GENE	GENE NAME	OMIM	MOI	OMIM DISEASE NAME	REASON FOR IN/EXCLUDING THE VARIANT AS PATHOGENIC	ENSEMBL TRANSCRIPT	CHR	POS [HG19]	VARIANT	AAE	COVERAGE [fold]	VARIANT FREQUENCY [%]	dbSNP	MAJOR ALLELE HOMO *	HET *	MINOR ALLELE HOMO *	ExAC SERVER: VARIANTS PER ALLELES (TOTAL)	gnomAD SERVER: VARIANTS PER ALLELES (TOTAL)
SLC25A4	solute carrier family 25 member 4	#615418	AR	Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR	**confirmed:** missense mutation of a conserved amino acid, known disease association with mutations in *SLC25A4,* new mutation, clinical phenotype of the patient corresponds to known clinical features.	ENST00000281456	4	186.066.967	A>C	Q218P	85	99					121.190 / 0	277.200 / 0
FAT1	FAT tumor suppressor homolog 1 *(Drosophila)*	*600976	AR AD		**excluded:** 238 heterozygous and 1 homozygous variants in ExAC. **NO** increased tumor growth in our patient as seen in homozygous loss-of function mutations in *Drosophila* and for spontaneous mutations in human glioblastoma multiforme brain tumors.	ENST00000441802	4	187.557.962	T>C	Y1250C	254	99	rs142805532	2496	8	0	120.738 / 240	277.168 / 546
NIPBL	Nipped-B homolog (*Drosophila*)	#122470	AR	Cornelia de Lange syndrome type 1	**excluded:** the phenotype does not fit with the disease description in OMIM. **NO** characteristic facial dysmorphism, no prenatal growth retardation, or upper limb anomalies.	ENST00000282516	5	37.058.994	C>T	S2471F	126	99					120.774 / 0	243.406 / 0
DCDC5	doublecortin domain containing 5	*612321			**excluded:** 532 heterozygous and 18 homozygous variants in ExAC. **NO** brain malformation as could be expected for muations in proteins similar to doublecortin.	ENST00000339794	11	30.926.525	C>T	A510T	124	99	rs61747656	2435	66	3	121.282 / 568	274.498 / 1396
DNAJC24	DnaJ (Hsp40) homolog, subfamily C, member 24	*611072	AR		**excluded:** homozygous knock-out mice show increased embryonic lethality, decreased embryonic size, and polydactyly. **NO** such dysmorphic features seen in our patient	ENST00000465995	11	31.436.491	G>A	R82Q	67	100					110.560 / 3	244.280 / 4
ARHGAP1	Rho GPTase activating protein 1	*602732			**excluded:** 889 heterozygous and 10 homozygous variants in ExAC	ENST00000311956	11	46.702.070	G>A	L263F	85	99	rs144801476	2474	29	1	120.888 / 909	277.142 / 2041
EML3	echinoderm microtubule associated protein like 3				**excluded:** *EML3* is a nuclear microtubule-binding protein required for the correct alignment of chromosomes in the metaphase. **NO** brain malformation or band heterotopia nor chromosomal rearrange-ments found in our patient as seen for mutations in other homologs in the EML family.	ENST00000529309	11	62.369.890	A>AAGGGC	S880Afs*18	150	99	rs573593516	2501	3	0	30.095 / 49	179.480 / 361
USP3	ubiquitin specific peptidase 3	*604728			**excluded:** deletion of *USP3* in mice increases the incidence of spontaneous tumors and affects hematopoiesis. **NO** oncological or hematological symptoms present in our patient	ENST00000380324	15	63.881.000	T>C	C439R	116	98					32.767 / 4	246.128 / 7

Supplemental table 1: Homozygous variants in the autozygous region that cause an amino acid exchange and were predicted to be potentially disease causing by the MutationTaster software. MOI, mode of inheritance; **AR,** autosomal recessive; **AD,** autosomal dominant; **AAE,** amino acid exchange, ***** Frequencies refer to the genotypes of the 1000 Genome Project; **ExAC,** Exome Aggregation Consortium (http://http://exac.broadinstitute.org/); Mouse phenotype data were obtained from the mouse phenotype database: http://www.mousephenotype.org/data/search/gene

The following mitochondria associated genes were located in the authozygous region (n=17 out of 1130): *AMACR, ATL1, COQ4, DDHD1, ECHS1, ETFA, GCSH, GLDC, IDH2, MTFMT, PC, PDHX, POLG, SDHAF2,* ***SLC25A4,*** *UQCC3, WWOX*
=> **only in *SLC25A4* a potentially pathogenic variant was discovered by MutationTaster 2.**