Dr.

Country

Order no.:
Order received: dd.mm.yyyy
Sample type / Sample collection date:
blood, EDTA / dd.mm.yyyy
Report date: dd.mm.yyyy
Report type: Final Report

Patient no.:, First Name:, Last Name:
DOB: **dd.mm.yyyy**, Sex:, Your ref.: **-**

Additional report recipient(s):

Test(s) requested: CentoScreen Genome

CLINICAL INFORMATION

Unaffected.
Family history: Unknown.
Consanguineous parents: Unknown.

CARRIER STATUS CONFIRMED

Pathogenic and likely pathogenic variants identified; additional finding requiring confirmation identified

INTERPRETATION

A heterozygous pathogenic variant was identified in the *CA2* gene. Pathogenic variants in this gene are associated with autosomal recessive osteopetrosis with renal tubular acidosis.

A heterozygous pathogenic variant was identified in the *SI* gene. Pathogenic variants in this gene are associated with autosomal recessive congenital sucrose-isomaltase deficiency.

A heterozygous pathogenic variant was identified in the *SLC29A3* gene. Pathogenic variants in this gene are associated with autosomal recessive histiocytosis-lymphadenopathy plus syndrome.

A heterozygous likely pathogenic variant was identified in the *C6* gene. Pathogenic variants in this gene are associated with autosomal recessive C6 deficiency.

A heterozygous likely pathogenic variant was identified in the *C7* gene. Pathogenic variants in this gene are associated with autosomal recessive C7 deficiency.

The carrier status for the above identified variants is confirmed.

Additionally, a heterozygous pathogenic -$\alpha^{3.7}$ deletion resulting in the loss of one HBA copy was detected by the DRAGEN HBA caller (please see the Methods section below) indicating a high risk of the presence of this variant. **Orthogonal testing is recommended to confirm the presence and/or zygosity of this finding.** Pathogenic variants in this gene are associated with autosomal recessive alpha thalassemia (α-thalassemia).

No further clinically relevant variants were detected.

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RECOMMENDATIONS

- MLPA analysis of the *HBA* genes is recommended to confirm the detected variant.
- Partner testing is recommended for reproductive risk assessment.
- Targeted testing for affected family members, if any, and familial cascade carrier testing are recommended.
- Genetic counselling, including reproductive counselling (discussing prenatal and preimplantation diagnoses, if relevant), is recommended.

MAIN FINDINGS

SEQUENCE VARIANTS

GENE	VARIANT COORDINATES	AMINO ACID CHANGE	SNP IDENTIFIER	ZYGOSITY	IN SILICO PARAMETERS*	ALLELE FREQUENCIES**	TYPE AND CLASSIFICATION***
C6	NM_000065.3:c.2381+2T>C	p.(?)	rs76202909	heterozygous	PolyPhen: N/A SIFT: N/A MutationTaster: N/A Conservation_nt: high 2/2 likely splice effect	gnomAD: 0.0024 ESP: - 1000 G: -	Splicing Likely Pathogenic
C7	NM_000587.2:c.1561C>A	p.(Arg521Ser)	rs121964920	heterozygous	PolyPhen: Probably damaging SIFT: Deleterious MutationTaster: Disease causing Conservation_nt: weak	gnomAD: 0.0026 ESP: - 1000 G: -	Missense Likely Pathogenic
CA2	NM_000067.2:c.232+1G>A	p.(?)	rs573750741	heterozygous	PolyPhen: N/A SIFT: N/A MutationTaster: N/A Conservation_nt: high 2/2 likely splice effect	gnomAD: 0.0000040 ESP: - 1000 G: -	Splicing Pathogenic
SI	NM_001041.3:c.3218G>A	p.(Gly1073Asp)	rs121912616	heterozygous	PolyPhen: Probably damaging SIFT: Deleterious MutationTaster: Disease causing Conservation_nt: high	gnomAD: 0.0015 ESP: - 1000 G: -	Missense Pathogenic
SLC29A3	NM_018344.5:c.1088G>A	p.(Arg363Gln)	rs387907066	heterozygous	PolyPhen: Probably damaging SIFT: Deleterious MutationTaster: Disease causing Conservation_nt: high	gnomAD: 0.0000080 ESP: - 1000 G: -	Missense Pathogenic

Variant annotation based on CentoCloud Bioinformatics pipeline. * Splicing predictions: Ada and RF scores. ** Genome Aggregation Database (gnomAD), Exome Sequencing Project (ESP), 1000Genome project (1000G). *** Based on ACMG recommendations.

COPY NUMBER VARIATIONS

CNV DESCRIPTION*	SIZE (KB)	GENE COUNT**	INTERPRETATION***	RELATED DISORDER
Heterozygous deletion resulting in the loss of one HBA copy (-α3.7 deletion)	-	2	Pathogenic	Thalassemia, alpha- (604131), AR

CNV DESCRIPTION*	RefSeq GENES
Heterozygous deletion resulting in the loss of one HBA copy (-α3.7 deletion)	*HBA1, HBA2*

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VARIANT INTERPRETATION

C6, c.2381+2T>C p.(?)

The *C6* variant c.2381+2T>C p.(?) affects the canonical donor splice-site and skipping of the exon is predicted. According to HGMD Professional 2024.4, this variant has previously been described as disease causing for Complement C6 deficiency, subtotal (PMID:7535801, 38096369, 31589614). ClinVar lists this variant (Interpretation: Conflicting interpretations of pathogenicity; Pathogenic (4), Likely pathogenic (4), Uncertain significance (2); Variation ID: 379370). It is classified as likely pathogenic based on CENTOGENE's implementation of the ACMG/AMP/ClinGen SVI guidelines.

Deficiency of the sixth component of complement (C6D) is frequently associated with recurrent neisserial infections, especially meningitis caused by Neisseria meningitidis (PMID: 8690922). Mode of inheritance: autosomal recessive (OMIM®: 612446)

C7, c.1561C>A p.(Arg521Ser)

The *C7* variant c.1561C>A p.(Arg521Ser) causes an amino acid change from Arg to Ser at position 521 in exon(s) no. 12 (of 18). According to HGMD Professional 2024.4, this variant has previously been described as disease causing for Complement C7 deficiency (PMID:8871666, 38096369, 31589614). ClinVar lists this variant (Interpretation: Conflicting interpretations of pathogenicity; Pathogenic (6), Likely pathogenic (2), Uncertain significance (3); Variation ID: 12105). It is classified as likely pathogenic based on CENTOGENE's implementation of the ACMG/AMP/ClinGen SVI guidelines.

Patients with *C7* deficiency have an increased susceptibility to recurrent bacterial infections, especially meningitis caused by Neisseria meningitidis (Nishizaka et al., 1996; PMID:8892662). Mode of inheritance: autosomal recessive (OMIM®: 610102)

CA2, c.232+1G>A p.(?)

The *CA2* variant c.232+1G>A p.(?) affects the canonical donor splice-site and skipping of the exon is predicted. According to HGMD Professional 2024.4, this variant has previously been described as disease causing for Carbonic anhydrase deficiency (PMID:1301935, 37644014, 27717089). ClinVar lists this variant (Interpretation: Pathogenic; Variation ID: 288909). It is classified as pathogenic based on CENTOGENE's implementation of the ACMG/AMP/ClinGen SVI guidelines.

Pathogenic variants in the *CA2* gene are associated with autosomal recessive osteopetrosis with renal tubular acidosis. It is is a rare disorder characterized by osteopetrosis, renal tubular acidosis (RTA), and neurological disorders related to cerebral calcifications. Other clinical manifestations include fractures, growth failure and short stature, developmental delay, intellectual deficit, dental malocclusions/malalignment, cranial nerve compression and hearing impairment (ORPHA: 2785). Mode of inheritance: Autosomal recessive (OMIM®: 259730)

SI, c.3218G>A p.(Gly1073Asp)

The *SI* variant c.3218G>A p.(Gly1073Asp) causes an amino acid change from Gly to Asp at position 1073 in exon(s) no. 27 (of 48). According to HGMD Professional 2024.4, this variant has previously been described as possibly disease causing for Sucrase isomaltase deficiency (PMID:16329100, 38682389, 19121318). ClinVar lists this variant (Interpretation: Conflicting interpretations of pathogenicity; Pathogenic (4), Likely pathogenic (10), Uncertain significance (2); Variation ID: 1419). It is classified as pathogenic based on CENTOGENE's implementation of the ACMG/AMP/ClinGen SVI guidelines.

Congenital sucrose-isomaltase deficiency (CSID) is an autosomal recessive disorder characterized by absence of sucrase and most of the maltase digestive activity within the sucrase-isomaltase enzyme complex, with the isomaltase activity varying from absent to normal. The large amounts of unabsorbed disaccharides create osmotic-fermatative diarrhea with symptoms such as vomiting, flatulence, and abdominal pain (summary by Sander et al., 2006; PMID:16329100). Mode of Inheritance: Autosomal recessive (OMIM®: 222900)

SLC29A3, c.1088G>A p.(Arg363Gln)

The *SLC29A3* variant c.1088G>A p.(Arg363Gln) causes an amino acid change from Arg to Gln at position 363 in

exon(s) no. 6 (of 6). According to HGMD Professional 2024.4, this variant has previously been described as disease causing for H syndrome (PMID:19889517, 31589614, 34657628). ClinVar lists this variant (Interpretation: Pathogenic; Variation ID: 30948). It is classified as pathogenic based on CENTOGENE's implementation of the ACMG/AMP/ClinGen SVI guidelines.

The histiocytosis-lymphadenopathy plus syndrome comprises features of 4 histiocytic disorders previously thought to be distinct: Faisalabad histiocytosis (FHC), sinus histiocytosis with massive lymphadenopathy (SHML), H syndrome, and pigmented hypertrichosis with insulin-dependent diabetes mellitus syndrome (PHID). FHC was described as an autosomal recessive disease involving joint deformities, sensorineural hearing loss, and subsequent development of generalized lymphadenopathy and swellings in the eyelids that contain histiocytes (summary by Morgan et al., 2010; PMID:20140240). SHML, or familial Rosai-Dorfman disease, was described as a rare cause of lymph node enlargement in children, consisting of chronic massive enlargement of cervical lymph nodes frequently accompanied by fever, leukocytosis, elevated erythrocyte sedimentation rate, and polyclonal hypergammaglobulinemia. Mode of Inheritance: Autosomal recessive (OMIM®: 602782)

HBA, -α3.7 deletion

A heterozygous deletion encompassing the *HBA* region corresponding to the -$\alpha^{3.7}$ deletion was detected by the DRAGEN HBA caller (please see the Methods section below). Orthogonal testing is recommended to confirm the presence and/or zygosity of this finding. This is the most common α-globin gene deletion, which is caused by the breakage of DNA molecules in the α-globin genes (*HBA2* and *HBA1*) region and rejoining of the broken ends by leaving a globin genes region with single functional gene. It has previously been described as disease-causing for Alpha Thalassemia by Sakai et al., 2000 (PMID: 10807536) and other authors. It is classified as pathogenic based on CENTOGENE's implementation of the ACMG/AMP/ClinGen SVI guidelines

Pathogenic variants in *HBA1* and *HBA2*, the genes encoding α1- and α2-globin, are associated with alpha thalassemia (α-thalassemia). This phenotype is usually inherited in an autosomal recessive manner. The number of normal alpha genes (3, 2, 1 or none) results in 4 different α-thalassemia syndromes. Three normal alpha genes give a silent carrier state (α+-thalassemia). Two normal alpha genes result in hematologically recognizable α thalassemia trait (α°-thalassemia). Carriers of α°-thalassemia show microcytosis (low MCV), hypochromia (low MCH), normal percentages of HbA2 and HbF, and RBC inclusion bodies. One normal alpha gene results in clinically relevant HbH disease. No normal alpha gene results in 'homozygous alpha thalassemia' manifested as fatal hydrops fetalis (OMIM® 141800, Origa *et al*., 2013, PMID: 20301608, Wadi et al., 2011, PMID:21410534). Other genetic and acquired confounding factors can also contribute to phenotype diversity. A co-inherited beta thalassemia allele could alleviate the degree of globin chain imbalance. Another potential genetic modifier is the alpha hemoglobin stabilizing protein (AHSP). The AHSP, a molecular chaperone, stabilizes the α-globin chain in native structure to facilitate its interaction with the b-globin chain (Wadi et al., 2011, PMID:21410534)

METHODS

CentoScreen Genome

Genomic DNA is enzymatically fragmented and tagged with Illumina compatible adapter sequences. The libraries are paired-end sequenced on an Illumina platform to yield an average coverage depth of ≥ 30x. The panel gene list can be obtained in the appendix of this report. A bioinformatics pipeline based on the DRAGEN pipeline from Illumina, as well as CENTOGENE's in-house pipeline is applied. The sequencing reads are aligned to the Genome Reference Consortium Human Build 37 (GRCh37/hg19), as well as the revised Cambridge Reference Sequence (rCRS) of the Human Mitochondrial DNA (NC_012920). Sequence variants (SNVs/indels) and copy number variations (CNVs) are called using DRAGEN, Manta and in-house algorithms. Variants with a minor allele frequency (MAF) of less than 1% in gnomAD database, or disease-causing variants reported in HGMD®, in ClinVar or in CENTOGENE's in-house Biodatabank are evaluated. Although the evaluation is focused on coding exons and flanking intronic regions, the complete gene is interrogated for candidate pathogenic and likely pathogenic variants. For the panel genes, only carrier screening-appropriate modes of inheritance are considered for analysis. Sequence variants are categorized into five classes (pathogenic, likely pathogenic, variant of uncertain significance [VUS], likely benign, and benign) according to ACMG/AMP guidelines for classification of variants in addition to ClinGen recommendations. CNVs are categorized into five classes (pathogenic, likely pathogenic, variant of uncertain significance [VUS], likely benign, and benign) according to ACMG/ClinGen guidelines for classification of CNVs. Pathogenic or likely pathogenic variants detected in the panel genes are reported. Likely benign and benign variants are not reported. Although VUS are generally not reported, exceptions may apply. For detection of SNVs and indels in the regions targeted for downstream analysis a sensitivity of 99.9%, a specificity of 99.9%, and an accuracy of 99.9% is achieved. CNV detection software has a sensitivity of more than 95%. CENTOGENE has established stringent quality criteria and validation processes for variants detected by NGS. Variants with low sequencing quality and/or unclear zygosity are reported as "high risk" as they represent lower confidence calls and additional

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confirmatory testing is recommended to confirm their presence and/or zygosity. Screening of repeat expansions is performed by the ExpansionHunter algorithm for the following genes: *AR*, *CSTB*, *FMR1*, and *FXN*. *GBA1* screening is performed using Gauchian algorithm to detect recombination events affecting the region encompassing exons 9-11 (NM_000157.3), a region which has the highest homology to *GBAP1*. Spinal muscular atrophy (SMA) screening is performed using SMN Caller algorithm to detect the copy number of the *SMN1* gene. *CYP21A2* screening is performed using the *CYP21A2* Caller to detect the presence of nonallelic homologous recombination variants. HBA variant screening is performed using the HBA Caller. Results from ExpansionHunter and gene-specific callers are reported as screening results and additional confirmatory testing is recommended. If no information is shown from ExpansionHunter and gene-specific callers, the screening result can be assumed “low risk” result. If these callers are unable to analyze the data (no call), this information will be reported.

Inversion analysis of intron 1 and intron 22 of the F8 gene
Inverse shifting-PCR (IS-PCR) is used for analysis of the well-known inversions in intron 1 (Inv1) and intron 22 (Inv22) of the F8 gene. This IS-PCR includes 3 steps: (a) BclI restriction, (b) self-ligation of restriction fragments providing BclI rings, and (c) standard PCR analysis. For analysis of Inv1, multiplex PCR is performed using a set of 3 primers that yields a 304-bp amplicon for the non-rearranged allele and a 224-bp amplicon for the inverted allele Inv1. For analysis of Inv22, two separate PCRs are performed using a set of 3 primers that yields a 487-bp amplicon for the non-rearranged allele and a 559-bp amplicon for the inverted allele Inv22 (PMIDs: 15860568, 18284600). Reference sequence for F8: NM_000132.3.

ANALYSIS STATISTICS

CentoScreen Genome

Targeted nucleotides covered	≥ 10x	99.89%

LIMITATIONS

The genetic results are interpreted in the context of the provided clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the provided genetic data or patient information is inaccurate and/or incomplete. If the obtained genetic results are not compatible with the clinical findings, additional testing should be considered.

CentoScreen Genome
CentoScreen Genome is a screening test designed to assess the risk of the consultant being a carrier of a pathogenic or likely pathogenic variant for autosomal recessive or X-linked recessive disorders. The test is not intended to establish a genetic diagnosis for the consultant – whether they are unaffected or affected. However, the test result may include information about a medical condition of the consultant that requires follow-up. A negative result for this panel does not rule out the possibility of a genetic condition in the consultant or their offspring. Additionally, while a negative screening result significantly reduces the risk of being a carrier of a medical condition related to the panel genes, it does not eliminate the risk since variants may not be detected by this screening test. If the results obtained do not match the clinical or family history, additional testing should be considered. The genetic results are interpreted in the context of carrier screening. No phenotypic information or genetic results of this patient or other family members are considered for the analysis. CentoScreen Genome is intended for the screening of pathogenic and likely pathogenic variants in the consultant and results from their partner are not considered in the analysis. Evaluation and reporting are limited to pathogenic and likely pathogenic variants within the panel genes. Genes with mapping issues in the genome assembly used, and genomic regions that are hard to sequence by current technology and are without evidenced relevance for monogenic disorders, are excluded from this analysis. More complex genetic events not mentioned in the methods section, such as inversions and translocations, are not analyzed in this test. In addition, due to technology limitations, certain regions may be poorly covered, or not covered at all. In these regions and others encompassing repetitive, high-homology (such as pseudogene homology), and GC-rich sequences, relevant variants can be missed. Extremely low-coverage calls are expected to be artifacts based on our extensive validations and are consequently not considered during the analysis. Potential aberrant splicing is assessed with splice prediction tools. Deep intronic variants without strong prediction of aberrant splicing may not be reported, with the exception of known pathogenic splicing variants evidenced by external sources. The CNV detection sensitivity is decreased for repetitive regions, homologous regions such as pseudogenes, and for events spanning 2 or less exons. It is expected that lower quality samples (e.g., prenatal, product of conception, blood from patients with hematologic disorders, and highly degraded DNA) may generate lower quality NGS data; in these cases, CNV analysis, and/or additional integrated screening analyses in this test may not be possible to perform. The repeat expansion algorithm used is not designed to handle complex loci that harbor multiple repeats. Repeats are only genotyped if the coverage at the locus is at least 10x. The Gauchian algorithm can only detect non-recombinant-like variants from a set of 111 known *GBA1* variants and can detect recombination events affecting exons 9-11 only. Therefore, recombinations affecting other regions are not in the scope of this screening. Repeat expansion screening in the *AFF2* and *ARX* genes is not part of the analysis. Silent carriers may be missed with the SMN Caller algorithm (two *SMN1* copies on one chromosome, 2+0 genotype). The *CYP21A2* Caller can only detect the presence of nonallelic homologous recombination variants, but not their zygosity. Nonrecombinant-like *CYP21A2* variant calling is not included in the analysis. The HBA Caller can only detect genotypes from a set of 14 known HBA genotypes. Variants that fall outside these results will be reported as unclear. ExpansionHunter and gene-specific caller results (repeat expansion screening, *GBA1* variant screening, *SMN1* CNV screening, HBA screening and *CYP21A2* screening) are not confirmed by orthogonal methods. Any result should be interpreted in the context of screening and additional confirmatory testing is necessary to obtain a conclusive result. Negative results for the aforementioned complex regions do not eliminate the possibility of a pathogenic or likely pathogenic variant being present in that region. Since this test does not include orthogonal confirmation of low confidence calls or calls from screening

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algorithms, any such variants should be confirmed by an additional diagnostic method before being used for clinical purposes. Additionally, variant classification may change over time due to advances in scientific and medical knowledge. For example, a variant of uncertain clinical significance (not reported in this test), may later be reclassified as a pathogenic or likely pathogenic variant, or a variant reported as pathogenic or likely pathogenic may later be reclassified as of uncertain clinical significance. Therefore, this screening test may produce false-positive and false-negative results, which should be interpreted in the context of current medical knowledge.

Inversion analysis of intron 1 and intron 22 of the *F8* gene
Allele drop-out cannot be completely excluded by this method; polymorphic/normal genomic variation in the patient sample may interfere with variant detection and lead to reduced sensitivity of the assay. This limitation is even more relevant, when a (familial) positive control is not included in the test.

ADDITIONAL INFORMATION

This test was developed, and its performance was validated, by CENTOGENE. This test has been developed for clinical purposes. All test results are reviewed, interpreted and reported by our scientific and medical experts.

To exclude mistaken identity in your clinic, several guidelines recommend testing a second sample that is independently obtained from the consultand. 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
CentoScreen Genome

AAAS, AARS1, AARS2, AASS, ABAT, ABCA1, ABCA12, ABCA3, ABCA4, ABCB11, ABCB4, ABCB7, ABCC2, ABCC6, ABCC8, ABCD1, ABCD4, ABCG5, ABCG8, ABHD12, ABHD5, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACE, ACO2, ACOX1, ACOX2, ACP5, ACSF3, ACSL4, ACTA1, ACTL6B, ACY1, ADA, ADA2, ADAM17, ADAM9, ADAMTS10, ADAMTS13, ADAMTS17, ADAMTS18, ADAMTS19, ADAMTS2, ADAMTSL2, ADAMTSL4, ADAR, ADAT3, ADD3, ADGRG1, ADGRG6, ADGRV1, ADK, ADSL, ADSS1, AEBP1, AFF2, AFG3L2, AGA, AGBL5, AGK, AGL, AGPAT2, AGPS, AGRN, AGT, AGTPBP1, AGTR1, AGXT, AHCY, AHI1, AICDA, AIFM1, AIMP1, AIMP2, AIPL1, AIRE, AK1, AK2, AKR1D1, ALAD, ALB, ALDH18A1, ALDH1A3, ALDH3A2, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ALMS1, ALOX12B, ALOXE3, ALPK3, ALPL, ALS2, ALX1, ALX3, ALX4, AMACR, AMBN, AMER1, AMH, AMHR2, AMN, AMPD1, AMPD2, AMT, ANGPTL3, ANKS6, ANO10, ANO5, ANOS1, ANTXR1, ANTXR2, AP1B1, AP1S1, AP1S2, AP3B1, AP3B2, AP3D1, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APOC2, 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CAV1, CAVIN1, CBLIF, CBS, CC2D1A, CC2D2A, CCBE1, CCDC115, CCDC22, CCDC39, CCDC40, CCDC65, CCDC8, CCDC88A, CCDC88C, CCN6, CCND2, CCNO, CD19, CD247, CD27, CD2AP, CD320, CD3D, CD3E, CD3G, CD40, CD40LG, CD55, CD59, CD79A, CD79B, CD8A, CDAN1, CDC14A, CDC45, CDCA7, CDH11, CDH23, CDH3, CDHR1, CDIN1, CDK10, CDK5RAP2, CDKL5, CDON, CDSN, CDT1, CEBPE, CENPF, CEP104, CEP120, CEP135, CEP152, CEP164, CEP19, CEP290, CEP41, CEP55, CEP57, CEP63, CEP78, CEP83, CERKL, CERS1, CERS3, CFAP298, CFAP300, CFAP418, CFAP43, CFAP53, CFC1, CFD, CFH, CFI, CFL2, CFTR, CHAT, CHKB, CHM, CHMP1A, CHRDL1, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CHST3, CHST6, CHSY1, CHUK, CIB2, CIITA, CILK1, CISD2, CIT, CKAP2L, CLCF1, CLCN1, CLCN2, CLCN4, CLCN5, CLCN7, CLCNKA, CLCNKB, CLDN1, CLDN10, CLDN14, CLDN16, CLDN19, CLMP, CLN3, CLN5, CLN6, CLN8, CLP1, CLPB, CLPP, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CNKSR2, CNNM2, CNNM4, CNOT1, CNPY3, CNTNAP1, CNTNAP2, COA6, COA8, COASY, COG1, COG4, COG5, COG6, COG7, COG8, COL11A1, COL11A2, 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