>A	p.Glu123Lys	Heterozygous	Yang et al 2021
24	c.382dup	p.Asp128Glyfs*27	Heterozygous*	Zhao et al 2020
25	c.394G>T	p.Gly132Cys	Heterozygous	Yang et al 2021
26	c.456T>C	p.Phe133Ser	Heterozygous*	Zhao et al 2020
27	c.458C>T	p.Gln134*	Heterozygous*	Zhao et al 2020
28	c.422G>C	p.Gly141Ala	Heterozygous	Yang et al 2021
29	c.426dupG	p.Thr143Aspfs*12	Homozygous Heterozygous *	Feng et al 2016b Chen et al 2019

30	c.552C>T	p.Thr166Ile	Heterozygous	Zhao et al 2020
31	c.523G>A	p.Val175Met	Heterozygous	Chen et al 2019
32	c.527C>T	p.Ser176Leu	Heterozygous	Feng et al 2016a ; Feng et al 2016b, Chen et al 2019
33	c.535G>A	p.Val179Met	Heterozygous	Huang et al 2017
34	c.593G>T	p.Val179Leu	Heterozygous	Zhao et al 2020
35	c.539T>C	p.Val180Ala	Heterozygous	Yang et al 2021
36	c.544C>T	p.Pro182Ser	Heterozygous	Yang et al 2021
37	c.550A>G	p.Asn184Asp	Heterozygous	Yang et al 2021
38	c.580G>A	p.Glu194Lys	Heterozygous*	Chen et al 2017b
39	c.594G>C	p.Glu198Asp	Heterozygous	Yang et al 2021
40	c.600T>G	p.Phe200Leu	Heterozygous	Chen et al 2019
41	c.608A>G	p.Asp203Gly	Heterozygous	Zhao et al 2020
42	c.613G>A	p.Glu205Lys	Heterozygous	Chen et al 2017b
43	c.628A>G	p.Ile210Val	Heterozygous	Feng et al 2016b
44	c.629T>A	p.Ile210Lys	Heterozygous	Yang et al 2021
45	c.720C>T	p.Thr221Ile	Heterozygous	Zhao et al 2020
46	c.686T>C	p.Val229Ala	Heterozygous	Feng et al 2016a
47	c.713C>T	p.Thr238Met	Heterozygous	Feng et al 2016b
48	c.715T>A	p.Cys239Ser	Heterozygous	Zhao et al 2020
49	c.721C>T	p.Arg241Cys	Homozygous	Chen et al 2019
50	c.722G>A	p.Arg241His	Heterozygous	Chen et al 2019
51	c.728C>T	p.Pro243Leu	Heterozygous	Zhao et al 2020
52	c.728C>G	p.Pro243Arg	Heterozygous	Zhao et al 2020
53	c.735G>C	p.Gln245His	Heterozygous	Chen et al 2019
54	c.736C>G	p.Leu246Val	Heterozygous	Zhao et al 2020
55	c.743C>T	p.Ala248Val	Heterozygous	Zhao et al 2020
56	c.763G>A	p.Val255Met	Heterozygous	Feng et al 2016b, Chen et al 2019
57	c.784C>T	p.Arg262Trp	Heterozygous	Feng et al 2016b
58	c.785G>A	p.Arg262Gln	Heterozygous	Feng et al 2016a
59	c.845G>C	p.Arg282Pro	Heterozygous	Zhao et al 2020
60	c.853A>C	p.Thr285Pro	Heterozygous	Feng et al 2016b
61	c.944G>C	p.Asp296His	Heterozygous	Chen et al 2019
62	c.950A>G	p.Asn298Ser	Heterozygous	Zhao et al 2020
63	c.958G>A	p.Met300Ile	Heterozygous	Feng et al 2016a
64	c.925C>T	p.Arg309Cys	Heterozygous	Zhao et al 2020
65	c.940G>T	p.Ala314Ser	Heterozygous	Zhao et al 2020
66	c.990G>A	p.Met330Ile	Heterozygous	Chen et al 2017b
67	c.1000C>G	p.Gln334Glu	Heterozygous	Chen et al 2019
68	c.1043A>G	p.Asn348Ser	Heterozygous	Feng et al 2016b
69	c.1054G>T	p.Ala352Ser	Heterozygous	Xiang et al 2018
70	c.1057G>A	p.Val353Ile	Heterozygous	Chen et al 2017b ; Chen et al 2019
71	c.1061G>A	p.Cys354Tyr	Heterozygous	Chen et al 2019
72	c.1072C>G	p.Pro358Ala	Heterozygous	Chen et al 2019
73	c.1073C>T	p.Pro358Leu	Heterozygous	Chen et al 2019
74	c.1076G>A	p.Arg359Gln	Homozygous	Zhao et al 2020
75	c.1088T>C	p.Met363Thr	Heterozygous	Feng et al 2016a
76	c.1099T>C	p.Phe367Leu	Heterozygous	Chen et al 2019
77	c.1103T>C	p.Ile368Thr	Heterozygous*	Zhao et al 2020
78	c.1130T>C	p.Leu377Pro	Heterozygous	Zhao et al 2020
79	c.1139G>A	p.Arg380His	Heterozygous	Zhao et al 2020
80	c.1163T>C	p.Met388Thr	Heterozygous	Zhao et al 2020
81	c.1171C>T	p.Arg391Cys	Heterozygous	Chen et al 2019
82	c.1172G>A	p.Arg391His	Heterozygous	Zhao et al 2020
83	c.1178C>A	p.Ala393Asp	Heterozygous	Yang et al 2021

84	c.1187A>G	p.His396Arg	Heterozygous	Zhao et al 2020
85	c.1189T>G	p.Trp397Glu	Heterozygous	Yang et al 2021
86	c.1203_1204insCT	p.G402Lfs*15	Heterozygous	Yang et al 2021
87	c.1205dupG	p.Met403Hisfs*3	Homozygous	Chen et al 2019
88	c.1228G>A	p.Glu410Lys	Heterozygous	Chen et al 2019
89	c.1242C>G	p.Asn414Lys	Heterozygous	Yang et al 2021
90	c.1245G>A	p.Met415Ile	Heterozygous*	Chen et al 2017b
91	c.1249G>T	p.Asp417Tyr	Heterozygous	Chen et al 2019
92	c.1249G>A	p.Asp417Asn	Heterozygous	Feng et al 2016a
93	c.1270C>T	p.Gln424*	Homozygous	Chen et al 2019
94	c.1271A>G	p.Gln424Arg	Heterozygous	Yang et al 2021
95	c.736C>G	p.Leu426Val	Heterozygous	Zhao et al 2020
96	c.1286C>T	p.Thr429Met	Heterozygous*	Chen et al 2019
97	c.1301_1327del	p.E434_E442del	Heterozygous*	Chen et al 2019
98	ND	Exon1-4 del	Homozygous	Chen et al 2017b

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