CENTOGENE AG Am Strande 7 • 18055 Rostock • Germany

xxx

Order no.: xxx
Order received: xxx
Sample type: DNA
Sample collection date: xxx
Report date: xxx
Report type: Final Report

Patient no.: **xxx**, First Name: **xxx**, Last Name: **xxx**
DOB: **xxx**, Sex: **male**, Your ref.: **xxx**

Additional report recipient(s): xxx

Test(s) requested: CentoXome®

CLINICAL INFORMATION

Abnormal cortical gyration; Abnormal facial shape; Abnormal lung morphology; Abnormal temporal bone morphology; Abnormality of bony orbit of skull; Abnormality of the acetabulum; Abnormality of the basal ganglia; Abnormality of the maxilla; Abnormality of the optic disc; Agenesis of corpus callosum; Blindness; Delayed myelination; Encephalocele; Encephalomalacia; Global developmental delay; Hydronephrosis; Hydroureter; Microcephaly; Nephrocalcinosis; Optic disc pallor; Seizures; Vesicoureteral reflux
(Clinical information indicated above follows HPO nomenclature.)

Family history: Consanguineous parents.

Please see our concurrent reports xxx and xxx for the parents of the patient.

POSITIVE RESULT
Pathogenic variant identified

INTERPRETATION

A homozygous pathogenic variant was identified in the CPLANE1 gene.

A genetic diagnosis of autosomal recessive Joubert syndrome type 17 is confirmed.

RECOMMENDATIONS

- We recommend genetic counselling.

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RESULT SUMMARY

GENE	VARIANT COORDINATES	AMINO ACID CHANGE	SNP IDENTIFIER	ZYGOSITY	IN SILICO PARAMETERS*	ALLELE FREQUENCIES**	TYPE AND CLASSIFICATION***
CPLANE1	NM_023073.3:c.7778G>A	p.(Trp2593*)	rs863225159	homozygous	PolyPhen: N/A Align-GVGD: N/A SIFT: N/A MutationTaster: Disease causing Conservation_nt: weak Conservation_aa:	gnomAD: - ESP: - 1000 G: 0.00031 CentoMD: 0.00026	Nonsense Pathogenic (class 1)

Variant description based on OTFA (using VEP v93). * AlignGVD: C0: least likely to interfere with function, C65: most likely to interfere with function; splicing predictions: Ada and RF scores. ** Genome Aggregation Database (gnomAD), Exome Sequencing Project (ESP), 1000Genome project (1000G) and CentoMD® (latest database available). *** based on ACMG recommendations.

VARIANT INTERPRETATION

CPLANE1, c.7778G>A p.(Trp2593*)

The CPLANE1 variant c.7778G>A p.(Trp2593*) creates a premature stop codon. According to HGMD Professional 2019.1, this variant has previously been described as disease causing for Joubert syndrome by Bachmann-Gagescu et al., 2015 (PMID: 26092869). ClinVar lists this variant as pathogenic (clinical testing/research, Variation ID: 217574). Homozygosity has been confirmed by parental testing. It is classified as pathogenic (class 1) according to the recommendations of Centogene and ACMG (please, see additional information below).

Pathogenic variants in the CPLANE1 gene are associated with Joubert syndrome type 17 (JS17), an autosomal recessive disorder. Joubert syndrome is a clinically and genetically heterogeneous group of disorders characterized by hypoplasia of the cerebellar vermis with the characteristic neuroradiologic 'molar tooth sign,' and accompanying neurologic symptoms, including dysregulation of breathing pattern and developmental delay. Other variable features include retinal dystrophy and renal anomalies as well a characteristic appearance with large head, prominent forehead, high rounded eyebrows, epicanthal folds, upturned nose with prominent nostrils, an open mouth, tongue protrusion and rhythmic tongue motions, and occasionally low-set and tilted ears.
In addition, pathogenic CPLANE1 variants are associated with Orofaciodigital syndrome type VI (OFD6), or Varadi syndrome, which is a rare autosomal recessive disorder distinguished from other orofaciodigital syndromes by metacarpal abnormalities with central polydactyly and by cerebellar abnormalities, including the molar tooth sign. (OMIM®: *614571)

INCIDENTAL FINDINGS

We did not detect any class 1 or 2 variants in the genes for which incidental findings are reported based on the ACMG guidelines.

TABULAR LIST OF ADDITIONAL PATHOGENIC AND LIKELY PATHOGENIC VARIANTS

To provide the most comprehensive and relevant genetic information, we below list selected variants found in severe and early onset disease genes of your patient. We consider these at the time of reporting as "pathogenic" and "likely pathogenic" (see our mutation database CentoMD® for further information). Variants not included and classified in the current release of CentoMD®, and low quality variants which usually represent technical artifacts, are not included. In our review, we follow an internally expert-curated list of more than 1.700 genes currently, as attached hereto.
The listed variants may not directly answer the diagnostic request, at least not with the clinical information provided to CENTOGENE or current scientific understanding of relevant genetic disease mechanisms. However, these variants may help to close a potential diagnostic gap with regard to the current disease and are therefore provided here for a full diagnostic overview.

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Variants in genes related to late-onset diseases with unclear (considerably reduced) penetrance and/or cancer related genes with typically onset in adulthood only are not included in this list. This is therefore not a complete list of potentially relevant genetic variants in the patient and the classification of these variants may also change over time. CENTOGENE is not liable for any missing variant in this list and/or any provided classification of the variants at a certain point of time.
Insofar, as the identified variants may indicate (additional) genetic risks or diagnoses in the patient and/or his family and/or inform about reproductive risks, we strongly recommend to follow applicable local guidelines with regard to informing the patient about such findings. Particularly, if the patient decided not to be informed about "incidental findings" (to avoid any misunderstanding the list given here is not covering "incidental findings" according to ACMG), such are not reported in this variant list either, and you should carefully inform and check with the patient whether he/she wants to be informed about these additional variants.

GENE	VARIANT COORDINATES	AMINO ACID CHANGE	SNP IDENTIFIER	ZYGOSITY	IN SILICO PARAMETERS*	ALLELE FREQUENCIES**	TYPE AND CLASSIFICATION***
ANTXR2	NM_001145794.1:c.134T>C	p.(Leu45Pro)	rs886041401	heterozygous	PolyPhen: Probably damaging Align-GVGD: C0 SIFT: Deleterious MutationTaster: Disease causing Conservation_nt: high Conservation_aa: high	gnomAD: - ESP: - 1000 G: 0.00059 CentoMD: 0.00050	Missense Pathogenic (class 1)

Variant description based on OTFA (using VEP v93). * AlignGVD: C0: least likely to interfere with function, C65: most likely to interfere with function; splicing predictions: Ada and RF scores. ** Genome Aggregation Database (gnomAD), Exome Sequencing Project (ESP), 1000Genome project (1000G) and CentoMD® (latest database available). *** based on ACMG recommendations.

ANALYSIS STATISTICS WES

% TARGET NUCLEOTIDES COVERED			
0X	**≥ 10X**	**≥ 20X**	**≥ 50X**
0.03	**99.82**	**99.43**	**86.02**

CENTOGENE VARIANT CLASSIFICATION (BASED ON ACMG RECOMMENDATIONS)

Class 1 – Pathogenic
Class 2 – Likely pathogenic
Class 3 – Variant of uncertain significance (VUS)
Class 4 – Likely benign
Class 5 – Benign

Additionally, other types of clinical relevant variants can be identified (e.g. risk factors, modifiers).

METHODS

Double stranded DNA capture baits against approximately 36.5 Mb of the human coding exome (targeting >98% of the coding RefSeq and Gencode v28 regions, which was obtained from the human genome build GRCh37/hg19 on May 2018) were used to enrich target regions from fragmented genomic DNA with the Twist Human Core Exome Plus kit . The generated library is sequenced on an Illumina platform to obtain at least 20x coverage depth for >98% of the targeted bases. An in-house bioinformatics pipeline, including read alignment to GRCh37/hg19 genome assembly, variant calling and annotation, and comprehensive variant filtering is applied. All disease-causing variants reported in HGMD®, in ClinVar and in CentoMD® as well as all variants with minor allele frequency (MAF) below 1% in gnomAD database are considered. The investigation for relevant variants is focused on coding exons and flanking +/-20 intronic bases. All potential modes of inheritance patterns are considered. In addition, provided family history and clinical information are used to evaluate identified variants with respect to their pathogenicity and causality, and are categorized into classes 1 - 5 (see above). All variants related to the phenotype of the patient, except benign or likely benign variants, are reported.

Centogene has established stringent quality criteria and validation processes for variants detected by NGS. Low quality single nucleotide variants and all relevant deletion/insertion variants are confirmed by Sanger sequencing. Consequently, we warrant a specificity of >99.9% for all reported variants.

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LIMITATIONS

The genetic results are interpreted in the context of the provided clinical findings, family history, and other laboratory data. Only variants in genes potentially related to the proband's medical condition are reported. Misinterpretation of results may occur if the provided information is inaccurate and/or incomplete. If the obtained genetic results do not concur with the clinical findings, additional testing should be considered.

Complex genetic events such as copy number variants, inversions, translocations and repeat expansions, may not be reliably detected with Exome Sequencing. In addition, due to technology limitations, certain regions may be either not or poorly covered. In these regions variants cannot be confidently detected. Extremely low coverage calls (homo/hemizygous or heterozygous calls with less than three or four reads, respectively) are expected to be artifacts based on our extensive validations and consequently are not considered during the analysis.

ADDITIONAL INFORMATION

This test was developed and its performance validated by CENTOGENE AG. The US Food and Drug Administration (FDA) has determined that clearance or approval of this method is not necessary and thus neither have been obtained. This test has been developed for clinical purposes. All test results are reviewed, interpreted and reported by our scientific and medical experts.

In line with ACMG recommendations for reporting of incidental findings in clinical exome and genome sequencing (Genetics in Medicine, 2017; PMID: 27854360), we report incidental findings, i.e. pathogenic variants (class 1) and likely pathogenic variants (class 2) only in the recommended genes for the recommended phenotypes. For children (<15 years old), pathogenic and likely pathogenic variants in BRCA1/2, MLH1, MSH2, MSH6, PMS2 and MUTYH genes related to adult onset phenotypes are not reported.

To also exclude mistaken identity in your clinic, several guidelines recommend testing a second sample that is independently obtained from the proband. Please note that any further analysis will result in additional costs.

The classification of variants can change over the time. Please feel free to contact CENTOGENE (customer.support@centogene.com) in the future to determine if there have been any changes in classification of any reported variants.

DISCLAIMER

Any preparation and processing of a sample from patient material provided to CENTOGENE by a physician, clinical institute or a laboratory (by a "Partner") and the requested genetic and/or biochemical testing itself is based on the highest and most current scientific and analytical standards. However, in very few cases genetic or biochemical tests may not show the correct result, e.g. because of the quality of the material provided by a Partner to CENTOGENE or in cases where any test provided by CENTOGENE fails for unforeseeable or unknown reasons that cannot be influenced by CENTOGENE in advance. In such cases, CENTOGENE shall not be responsible and/or liable for the incomplete, potentially misleading or even wrong result of any testing if such issue could not be recognized by CENTOGENE in advance.

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Appendix - Current Gene List used for tabular list of additional pathogenic and likely pathogenic variants:

AAAS, AARS, ABAT, ABCA12, ABCB11, ABCB7, ABCC6, ABCC9, ABCD1, ABCD4, ABHD12, ABHD5, ACAD9, ACADM, ACADS, ACADVL, ACAN, ACAT1, ACE, ACO2, ACOX1, ACP5, ACSL4, ACTA1, ACTA2, ACTB, ACTG1, ACY1, ADA, ADAMTS2, ADAMTSL2, ADAR, ADGRG1, ADGRG6, ADGRV1, ADK, ADNP, ADPRHL2, ADSL, AFF2, AFG3L2, AGA, AGK, AGL, AGPS, AGRN, AGT, AGTR1, AGXT, AHDC1, AHI1, AIFM1, AIMP1, AIPL1, AIRE, AK2, AKR1D1, AKT3, ALDH18A1, ALDH1A3, ALDH3A2, ALDH4A1, ALDH5A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, ALMS1, ALPL, ALS2, ALX3, ALX4, AMER1, AMPD1, AMPD2, AMT, ANK2, ANKH, ANKRD11, ANO5, ANTXR1, ANTXR2, AP1S2, AP3B2, AP4B1, AP4E1, AP4M1, AP4S1, APOPT1, APTX, AR, ARCN1, ARFGEF2, ARG1, ARHGAP31, ARHGEF9, ARID1A, ARID1B, ARID2, ARL13B, ARL3, ARL6, ARMC4, ARMC9, ARSA, ARSB, ARSE, ARX, ASAH1, ASCC1, ASH1L, ASL, ASPA, ASPH, ASPM, ASS1, ASXL1, ASXL2, ASXL3, ATAD3A, ATIC, ATL1, ATOH7, ATP13A2, ATP1A1, ATP1A3, ATP6AP2, ATP6V0A2, ATP6V1A, ATP6V1B1, ATP6V1B2, ATP7A, ATP7B, ATP8A2, ATP8B1, ATRX, AUH, AUTS2, B3GALNT2, B3GALT6, B4GALT7, B9D1, BANF1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BCKDHA, BCKDHB, BCKDK, BCL11A, BCOR, BCS1L, BGN, BHLHA9, BICD2, BIN1, BLM, BMP2, BMP4, BMPER, BMPR1B, BOLA3, BPTF, BRAT1, BRD4, BRPF1, BRWD3, BSCL2, BSND, BTD, BTK, C12orf57, C12orf65, C1QBP, C2CD3, C8orf37, CA2, CA5A, CA8, CACNA1A, CACNA1C, CACNA1D, CACNA1E, CACNA1G, CAD, CAMK2A, CAMK2B, CAMTA1, CARS2, CASK, CAV1, CBL, CBS, CC2D1A, CC2D2A, CCBE1, CCDC103, CCDC114, CCDC115, CCDC151, CCDC22, CCDC39, CCDC40, CCDC47, CCDC65, CCDC78, CCDC8, CCDC88A, CCDC88C, CCND2, CCNK, CCNO, CCNQ, CD40LG, CDC45, CDH23, CDH3, CDK10, CDK13, CDK5RAP2, CDKL5, CDKN1C, CDON, CDT1, CENPF, CENPJ, CEP104, CEP135, CEP152, CEP290, CEP41, CEP57, CEP63, CEP83, CFAP298, CFAP300, CFAP410, CFC1, CFL2, CFTR, CHAMP1, CHAT, CHD2, CHD3, CHD4, CHD7, CHD8, CHM, CHMP1A, CHRDL1, CHRNA1, CHRNA2, CHRNA4, CHRND, CHRNE, CHRNG, CHST14, CHST3, CHSY1, CHUK, CIB2, CISD2, CIT, CKAP2L, CLCN1, CLCN4, CLCN7, CLCNKA, CLCNKB, CLDN1, CLDN19, CLMP, CLN3, CLN5, CLN6, CLN8, CLP1, CLPB, CLPP, CLRN1, CLTC, CNKSR2, CNPY3, CNTNAP1, CNTNAP2, COASY, COG1, COG4, COG5, COG7, COG8, COL10A1, COL11A1, COL11A2, COL17A1, COL18A1, COL1A1, COL1A2, COL2A1, COL4A1, COL4A2, COL4A3, COL4A3BP, COL4A4, COL4A5, COL6A1, COL6A2, COL6A3, COL7A1, COL9A1, COL9A2, COL9A3, COLEC10, COLEC11, COMP, COQ2, COQ4, COQ8A, COQ9, COX10, COX15, COX6B1, COX7B, CPLANE1, CPS1, CPT1A, CPT2, CRADD, CRB1, CRB2, CRBN, CREBBP, CRELD1, CRIPT, CRKL, CRLF1, CRTAP, CRX, CRYAA, CSNK2A1, CSPP1, CSTA, CSTB, CTC1, CTCF, CTDP1, CTNNA2, CTNNB1, CTNND1, CTNS, CTSA, CTSD, CTSK, CUL3, CUL4B, CUL7, CWC27, CYB5R3, CYC1, CYP11A1, CYP21A2, CYP24A1, CYP27A1, CYP27B1, CYP2U1, CYP7B1, D2HGDH, DAG1, DARS, DARS2, DBT, DCAF17, DCDC2, DCHS1, DCLRE1C, DCX, DDB2, DDC, DDHD1, DDHD2, DDR2, DDX11, DDX3X, DDX59, DEAF1, DENND5A, DEPDC5, DGAT1, DGUOK, DHCR24, DHCR7, DHDDS, DHODH, DHTKD1, DHX30, DIAPH1, DIS3L2, DKC1, DLAT, DLD, DLG3, DLG4, DLL3, DLL4, DLX5, DMD, DMP1, DMPK, DNA2, DNAAF3, DNAAF4, DNAAF5, DNAH5, DNAH9, DNAJC12, DNAJC19, DNAJC3, DNM1, DNMT3A, DNMT3B, DOCK6, DOCK7, DOCK8, DOK7, DOLK, DPAGT1, DPF2, DPM1, DPYD, DRC1, DSE, DSG1, DSP, DSTYK, DTNA, DVL1, DVL3, DYM, DYNC1H1, DYNC2H1, DYRK1A, EBF3, EBP, ECEL1, EDA, EDAR, EDNRA, EDNRB, EED, EEF1A2, EFEMP2, EFNB1, EFTUD2, EGR2, EHMT1, EIF2AK3, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF2S3, EIF4A3, ELMO2, ELN, ELOVL4, ELP1, ELP2, EMC1, EMD, EMG1, EMX2, ENPP1, ENTPD1, EOGT, EPG5, EPHB4, EPM2A, EPRS, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6L2, ERCC8, ERF, ERLIN2, ESCO2, ETFA, ETFB, ETFDH, ETHE1, EVC, EVC2, EXOSC3, EXOSC9, EXPH5, EXT1, EXT2, EXTL3, EYA1, EZH2, FA2H, FAH, FAM111A, FAM126A, FAM161A, FAM20A, FAM20C, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAR1, FARS2, FASTKD2, FAT4, FBLN5, FBN1, FBN2, FBP1, FBXL4, FBXO11, FBXW4, FEZF1, FGA, FGB, FGD1, FGD4, FGF10, FGF12, FGF3, FGF8, FGF9, FGFR1, FGG, FHL1, FIG4, FKBP14, FKRP, FKTN, FLAD1, FLNA, FLNB, FLT4, FLVCR1, FLVCR2, FMN2, FMR1, FN1, FOLR1, FOXC1, FOXC2, FOXE3, FOXF1, FOXG1, FOXL2, FOXN1, FOXP1, FOXP3, FOXRED1, FRAS1, FREM1, FREM2, FRMPD4, FRRS1L, FTCD, FTL, FTSJ1, FUCA1, FUT8, FXN, G6PC3, GAA, GABBR2, GABRA1, GABRB2, GABRB3, GABRG2, GALC, GALE, GALK1, GALNS, GALT, GAMT, GAN, GAS8, GATA1, GATA2, GATA3, GATA4, GATA6, GATAD2B, GATM, GBA, GBA2, GBE1, GCDH, GCH1, GDF1, GDF3, GDF5, GDF6, GDI1, GFAP, GFER, GFM1, GHR, GJA1, GJA8, GJB2, GJB3, GJC2, GK, GLA, GLB1, GLDC, GLDN, GLE1, GLI2, GLI3, GLIS2, GLIS3, GLMN, GLUD1, GLUL, GM2A, GMNN, GMPPA, GMPPB, GNA11, GNAI3, GNAO1, GNAQ, GNAS, GNB1, GNB5, GNPAT, GNPTAB, GNPTG, GNS, GORAB, GPAA1, GPC3, GPC6, GPHN, GPSM2, GPX4, GREB1L, GRHL2, GRHL3, GRIA3, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIN2D, GRIP1, GRM1, GSS, GTF2H5, GTPBP3, GUCY2C, GUSB, GZF1, HACE1, HADH, HADHA, HADHB, HARS, HAX1, HBA1, HBB, HCCS, HCFC1, HCN1, HDAC4, HDAC8, HECW2, HESX1, HEXA, HEXB, HGSNAT, HIBCH, HINT1, HIST1H1E, HIVEP2, HLCS, HMGCL, HMGCS2, HMX1, HNF1B, HNF4A, HNRNPH2, HNRNPK, HNRNPU, HOXA1, HOXA13, HOXC13, HOXD13, HPD, HPGD, HPRT1, HPS1, HPSE2, HSD17B10, HSD17B4, HSD3B7, HSPD1, HSPG2, HUWE1, HYAL1, HYDIN, HYLS1, IARS, IARS2, ICK, IDS, IDUA, IFIH1, IFITM5, IFT122, IFT140, IFT172, IFT43, IFT80, IGF1, IGF1R, IGF2, IGFBP7, IGHMBP2, IGSF1, IHH, IKBKG, IL11RA, IL1RAPL1, IL2RG, IMPAD1, INPP5E, INPP5K, INPPL1, INSR, INVS, IQCB1, IQSEC2, IRF6, IRX5, ISPD, ITGA3, ITGA6, ITGA7, ITGA8, ITGB4, ITPR1, IVD, JAG1, JAGN1, JAK3, JAM3, KANK1, KANSL1, KARS, KAT6A, KAT6B, KBTBD13, KCNA2, KCNB1, KCNC1, KCNC3, KCNE1, KCNH1, KCNJ1, KCNJ10, KCNJ11, KCNJ6, KCNMA1, KCNQ1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCTD1, KCTD7, KDM5C, KDM6A, KIAA0586, KIAA1109, KIAA1279, KIDINS220, KIF11, KIF1A, KIF22, KIF2A, KIF5C, KIF7, KIRREL3, KIT, KLF1, KLHL15, KLHL40, KLHL7, KMT2A, KMT2B, KMT2C, KMT2D, KMT2E, KMT5B, KPTN, KRIT1, KRT8, L1CAM, L2HGDH, LAMA1, LAMA2, LAMA3, LAMB1, LAMB2, LAMB3, LAMC2, LAMC3, LAMP2, LARGE1, LARP7, LARS2, LBR, LDB3, LEMD3, LFNG, LGI4, LHX3, LHX4, LIAS, LIFR, LIG4, LINS1, LIPT1, LMBRD1, LMNA, LMX1B, LONP1, LRBA, LRP2, LRP4, LRP5, LRPPRC, LRRC56, LRRC6, LTBP2, LTBP3, LYST, LZTR1, MAB21L2, MACF1, MAF, MAFB, MAGEL2, MAGI2, MAGT1, MAMLD1, MAN1B1, MAN2B1, MANBA, MAOA, MAP2K1, MAP2K2, MAP3K1, MAP3K7, MAPK8IP3, MAPRE2, MASP1, MATN3, MBD5, MBOAT7, MC2R, MCCC1, MCCC2, MCEE, MCOLN1, MCPH1, MDH2, MECOM, MECP2, MECR, MED12, MED13, MED13L, MED17, MED23, MEF2C, MEGF10, MEGF8, MEOX1, MESP2, MFRP, MFSD2A, MFSD8, MGAT2, MGP, MICU1, MID1, MIR17HG, MITF, MKKS, MKS1, MLC1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMP13, MMP21, MNX1, MOCS1, MOCS2, MOGS, MORC2, MPDU1, MPDZ, MPI, MPLKIP, MPV17, MPZ, MRPS2, MRPS22, MRPS34, MSL3, MSX1, MSX2, MTHFR, MTM1, MTO1, MTOR, MTR, MTRR, MTTP, MUSK, MUT, MUTYH, MVK, MYCN, MYH3, MYH6, MYH8, MYH9, MYO5A, MYO5B, MYO7A, MYPN, MYT1L, NAA10, NAA15, NACC1, NAGA, NAGLU, NAGS, NALCN, NANS, NARS2, NAXD, NAXE, NBAS, NBEA, NBN, NCAPD3, NDE1, NDP, NDST1, NDUFA1, NDUFA10, NDUFA6, NDUFAF2, NDUFAF5, NDUFAF6, NDUFB8, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NEB, NECTIN1, NECTIN4, NEDD4L, NEK1, NEK8, NEU1, NEUROG3, NEXMIF, NF1, NFIA, NFIB, NFIX, NFU1, NGLY1, NHLRC1, NHP2, NHS, NIPBL, NKX2-5, NKX3-2, NKX6-2, NMNAT1, NODAL, NOG, NONO, NOTCH1, NOTCH2, NOTCH3, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NPR2, NR0B1, NR2F1, NR2F2, NR5A1, NRXN1, NRXN3, NSD1, NSDHL, NSUN2, NT5C3A, NTRK1, NTRK2, NUBPL, NUP107, NYX, OBSL1, OCLN, OCRL, ODAPH, OFD1, OPA1, OPA3, OPHN1, ORC1, ORC4, ORC6, OSGEP, OSTM1, OTC, OTOGL, OTUD6B, OTULIN, OTX2, OXCT1, P3H1, P4HB, PACS1, PACS2, PAFAH1B1, PAH, PAK3, PANK2, PAPSS2, PARN, PAX2, PAX3, PAX6, PAX8, PAX9, PC, PCARE, PCBD1, PCCA, PCCB, PCDH15, PCDH19, PCGF2, PCNT, PCYT1A, PDCD10, PDE10A, PDE4D, PDE6G, PDGFRB, PDHA1, PDHB, PDHX, PDSS2, PEPD, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGAP1, PGAP2, PGAP3, PGK1, PGM1, PGM3, PHACTR1, PHF21A, PHF6, PHF8, PHGDH, PHIP, PHOX2B, PIEZO1, PIEZO2, PIGA, PIGG, PIGL, PIGN, PIGO, PIGS, PIGT, PIGV, PIGY, PIH1D3, PIK3R1, PIK3R2, PITX1, 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