BAYLOR GENETICS

BAYLOR GENETICS
2450 HOLCOMBE BLVD.
GRAND BLVD. RECEIVING DOCK
HOUSTON, TX 77021-2024

PHONE
1.800.411.4363
FAX
1.800.434.9850

CONNECT

PRENATAL COMPREHENSIVE REQUISITION

PATIENT INFORMATION (COMPLETE ONE FORM FOR EACH PERSON TESTED)

Fetus of: Patient Last Name | Patient First Name | MI | Date of Birth (MM / DD / YYYY)

Address | City | State | Zip | Phone

Accession # | Hospital / Medical Record #

Patient discharged from the hospital/facility: ◯ Yes ◯ No

Biological Sex: ◯ Female ◯ Male ◯ Unknown

Gender identity (if different from above):

REPORTING RECIPIENTS

Ordering Physician | Institution Name

Email (Required for International Clients) | Phone | Fax

ADDITIONAL RECIPIENTS

Name | Email | Fax

Name | Email | Fax

PAYMENT (FILL OUT ONE OF THE OPTIONS BELOW)

◯ **SELF PAYMENT**

☐ Pay With Sample ☐ Bill To Patient

◯ **INSTITUTIONAL BILLING**

Institution Name | Institution Code | Institution Contact Name | Institution Phone | Institution Contact Email

◯ **INSURANCE**

☐ Do Not Perform Test Until Patient is Aware of Out-Of-Pocket Costs (excludes prenatal testing)

REQUIRED ITEMS 1. Copy of the Front/Back of Insurance Card(s) 2. ICD10 Diagnosis Code(s) 3. Name of Ordering Physician 4. Insured Signature of Authorization

Name of Insured | Insured Date of Birth (MM / DD / YYYY)

Patient's Relationship to Insured | Phone of Insured

Address of Insured

City | State | Zip

Primary Insurance Co. Name | Primary Insurance Co. Phone

Primary Member Policy # | Primary Member Group #

Name of Insured | Insured Date of Birth (MM / DD / YYYY)

Patient's Relationship to Insured | Phone of Insured

Address of Insured

City | State | Zip

Secondary Insurance Co. Name | Secondary Insurance Co. Phone

Secondary Member Policy # | Secondary Member Group #

By signing below, I hereby authorize Baylor Genetics to provide my insurance carrier any information necessary, including test results, for processing my insurance claim. I understand that I am responsible for any co-pay, co-insurance, and unmet deductible that the insurance policy dictates, as well as any amounts not paid by my insurance carrier for reasons including, but not limited to, non-covered and non-authorized services. I understand that I am responsible for sending Baylor Genetics any and all payments that I receive directly from my insurance company in payment for this test. Please note that Medicare does not cover routine screening tests.

Patient's Printed Name | Patient's Signature | Date (MM / DD / YYYY)

STATEMENT OF MEDICAL NECESSITY (REQUIRED)

This test is medically necessary for the risk assessment, diagnosis, or detection of a disease, illness, impairment, symptom, syndrome, or disorder. The results will determine my patient's medical management and treatment decisions. The person listed as the Ordering Physician is authorized by law to order the test(s) requested herein. I confirm that I have provided genetic testing information to the patient and they have consented to genetic testing.

Physician's Printed Name | Physician's Signature | Date (MM / DD / YYYY)

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Fetus of: _____ Patient Last Name _____ Patient First Name _____ MI _____ / _____ / _____ Date of Birth (MM / DD / YYYY) _____ Biological Sex

ETHNICITY

- ◯ African American
- ◯ Ashkenazi Jewish
- ◯ East Asian (China, Japan, Korea)
- ◯ Finnish
- ◯ French Canadian
- ◯ Hispanic American
- ◯ Mennonite
- ◯ Middle Eastern (Saudi Arabia, Qatar, Iraq, Turkey)
- ◯ Native American
- ◯ Northern European Caucasian (Scandinavian, UK, Germany)
- ◯ Pacific Islander (Philippines, Micronesia, Malaysia, Indonesia)
- ◯ South Asian (India, Pakistan)
- ◯ Southeast Asian (Vietnam, Cambodia, Thailand)
- ◯ Southern European Caucasian (Spain, Italy, Greece)
- ◯ Other (Specify): _____

SAMPLE

Date of Collection (MM / DD / YYYY) _____ / _____ / _____

SAMPLE TYPE

- ◯ Amniotic Fluid _____ cc
- ◯ CVS _____ mg ☐ TA ☐ TC
- ◯ Fetal Blood _____ cc
- ◯ Cultured Amniocytes
- ◯ Cultured CVS

GESTATIONAL INFORMATION*

U/S Date (MM/DD/YYYY) _____ / _____ / _____

Gestational Age on U/S Date: _____ weeks _____ days

LMP Date (MM/DD/YYYY) _____ / _____ / _____

* NOTE: U/S dating increases Amniotic Fluid Alpha Fetoprotein (AFAFP) and Acetylcholinesterase (AChE) performance.

PARENTAL BLOODS (REQUIRED FOR CHROMOSOMAL MICROARRAY ANALYSIS (CMA))

- ◯ Maternal Blood _____ / _____ / _____ Date of Collection (MM/DD/YYYY) _____ Paternal Last Name _____ Paternal First Name
- ◯ Paternal Blood _____ / _____ / _____ Date of Collection (MM/DD/YYYY) _____ / _____ / _____ Date of Birth (MM/DD/YYYY)

NOTE: Parental bloods should be collected in an EDTA tube (5-7 cc) and labeled with name and DOB.

INDICATION FOR TESTING (REQUIRED)

- ◯ Pregnancy at Risk for Specific Genetic Disorder (Complete Familial Mutation information to the right)
- ◯ Advanced Maternal Age (AMA)
- ◯ Abnormal Maternal Screen
  - ☐ NTD ☐ TRI 21 ☐ TRI 18 ☐ Other: _____
- ◯ Abnormal NIPT (attach report)
  - ☐ NTD ☐ TRI 21 ☐ TRI 18 ☐ Other: _____
- ◯ Abnormal U/S (Specify)

_____

- ◯ Multiple Pregnancy Losses
- ◯ Parental Concern
- ◯ Other Indication (Attach Report and Specify)

_____

ICD-10 Diagnosis Code(s): _____

KNOWN FAMILIAL MUTATION/DISORDER SPECIFIC PRENATAL TESTING

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_____ Name of Baylor Genetic Counselor _____ / _____ / _____ Date (MM/DD/YYYY)

Additional Cultures to be sent later: ◯ Yes ◯ No

Cultures will be sent from: _____ Name of Laboratory

Gene Name: _____ Baylor Genetics Family #: _____

Please mark corresponding gene related disorder on pages 4 - 13

◯ (REQUIRED) Attached Familial Mutation Report

NOTICE FOR PRENATAL BIOCHEMICAL AND DNA TESTS: Please be aware that our specimen requirements and quality control measures are compliant with the American College of Medical Genetics (ACMG) Standards and Guidelines for Clinical Genetics Laboratories. While these requirements are intended to provide the highest level of assurance that a single laboratory can offer, the ideal practice to assure the accuracy of prenatal diagnosis testing is through duplicate testing conducted by independent laboratories. We recommend that referring medical professionals make the necessary arrangements for these two independent analyses for their patients prior to performing the prenatal diagnostic procedure.

Physician/Counselor Acknowledgement: _____

OTHER PRENATAL TESTING OPTIONS

IMPORTANT INSTRUCTIONS FOR CHROMOSOMAL MICROARRAY ANALYSIS (CMA) and/or FETAL MOLECULAR STUDIES: Parental Bloods (Draw 5-7cc in an EDTA tube) are required for control and MCC studies. Label with name, DOB, and complete Parental Bloods information above.

- ◯ AF - AFP
- ◯ AChE
- ◯ Aneuploidy FISH (24-48hrs for 13, 18, 21, X, Y)
- ◯ Chromosome Analysis
- ◯ COL1A1 & COL1A2-Related Disorders Panel
- ◯ Expanded CMA
- ◯ Expanded CMA + Limited Chromosome Karyotype (5 cell karyotype) *
- ◯ Noonan Spectrum Disorders Panel
- ◯ Targeted CMA
- ◯ Targeted CMA + Limited Chromosome Analysis (5 cell karyotype) *

Note: Cultured Fetal Samples are not accepted for CMA + Limited Karyotype.

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- ◯ 2-Methyl-3-Hydroxybutyryl-CoA Dehydrogenase Deficiency HSD17B10
- ◯ 3-Hydroxy-3-Methylglutaryl CoA lyase Deficiency HMGCL
- ◯ 3-Hydroxy-3-Methylglutaryl-CoA Synthase 2 Deficiency HMGCS2
- ◯ 3-Methylcrotonyl-CoA Carboxylase Deficiency, MCCC1-Related
- ◯ 3-Methylcrotonyl-CoA Carboxylase Deficiency, MCCC2-Related
- ◯ 3-Methylglutaconic Aciduria Type 1, AUH-Related
- ◯ ABCA4-Related Disorders
- ◯ ABCC8-Related Disorders (Diabetes Mellitus, Permanent Neonatal)
- ◯ ACACA-Related Disorders
- ◯ ACTA1-Related Disorders
- ◯ Acute Myeloid Leukemia CEBPA
- ◯ Acute Recurrent Myoglobinuria, LPIN1-Related
- ◯ Acyl-CoA Dehydrogenase, Short/Branched Chain Deficiency ACADSB
- ◯ Adenine Phosphoribosyltransferase Deficiency APRT
- ◯ Adenosine Deaminase Deficiency
- ◯ Adenylosuccinase Deficiency ADSL
- ◯ Adrenoleukodystrophy ABCD1
- ◯ AKT2-Related Disorders
- ◯ Alagille Syndrome JAG1
- ◯ Alpha-Mannosidosis MAN2B1
- ◯ ALPL-Related Disorders (Hypophosphatasia)
- ◯ AMACR-Related Disorders
- ◯ Androgen Insensitivity Syndrome AR
- ◯ Angelman Syndrome UBE3A
- ◯ ANO5-Related Disorders
- ◯ APC-Associated Polyposis Conditions
- ◯ Arginase Deficiency ARG1
- ◯ Argininosuccinate Lyase Deficiency (Argininosuccinic Aciduria) ASL
- ◯ ARL6-Related Disorders
- ◯ ARSACS SACS
- ◯ Arylsulfatase A Deficiency (Metachromatic Leukodystrophy) ARSA
- ◯ ARX-Related Disorders
- ◯ Aspartylglycosaminuria AGA
- ◯ Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia APTX
- ◯ Ataxia,Telangiectasia-like Disorder MRE11A
- ◯ Ataxia with Vitamin E Deficiency TTPA
- ◯ Atelosteogenesis Type 2 (SLC26A2-Related Disorders) SLC26A2 (DTDST)
- ◯ ATM-Related Disorders (Ataxia-Telangiectasia)
- ◯ ATP5A1-Related Disorders
- ◯ ATP6V0A2-Related Disorders
- ◯ Autoimmune Polyendocrinopathy 1 (APECED) AIRE
- ◯ Autosomal Recessive Congenital Ichthyosis, TGM1-Related
- ◯ Autosomal Recessive Polycystic Kidney Disease PKHD1
- ◯ B4GALT7-Related Disorders
- ◯ BAG3-Related Disorders
- ◯ Bardet-Biedl Syndrome 1, BBS1
- ◯ Bardet-Biedl Syndrome 2, BBS2
- ◯ Bardet-Biedl Syndrome 4, BBS4
- ◯ Bardet-Biedl Syndrome 5, BBS5
- ◯ Bardet-Biedl Syndrome 7, BBS7
- ◯ Bardet-Biedl Syndrome 9, BBS9
- ◯ Bardet-Biedl Syndrome 10, BBS10
- ◯ Bardet-Biedl Syndrome 12, BBS12
- ◯ Bardet-Biedl Syndrome 15, WDPCP
- ◯ Bardet-Biedl Syndrome, Modifier of, CCDC28B
- ◯ Bare Lymphocyte Syndrome Type I TAP1
- ◯ Bare Lymphocyte Syndrome Type II RFX5
- ◯ Bare Lymphocyte Syndrome Type II, CGA, CIITA
- ◯ Bare Lymphocyte Syndrome Type II, CGD, RFXAP
- ◯ Barth Syndrome TAZ
- ◯ Beta-Thalassaemia/Sickle Cell Anemia HBB
- ◯ BH4-Deficient Hyperphenylalaninemia A PTS
- ◯ Biotinidase Deficiency (BTD)
- ◯ Bloom Syndrome BLM
- ◯ BMPR1A-Related Disorders
- ◯ BRCA1-Related Disorders
- ◯ BRCA2-Related Disorders
- ◯ Breast Cancer BARD1
- ◯ Breast-Ovarian Cancer RAD51D
- ◯ BRIP1-Related Disorders
- ◯ BCS1L-Related Disorders (Complex III Deficiency; GRACILE Syndrome)
- ◯ Buschke-Ollendorff Syndrome LEMD3
- ◯ C10orf2/TWINKLE-Related Disorders
- ◯ Camurati-Engelmann Disease TGFB1
- ◯ Canavan Disease ASPA
- ◯ Carbamoyl Phosphate Synthetase I Deficiency CPS1
- ◯ Cardiofaciocutaneous Syndrome BRAF
- ◯ Carnitine-Acylcarnitine Translocase Deficiency SLC25A20 (CACT)
- ◯ Carnitine Deficiency, Systemic SLC22A5 (OCTN2)
- ◯ Carnitine Palmitoyltransferase IA Deficiency CPT1A
- ◯ Carnitine Palmitoyltransferase II Deficiency CPT2
- ◯ CASP8-Related Disorders
- ◯ CAV3-Related Disorders
- ◯ CD8 Deficiency, Familial CD8A

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DISORDER SPECIFIC TESTS

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- ◯ CDC73 -Related Disorders
- ◯ CDH1-Related Disorders
- ◯ CDH23-Related Disorders (Usher Syndrome 1D)
- ◯ CDKL5-Related Disorders
- ◯ CDKN1C-Related Disorders
- ◯ CDKN2A-Related Disorders
- ◯ Centronuclear Myopathy MTMR14
- ◯ Centronuclear Myopathy 3 MYF6
- ◯ Centronuclear Myopathy 4 CCDC78
- ◯ Centronuclear Myopathy, Autosomal Recessive BIN1
- ◯ Cerebrotendinous Xanthomatosis CYP27A1
- ◯ CFTR-Related Disorders (Cystic Fibrosis)
- ◯ CHD7-Related Disorders (CHARGE Syndrome)
- ◯ Chediak-Higashi Syndrome LYST
- ◯ CHEK2-Related Disorders
- ◯ CHRNA1-Related Disorders
- ◯ CHRNA7-Related Disorders
- ◯ CHRNB1-Related Disorders
- ◯ CHRND-Related Disorders
- ◯ Citrin Deficiency SLC25A13 (CTLN2)
- ◯ Citrullinemia I ASS1
- ◯ Cleidocranial Dysplasia RUNX2
- ◯ CLRN1-Related Disorders (Usher Syndrome 3A; Retinitis Pigmentosa)
- ◯ Coenzyme Q10 Deficiency ADCK3(CABC1)
- ◯ Coenzyme Q10 Deficiency COQ2
- ◯ Coenzyme Q10 Deficiency COQ6
- ◯ Coenzyme Q10 Deficiency PDSS2
- ◯ COG6-Related Disorders
- ◯ COL1A1-Related Disorders
- ◯ COL1A2-Related Disorders
- ◯ COL2A1-Related Disorders
- ◯ COL6A1-Related Disorders
- ◯ COL6A2-Related Disorders
- ◯ COL6A3-Related Disorders
- ◯ Combined Oxidative Phosphorylation Deficiency 1, GFM1-Related
- ◯ Combined Oxidative Phosphorylation Deficiency 3, TSFM-Related
- ◯ Combined Oxidative Phosphorylation Deficiency 5, MRPS22-Related
- ◯ Combined Oxidative Phosphorylation Deficiency 7, C12orf65-Related
- ◯ Combined Oxidative Phosphorylation Deficiency 8, AARS2-Related
- ◯ Complex I Deficiency, ACAD9-Related
- ◯ Complex I Deficiency, FOXRED1-Related
- ◯ Complex I Deficiency, NDUFA1-Related
- ◯ Complex I Deficiency, NDUFA11-Related
- ◯ Complex I Deficiency, NDUFAF1-Related
- ◯ Complex I Deficiency, NDUFAF2-Related
- ◯ Complex I Deficiency, NDUFAF3-Related
- ◯ Complex I Deficiency, NDUFB8-Related
- ◯ Complex I Deficiency, NDUFS1-Related
- ◯ Complex I Deficiency, NDUFS3-Related
- ◯ Complex I Deficiency, NDUFS4-Related
- ◯ Complex I Deficiency, NDUFS6-Related
- ◯ Complex I Deficiency, NDUFS8-Related
- ◯ Complex I Deficiency, NDUFV1-Related
- ◯ Complex I Deficiency, NUBPL-Related
- ◯ Complex II Deficiency, SDHA-Related
- ◯ Complex II Deficiency, SDHAF1-Related
- ◯ Complex II Deficiency, SDHB-Related
- ◯ Complex III Deficiency, TTC19-Related
- ◯ Complex IV (COX) Deficiency, COX4I1-Related
- ◯ Complex IV (COX) Deficiency, COX10-Related
- ◯ Complex IV (COX) Deficiency, SCO1-Related
- ◯ Complex IV (COX) Deficiency, SCO2-Related
- ◯ Complex IV (COX) Deficiency, SURF1-Related
- ◯ Complex IV (COX) Deficiency, TACO1-Related
- ◯ Complex V Deficiency, ATP5E-Related
- ◯ Compton-North Congenital Myopathy CNTN1
- ◯ Cone-rod Dystrophy 15 CDHR1
- ◯ Congenital Adrenal Hyperplasia CYP11B1
- ◯ Congenital Adrenal Hyperplasia CYP17A1
- ◯ Congenital Amegakaryocytic Thrombocytopenia MPL
- ◯ Congenital Bile Acid Synthesis Defect 2 AKR1D1
- ◯ Congenital Disorders of Glycosylation CDG1A, PMM2-Related
- ◯ Congenital Disorders of Glycosylation CDG1B, MPI-Related
- ◯ Congenital Disorders of Glycosylation CDG1C, ALG6-Related
- ◯ Congenital Disorders of Glycosylation CDG1D, ALG3-Related
- ◯ Congenital Disorders of Glycosylation CDG1F, MPDU1-Related
- ◯ Congenital Disorders of Glycosylation CDG1G, ALG12-Related
- ◯ Congenital Disorders of Glycosylation CDG1H, ALG8-Related
- ◯ Congenital Disorders of Glycosylation CDG1K, ALG1-Related
- ◯ Congenital Disorders of Glycosylation CDG1L, ALG9-Related
- ◯ Congenital Disorders of Glycosylation CDG1M, DOLK-Related
- ◯ Congenital Disorders of Glycosylation CDG1P, ALG11-Related
- ◯ Congenital Disorders of Glycosylation CDG1R, DDOST-Related
- ◯ Congenital Disorders of Glycosylation CDG1S, ALG13-Related

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DISORDER SPECIFIC TESTS

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- ◯ Congenital Disorders of Glycosylation CDG1U, DPM2-Related
- ◯ Congenital Disorders of Glycosylation CDG1V, NGLY1-Related
- ◯ Congenital Disorders of Glycosylation CDG2B, MOGS-Related
- ◯ Congenital Disorders of Glycosylation CDG2C, SLC35C1 (FUCT1)-Related
- ◯ Congenital Disorders of Glycosylation CDG2D, B4GALT1-Related
- ◯ Congenital Disorders of Glycosylation CDG2E, COG7-Related
- ◯ Congenital Disorders of Glycosylation CDG2F, SLC35A1 (CST)-Related
- ◯ Congenital Disorders of Glycosylation CDG2G, COG1-Related
- ◯ Congenital Disorders of Glycosylation CDG2H, COG8-Related
- ◯ Congenital Disorders of Glycosylation CDG2I, COG5-Related
- ◯ Congenital Disorders of Glycosylation CDG2J, COG4-Related
- ◯ Congenital Disorders of Glycosylation CDG2K, TMEM165-Related
- ◯ Congenital Disorders of Glycosylation CDG2M, SLC35A2 (UGALT)-Related
- ◯ Congenital Generalized Lipodystrophy Type 4 PTRF
- ◯ Congenital Muscular Dystrophy due to ITGA7 Deficiency ITGA7
- ◯ Congenital Muscular Dystrophy, Megaconial Type CHKB
- ◯ Congenital Muscular Dystrophy-Dystroglycanopathy with Brain and Eye Anomalies Type A 8 POMGNT2
- ◯ Congenital Muscular Dystrophy-Dystroglycanopathy with Brain and Eye Anomalies Type A 10 TMEM5
- ◯ Congenital Muscular Dystrophy-Dystroglycanopathy with Brain and Eye Anomalies Type A 11 B3GALNT2
- ◯ Congenital Muscular Dystrophy-Dystroglycanopathy with Brain and Eye Anomalies Type A 12 POMK
- ◯ Congenital Myasthenia with Tubular Aggregates 1 GFPT1
- ◯ Congenital Myasthenic Syndrome, AGRN-Related
- ◯ Congenital Myasthenic Syndrome, ALG14-Related
- ◯ Congenital Myasthenic Syndrome, CHAT-Related
- ◯ Congenital Myasthenic Syndrome, CHRNE-Related
- ◯ Congenital Myasthenic Syndrome, DOK7-Related
- ◯ Congenital Myasthenic Syndrome, RAPSN-Related
- ◯ Congenital Myopathy PTPLA
- ◯ Costello Syndrome HRAS
- ◯ COX15-Related Disorders
- ◯ CP-Related Disorders
- ◯ CPT1B-Related Disorders
- ◯ Creatine Transporter (CRTR) Deficiency SLC6A8 (CT1)
- ◯ Crigler-Najjar Syndrome UGT1A1
- ◯ CRYAB-Related Disorders
- ◯ Cutaneous Malignant Melanoma 3 CDK4
- ◯ CYP1B1-Related Disorders (Primary Congenital Glaucoma)
- ◯ Cystinosis CTNS
- ◯ Danon Disease LAMP2
- ◯ Deafness-Dystonia-Optic Neuropathy TIMM8A
- ◯ Complex I Deficiency, FOXRED1-Related
- ◯ Complex I Deficiency, NDUFA1-Related
- ◯ Desmoplastic Medulloblastoma SUFU
- ◯ DES-Related Disorders
- ◯ DGUOK Sequence Analysis
- ◯ Diamond-Blackfan Anemia RPS19
- ◯ Digenic Fascioscapulohumeral Muscular Dystrophy 2 SMCHD1
- ◯ DiGeorge Syndrome TBX1
- ◯ Dihydrolipoamide Dehydrogense Deficiency DLD
- ◯ Dihydropyrimidine Dehydrogenase Deficiency DPYD
- ◯ DNM2-Related Disorders
- ◯ DOCK8-Related Disorders
- ◯ DPAGT1-Related Disorders
- ◯ DYSF-Related Disorders
- ◯ Dystrophinopathies (Duchenne/Becker) DMD
- ◯ Early-Onset Distal Myopathy KLHL9
- ◯ Early-Onset Myopathy, Areflexia, Respiratory Distress, and Dysphagia (EMARDD) MEGF10
- ◯ Ehlers-Danlos Syndrome, Classic Type COL5A1
- ◯ Ehlers-Danlos Syndrome, Classic Type COL5A2
- ◯ Ehlers-Danlos Syndrome, Kyphoscoliotic form PLOD1
- ◯ Ehlers-Danlos Syndrome Type IV COL3A1
- ◯ Ehlers-Danlos Syndrome, Spondylocheiro Dysplastic Form SLC39A13 (ZnT)
- ◯ Emery-Dreifuss Muscular Dystrophy 1, X-Linked EMD
- ◯ Emery-Dreifuss Muscular Dystrophy 5, Autosomal Dominant SYNE2
- ◯ Endplate Acetylcholinesterase Deficiency COLQ
- ◯ Epileptic Encephalopathy, Early Infantile, Type 4 STXBP1
- ◯ Epileptic Encephalopathy, Early Infantile, Type 7 KCNQ2
- ◯ Erythrocytic AMP Deaminase Deficiency AMPD3
- ◯ Ethylmalonic Encephalopathy ETHE1
- ◯ Exudative Vitreoretinopathy 5 TSPAN12
- ◯ Fabry Disease GLA
- ◯ FAM20C-Related Disorders
- ◯ Familial Dysautonomia IKBKAP
- ◯ Fanconi Anaemia FANCC
- ◯ Fanconi Anemia, CGN, PALB2
- ◯ Fanconi Anemia, CGO, RAD51C
- ◯ Fanconi-Bickel Syndrome SLC2A2 (GLUT2)
- ◯ FARS2-Related Disorders
- ◯ FASTKD2-Related Disorders
- ◯ FBN1-Related Disorders
- ◯ FH-Related Disorders
- ◯ FHL1-Related Disorders
- ◯ Fibrodysplasia Ossificans Progressiva ACVR1
- ◯ Congenital Disorders of Glycosylation CDG1S, ALG13-Related

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- ◯ FLNC-Related Disorders
- ◯ FKRP-Related Disorders
- ◯ FLCN -Related Disorders
- ◯ FMR1-Related Disorders (Fragile X)
- ◯ Focal Dermal Hypoplasia PORCN
- ◯ FOXF1-Related Disorders
- ◯ Fructose 1,6 Bisphosphatase Deficiency FBP1
- ◯ Fukuyama Congenital Muscular Dystrophy FKTN
- ◯ FZD4-Related Disorders
- ◯ Galactosemia GALE
- ◯ Galactosemia GALT
- ◯ Galactokinase Deficiency GALK1
- ◯ GAMT Deficiency GAMT
- ◯ GATA2-Related Disorders
- ◯ GATA6-Related Disorders
- ◯ GATM Deficiency (Arginine:Glycine Amidinotransferase Deficiency) GATM
- ◯ Gaucher Disease GBA
- ◯ GBE1-Related Disorders
- ◯ GCK -Related Disorders
- ◯ GJB2-Related Hearing Loss and Deafness
- ◯ Glucose-6-Phosphate Dehydrogenase Deficiency G6PD
- ◯ Glucose Transporter Type 1 Deficiency Syndrome SLC2A1 (GLUT1)
- ◯ Glutaric Acidemia Type 1 GCDH
- ◯ Glutaric Acidemia Type 3 C7orf10
- ◯ Glycine Encephalopathy AMT
- ◯ Glycogen Storage Disease Type 0, Liver Isoform GYS2
- ◯ Glycogen Storage Disease Type 0, Muscle Isoform GYS1
- ◯ Glycogen Storage Disease Type 1a G6PC
- ◯ Glycogen Storage Disease Type 1 (b, c, d) SLC37A4 (GSD1B)
- ◯ Glycogen Storage Disease Type II (Pompe Disease) GAA
- ◯ Glycogen Storage Disease Type III AGL
- ◯ Glycogen Storage Disease Type V PYGM
- ◯ Glycogen Storage Disease Type VI PYGL
- ◯ Glycogen Storage Disease Type VII PFKM
- ◯ Glycogen Storage Disease Type IX PHKA1
- ◯ Glycogen Storage Disease Type IX PHKA2
- ◯ Glycogen Storage Disease Type IX PHKB
- ◯ Glycogen Storage Disease Type IX PHKG2
- ◯ Glycogen Storage Disease Type X PGAM2
- ◯ Glycogen Storage Disease Type XI LDHA
- ◯ Glycogen Storage Disease Type XIII ENO3
- ◯ Glycogen Storage Disease Type XIV PGM1
- ◯ Glycogen Storage Disease Type XV GYG1
- ◯ GMPPB-Related Disorders
- ◯ GNE-Related Disorders (Inclusion Body Myopathy Type 2)
- ◯ GPC3-Related Disorders
- ◯ Gyrate Atrophy of Choroid and Retina OAT
- ◯ HADH-Related Disorders
- ◯ HADHA-Related Disorders (LCHAD Deficiency)
- ◯ HADHB-Related Disorders
- ◯ HARS2-Related Disorders
- ◯ Hearing Loss and Deafness, Nonsyndromic, GJB2-Related
- ◯ Hearing Loss, X-Linked Nonsyndromic, POU3F4
- ◯ Hemochromatosis Type 1 HFE
- ◯ Hemochromatosis Type 2A HFE2
- ◯ Hemochromatosis Type 2B HAMP
- ◯ Hemochromatosis Type 3 TFR2
- ◯ Hemochromatosis Type 4 SLC40A1 (HFE4)
- ◯ Hemophagocytic Lymphohistiocytosis 3, Familial, UNC13D
- ◯ Hemophagocytic Lymphohistiocytosis 4, Familial, STX11
- ◯ Hemophagocytic Lymphohistiocytosis 5, Familial, STXBP2
- ◯ Hereditary Fructose Intolerance ALDOB
- ◯ Hereditary Hemorrhagic Telangiectasia Type 1 ENG
- ◯ Hereditary Motor and Sensory Neuropathy with Agenesis of the Corpus Callosum SLC12A6 (KCC3A)
- ◯ Herlitz Junctional Epidermolysis Bullosa, LAMA3-Related
- ◯ Herlitz Junctional Epidermolysis Bullosa, LAMB3-Related
- ◯ Herlitz Junctional Epidermolysis Bullosa, LAMC2-Related
- ◯ Hermansky-Pudlak Syndrome 1 HPS1
- ◯ Hermansky-Pudlak Syndrome 2 AP3B1
- ◯ Hermansky-Pudlak Syndrome 3 HPS3
- ◯ Hermansky-Pudlak Syndrome 4 HPS4
- ◯ Hermansky-Pudlak Syndrome 5 HPS5
- ◯ Hermansky-Pudlak Syndrome 6 HPS6
- ◯ Hermansky-Pudlak Syndrome 7 DTNBP1
- ◯ Hermansky-Pudlak Syndrome 8 BLOC1S3
- ◯ HNF1A-Related Disorders
- ◯ HNF1B-Related Disorders
- ◯ HNRNPA1-Related Disorders
- ◯ Holocarboxylase Synthetase Deficiency HLCS
- ◯ Homocystinuria Caused by Cystathionine Beta-Synthase Deficiency CBS
- ◯ HPD Related Disorders HPD
- ◯ HSD17B4-Related Disorders (D-Bifunctional Protein Deficiency)
- ◯ Huntington Disease
- ◯ Congenital Disorders of Glycosylation CDG1S, ALG13-Related

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- ◯ Hyperinsulinism-Hyperammonemia Syndrome GLUD1
- ◯ Hypermethioninemia GNMT
- ◯ Hypermethioninemia with S-Adenosylhomocysteine Hydrolase Deficiency AHCY
- ◯ Hyperornithinemia-Hyperammonemia-Homocitrullinuria (HHH) Syndrome SLC25A15 (HHH)
- ◯ Hyperprolinemia Type II ALDH4A1
- ◯ Hypophosphatemic Nephrolithiasis/Osteoporosis, 1 SLC34A1 (NPT2)
- ◯ Hypothyroidism, Congenital, IYD
- ◯ Ichthyosis, X-Linked (STS Deficiency) FISH
- ◯ Ichthyosis, X-Linked BIOCHEMICAL
- ◯ IKBKG-Related Disorders
- ◯ IKZF1-Related Disorders
- ◯ Immunodeficiency Type 8 CORO1A
- ◯ Immunodeficiency Type 9 ORAI1
- ◯ Immunodeficiency Type 17 CD3G
- ◯ Immunodeficiency Type 18 CD3E
- ◯ Immunodeficiency Type 19 CD3D
- ◯ Immunodeficiency Type 22 LCK
- ◯ Immunodysregulation, Polyendocrinopathy, and Enteropathy, X-linked FOXP3
- ◯ Inclusion Body Myopathy 3 MYH2
- ◯ Inclusion Body Myopathy with Early-Onset Paget Disease with or without Frontotemporal Dementia 2 HNRNPA2B1
- ◯ INS-Related Disorders
- ◯ INSR-Related Disorders
- ◯ Intermediate Charcot-Marie-Tooth Neuropathy, KARS-Related
- ◯ Intrahepatic Cholestasis 1, Progressive Familial (PFIC1) ATP8B1
- ◯ Intrahepatic Cholestasis 2, Progressive Familial (PFIC2) ABCB11
- ◯ Intrahepatic Cholestasis 3, Progressive Familial (PFIC3) ABCB4
- ◯ Intrinsic Factor Deficiency GIF
- ◯ Isobutyryl-CoA Dehydrogenase Deficiency ACAD8
- ◯ Isovaleric Acidemia IVD
- ◯ ISPD-Related Disorders
- ◯ Joubert Syndrome TMEM216
- ◯ KCNJ11-Related Disorders
- ◯ Ketothiolase Deficiency ACAT1
- ◯ KIF11-Related Disorders
- ◯ Krabbe Disease GALC
- ◯ LAMA2-Related Disorders
- ◯ LAMB2-Related Disorders
- ◯ LARGE-Related Disorders
- ◯ LARS2-Related Disorders
- ◯ LCAD Deficiency ACADL
- ◯ LDB3-Related Disorders
- ◯ Leber Congenital Amaurosis, AIPL1-Related
- ◯ Leber Congenital Amaurosis, CABP4-Related
- ◯ Leber Congenital Amaurosis, CEP290-Related
- ◯ Leber Congenital Amaurosis, CRB1-Related
- ◯ Leber Congenital Amaurosis, CRX-Related
- ◯ Leber Congenital Amaurosis, GUCY2D-Related
- ◯ Leber Congenital Amaurosis, IMPDH1-Related
- ◯ Leber Congenital Amaurosis, IQCB1-Related
- ◯ Leber Congenital Amaurosis, LCA5-Related
- ◯ Leber Congenital Amaurosis, LRAT-Related
- ◯ Leber Congenital Amaurosis, RDH12-Related
- ◯ Leber Congenital Amaurosis, RPE65-Related
- ◯ Leber Congenital Amaurosis, RPGRIP1-Related
- ◯ Leber Congenital Amaurosis, SPATA7-Related
- ◯ Leber Congenital Amaurosis, TULP1-Related
- ◯ Leigh Syndrome, French-Canadian Type LRPPRC
- ◯ Lesch-Nyhan Disease HPRT
- ◯ Lethal Encephalopathy -Due to defective mitochondrial peroxisomal fission DNM1L
- ◯ Leukemia, Acute Lymphoblastic PAX5
- ◯ Leukoencephalopathy (LBSL), DARS2-Related
- ◯ Leukoencephalopathy (VWM), EIF2B5-Related
- ◯ Leukoencephalopathy with Dystonia and Motor Neuropathy SCP2
- ◯ LIG4-Related Disorders
- ◯ Limb-Girdle Muscular Dystrophy Type 1E DNAJB6
- ◯ Limb-Girdle Muscular Dystrophy Type 1F TNPO3
- ◯ Limb-Girdle Muscular Dystrophy Type 2A CAPN3
- ◯ Limb-Girdle Muscular Dystrophy Type 2C SGCG
- ◯ Limb-Girdle Muscular Dystrophy Type 2D SGCA
- ◯ Limb-Girdle Muscular Dystrophy Type 2E SGCB
- ◯ Limb-Girdle Muscular Dystrophy Type 2S TRAPPC11
- ◯ Liver Failure, Acute Infantile TRMU
- ◯ LMNA-Related Disorders
- ◯ Lowe Syndrome OCRL1
- ◯ LRP5-Related Disorders
- ◯ Lymphoproliferative Syndrome 1 ITK
- ◯ Lymphoproliferative Syndrome 1, X-linked, SH2D1A
- ◯ Lymphoproliferative Syndrome 2, X-linked, XIAP
- ◯ Lysinuric Protein Intolerance SLC7A7 (LAT1)
- ◯ Malabsorptive Congenital Diarrhea 4 NEUROG3
- ◯ Malonic & Methylmalonic Aciduria, Combined ACSF3

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- ◯ Maple Syrup Urine Disease Type 1A BCKDHA
- ◯ Maple Syrup Urine Disease Type 1B BCKDHB
- ◯ Maple Syrup Urine Disease Type 2 DBT
- ◯ MARS2-Related Disorders
- ◯ Maturity-Onset Diabetes of the Young (MODY) Type I HNF4A
- ◯ Maturity-Onset Diabetes of the Young (MODY) Type II BLK
- ◯ Maturity-Onset Diabetes of the Young (MODY) Type VI NEUROD1
- ◯ Maturity-Onset Diabetes of the Young (MODY) Type VII KLF11
- ◯ MCAD Deficiency ACADM
- ◯ MECP2-Related Disorders (Rett)
- ◯ Megalencephalic Leukoencephalopathy with Subcortical Cysts, MLC1-Related
- ◯ Menkes Disease ATP7A
- ◯ MET-Related Disorders
- ◯ Methylcobalamin Deficiency, cblE Type MTRR
- ◯ Methylcobalamin Deficiency, cblG Type MTR
- ◯ Methylmalonic Acidemia, MCEE-Related
- ◯ Methylmalonic Acidemia, MMAA-Related
- ◯ Methylmalonic Acidemia, MMAB-Related
- ◯ Methylmalonic Acidemia, MMADHC-Related
- ◯ Methylmalonic Acidemia, MUT-Related
- ◯ Methylmalonic Acidemia and Homocysteinemia, cblX Type HCFC1
- ◯ Methylmalonic Aciduria and Homocystinuria, cblF Type LMBRD1
- ◯ Methylmalonic Aciduria due to Transcobalamin Receptor Defect CD320
- ◯ MHC Class II Deficiency, CGB, RFXANK
- ◯ Microcephaly, Epilepsy, and Diabetes Syndrome IER3IP1
- ◯ Microphthalmia, Isolated 5, Disorder MFRP
- ◯ Mitchell-Riley Syndrome RFX6
- ◯ Mitochondrial Myopathy and Sideroblastic Anemia Type 1 PUS1
- ◯ Mitochondrial Myopathy and Sideroblastic Anemia Type 2 YARS2
- ◯ Mitochondrial Progressive Myopathy with Congenital Cataract, Hearing Loss, and Developmental Delay GFER
- ◯ MKKS-Related Disorders
- ◯ MKS1-Related Disorders
- ◯ MMACHC (cblC) -Related Disorders (Methylmalonic Aciduria and Homocystinuria, cblC Type)
- ◯ MNGIE Syndrome TYMP
- ◯ Molybdenum Cofactor Deficiency MOCS1
- ◯ Molybdenum Cofactor Deficiency MOCS2
- ◯ MPV17-Related Disorders
- ◯ MRPL44-Related Disorders
- ◯ MTFMT-Related Disorders
- ◯ mtDNA Depletion Syndrome 13, Encephalomyopathic Type FBXL4
- ◯ mtDNA Depletion Syndrome, Encephalomyopathic Form SUCLG2
- ◯ mtDNA Depletion Syndrome, Myopathic RRM2B
- ◯ mtDNA Depletion Syndrome, Myopathic SUCLA2
- ◯ mtDNA Depletion Syndrome, Myopathic TK2
- ◯ Mucolipidosis IV MCOLN1
- ◯ Mucopolysaccharidosis Type I IDUA
- ◯ Mucopolysaccharidosis Type II IDS
- ◯ Mucopolysaccharidosis Type IIIA (Sanfilippo Syndrome A) SGSH
- ◯ Mucopolysaccharidosis Type IVA GALNS
- ◯ Multiple Acyl-CoA Dehydrogenase Deficiency ETFA
- ◯ Multiple Acyl-CoA Dehydrogenase Deficiency ETFB
- ◯ Multiple Acyl-CoA Dehydrogenase Deficiency ETFDH
- ◯ Multiple Intestinal Atresia TTC7A
- ◯ Muscle-Eye-Brain Disease POMGNT1
- ◯ Muscular Dystrophy-Dystroglycanopathy 9 (Limb-Girdle) Type C DAG1
- ◯ MYBPC3 -Related Disorders
- ◯ MYH7 -Related Disorders
- ◯ MYO7A-Related Disorders (Usher Syndrome 1B)
- ◯ Myoclonic Dystonia-11 SGCE
- ◯ Myopathy due to Myoadenylate Deaminase Deficiency AMPD1
- ◯ Myopathy with Deficiency of ISCU
- ◯ MYOT Related Disorders MYOT
- ◯ Myotonic Dystrophy Type 1
- ◯ Myotubular Myopathy, X-linked MTM1
- ◯ N-Acetylglutamate Synthase Deficiency NAGS
- ◯ Nail-Patella Syndrome LMX1B
- ◯ NARS2-Related Disorders
- ◯ Native American Myopathy STAC3
- ◯ NBN-Related Disorders (Nijmegen Breakage Syndrome)
- ◯ NDP-Related Disorders
- ◯ Nemaline Myopathy Amish Type 5 TNNT1
- ◯ Nemaline Myopathy, Autosomal Dominant 6 KBTBD13
- ◯ Nemaline Myopathy, Autosomal Recessive 2 NEB
- ◯ Nemaline Myopathy, Autosomal Recessive 7 CFL2
- ◯ Nemaline Myopathy, Autosomal Recessive 8 KLHL40
- ◯ Neonatal Diabetes Mellitus with Congenital Hypothyroidism GLIS3
- ◯ Nephronophthisis 2, Infantile INVS
- ◯ Nephrotic Syndrome Type 1 NPHS1
- ◯ Nephrotic Syndrome Type 2 NPHS2
- ◯ Neuroblastoma ALK
- ◯ Neuronal Ceroid Lipofuscinosis, CLN3-Related
- ◯ Neuronal Ceroid Lipofuscinosis, CLN5-Related
- ◯ NF2-Related Disorders

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- ◯ Niemann-Pick Disease Type A SMPD1
- ◯ Niemann-Pick Disease Type C NPC1
- ◯ Niemann-Pick Disease Type C NPC2
- ◯ Nijmegen Breakage Syndrome-like Disorder RAD50
- ◯ Non-Polyposis Colorectal Cancer PMS1
- ◯ Noonan Syndrome CBL
- ◯ Noonan Syndrome KRAS
- ◯ Noonan Syndrome NRAS
- ◯ Noonan Syndrome PTPN11
- ◯ Noonan Syndrome RAF1
- ◯ Noonan Syndrome RIT1
- ◯ Noonan Syndrome SOS1
- ◯ Noonan-like Syndrome SHOC2
- ◯ NPHP1-Related Disorders
- ◯ NPHP3-Related Disorders
- ◯ NPHP4-Related Disorders
- ◯ Nuclear Encoded ATPase Deficiency TMEM70
- ◯ Oculocutaneous Albinism Type 1 TYR
- ◯ Oculocutaneous Albinism Type 2 OCA2
- ◯ Oculocutaneous Albinism Type 3 TYRP1
- ◯ Oculocutaneous Albinism Type 4 SLC45A2 (OCA4)
- ◯ Oculocutaneous Albinism, X-Linked GPR143
- ◯ Oculopharyngeal Muscular Dystrophy PABPN1
- ◯ OPA3-Related Disorders
- ◯ Optic Atrophy Type 1 OPA1
- ◯ OPTN-Related Disorders
- ◯ Osteogenesis Imperfecta CRTAP
- ◯ Osteogenesis Imperfecta LEPRE1
- ◯ Osteogenesis Imperfecta Type V IFITM5
- ◯ Osteogenesis Imperfecta Type VI SERPINF1
- ◯ Osteopathia Striata with Cranial Sclerosis FAM123B
- ◯ Osteopetrosis with Renal Tubular Acidosis CA2
- ◯ Osteogenesis Imperfecta Type VI SERPINF1
- ◯ Osteogenesis Imperfecta, Type XV WNT1
- ◯ Osteopathia Striata with Cranial Sclerosis FAM123B
- ◯ Osteopetrosis with Renal Tubular Acidosis CA2
- ◯ Osteopetrosis, CLCN7-Related
- ◯ Osteopetrosis, TCIRG1-Related
- ◯ OTC Deficiency OTC
- ◯ Paraganglioma/Pheochromocytoma TMEM127
- ◯ PAX4 -Related Disorders
- ◯ PAX6-Related Disorders
- ◯ PCDH19-Related X-Linked Female-Limited Epilepsy w/MR
- ◯ PDH Complex Deficiency DLAT
- ◯ PDH Complex Deficiency PDHA1
- ◯ PDH Complex Deficiency PDHB
- ◯ PDH Complex Deficiency PDHX
- ◯ PDH Complex Deficiency PDP1
- ◯ PDX1-Related Disorders PDX1
- ◯ Pelizaeus-Merzbacher-Like Disease GJC2
- ◯ Pendred Syndrome SLC26A4 (PENDRIN)
- ◯ Permanent Neonatal Diabetes Mellitus with Cerebellar Agenesis PTF1A
- ◯ Peroxisomal Acyl-CoA Oxidase Deficiency ACOX1
- ◯ Peroxisome Biogenesis Disorder 1 PEX1 (Zellweger Spectrum Disorders)
- ◯ Peroxisome Biogenesis Disorder 2 PEX5
- ◯ Peroxisome Biogenesis Disorder 3 PEX12
- ◯ Peroxisome Biogenesis Disorder 4 PEX6
- ◯ Peroxisome Biogenesis Disorder 5 PEX2
- ◯ Peroxisome Biogenesis Disorder 6 PEX10
- ◯ Peroxisome Biogenesis Disorder 7 PEX26
- ◯ Peroxisome Biogenesis Disorder 8 PEX16
- ◯ Peroxisome Biogenesis Disorder 10A (Zellweger) PEX3
- ◯ Peroxisome Biogenesis Disorder 11 PEX13
- ◯ Peroxisome Biogenesis Disorder 12A (Zellweger) PEX19
- ◯ Peroxisome Biogenesis Disorder 13A (Zellweger) PEX14
- ◯ Peroxisome Biogenesis Disorder 14B PEX11B
- ◯ PEX7-Related Disorders (Rhizomelic Chondrodysplasia Punctata Type I)
- ◯ PGM3-Related Disorders
- ◯ Phenylalanine Hydroxylase Deficiency (Phenylketonuria) PAH
- ◯ Pheochromocytoma MAX
- ◯ Phosphoenolpyruvate Carboxykinase Deficiency, Cytosolic, PCK1
- ◯ Phosphoenolpyruvate Carboxykinase Deficiency, Mitochondrial, PCK2
- ◯ PHOX2B-Related Disorders
- ◯ PITX2-Related Disorders
- ◯ PITX3-Related Disorders
- ◯ PLEC-Related Disorders
- ◯ PLP1-Related Disorders
- ◯ POLG-Related Disorders
- ◯ POLG2-Related Disorders
- ◯ POMT1-Related Disorders
- ◯ POMT2-Related Disorders
- ◯ Polyneuropathy, Hearing Loss, Ataxia, Retinitis Pigmentosa, and Cataract Disorder ABHD12
- ◯ Prader-Willi-like Syndrome; Intellectual Disability; Autism MAGEL

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- ◯ Related Disorders
- ◯ Primary Hyperoxaluria Type 1 AGXT
- ◯ Primary Hyperoxaluria Type 2 GRHPR
- ◯ Primary Open Angle Glaucoma 1A MYOC
- ◯ PRKAR1A-Related Disorders
- ◯ PRKDC-Related Disorders
- ◯ PROP1-Related Combined Pituitary Hormone Deficiency
- ◯ Propionic Acidemia, PCCA-Related
- ◯ Propionic Acidemia, PCCB-Related
- ◯ PTCH1-Related Disorders
- ◯ PTEN-Related Disorders
- ◯ Purine Nucleoside Phosphorylase Deficiency
- ◯ Pycnodysostosis CTSK
- ◯ Pyridoxine-Dependent Seizures ALDH7A1
- ◯ Pyruvate Carboxylase Deficiency PC
- ◯ RAG2-Related Disorders
- ◯ RECQL4 -Related Disorders (Rothmund-Thomson Syndrome)
- ◯ Refsum Disease PHYH
- ◯ Reticular Dysgenesis AK2
- ◯ Retinitis Pigmentosa, ABCA4-Related
- ◯ Retinitis Pigmentosa, ABHD12-Related
- ◯ Retinitis Pigmentosa, BEST1-Related
- ◯ Retinitis Pigmentosa, C2orf71-Related
- ◯ Retinitis Pigmentosa, CA4-Related
- ◯ Retinitis Pigmentosa, CDHR1-Related
- ◯ Retinitis Pigmentosa, CEP290-Related
- ◯ Retinitis Pigmentosa, CNGB1-Related
- ◯ Retinitis Pigmentosa, CRB1-Related
- ◯ Retinitis Pigmentosa, CRX-Related
- ◯ Retinitis Pigmentosa, DHDDS-Related
- ◯ Retinitis Pigmentosa, EYS-Related
- ◯ Retinitis Pigmentosa, FAM161A-Related
- ◯ Retinitis Pigmentosa, FLVCR1-Related
- ◯ Retinitis Pigmentosa, FSCN2-Related
- ◯ Retinitis Pigmentosa, GUCY2D-Related
- ◯ Retinitis Pigmentosa, IMPDH1-Related
- ◯ Retinitis Pigmentosa, IMPG2-Related
- ◯ Retinitis Pigmentosa, LCA5-Related
- ◯ Retinitis Pigmentosa, LRAT-Related
- ◯ Retinitis Pigmentosa, MERTK-Related
- ◯ Retinitis Pigmentosa, MFRP-Related
- ◯ Retinitis Pigmentosa, NR2E3-Related
- ◯ Retinitis Pigmentosa, PDE6B-Related
- ◯ Retinitis Pigmentosa, PRKCG-Related
- ◯ Retinitis Pigmentosa, PROM1-Related
- ◯ Retinitis Pigmentosa, PRPF3-Related
- ◯ Retinitis Pigmentosa, PRPH2-Related
- ◯ Retinitis Pigmentosa, RD3-Related
- ◯ Retinitis Pigmentosa, RDH12-Related
- ◯ Retinitis Pigmentosa, RGR-Related
- ◯ Retinitis Pigmentosa, Autosomal Recessive, Bothnia Type RLBP1
- ◯ Retinitis Pigmentosa, ROM1-Related
- ◯ Retinitis Pigmentosa, RP2-Related
- ◯ Retinitis Pigmentosa, RPE65-Related
- ◯ Retinitis Pigmentosa, RPGR-Related
- ◯ Retinitis Pigmentosa, RPGRIP1-Related
- ◯ Retinitis Pigmentosa, SAG-Related
- ◯ Retinitis Pigmentosa, TOPORS-Related
- ◯ Retinoschisis RS1
- ◯ Rett Syndrome, Congenital Variant FOXG1
- ◯ Rhizomelic Chondrodysplasia Punctata Type 2 GNPAT
- ◯ Rhizomelic Chondrodysplasia Punctata Type 3 AGPS
- ◯ RMRP-Related Disorders (Cartilage Hair Hypoplasia)
- ◯ RYR1-Related Disorders
- ◯ RYR2-Related Disorders
- ◯ Rubinstein-Taybi Syndrome CREBBP
- ◯ Salla Disease SLC17A5 (NSD)
- ◯ Sandhoff Disease HEXB
- ◯ SCAD Deficiency ACADS
- ◯ Schmid Metaphyseal Chondrodysplasia (SMCD) COL10A1
- ◯ SCN4A-Related Disorders
- ◯ Selective T-cell Defect ZAP70
- ◯ SEPN1-Related Disorders
- ◯ SERPINA1-Related Disorders SERPINA1
- ◯ Severe Combined Immunodeficiency, Athabascan type DCLRE1C
- ◯ Severe Combined Immunodeficiency, X-Linked IL2RG
- ◯ Severe Combined Immunodeficiency JAK3
- ◯ Severe Combined Immunodeficiency NHEJ1
- ◯ Severe Combined Immunodeficiency PTPRC
- ◯ Severe Combined Immunodeficiency RAG1
- ◯ SGCD-Related Disorders
- ◯ Shwachman-Bodian-Diamond Syndrome SBDS
- ◯ Sjogren-Larsson Syndrome ALDH3A2
- ◯ SLC16A1

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- ◯ SLC25A4/ANT1-Related Disorders
- ◯ SMAD4 -Related Disorders
- ◯ Smith-Lemli-Opitz Syndrome DHCR7
- ◯ Smith-Magenis Syndrome RAI1
- ◯ Spastic Paraplegia 7, Autosomal Recessive SPG7
- ◯ Spinocerebellar Ataxia 1 SCA1
- ◯ Spinocerebellar Ataxia 10 SCA10
- ◯ Spinocerebellar Ataxia 14 PRKCG
- ◯ SRD5A3-Related Disorders
- ◯ STAT5B-Related Disorders
- ◯ STIM1-Related Disorders
- ◯ STK11-Related Disorders
- ◯ Succinic Semialdehyde Dehydrogenase Deficiency ALDH5A1
- ◯ SUCLG1-Related Disorders
- ◯ SYNE1-Related Disorders
- ◯ Tay-Sachs Disease (Hexosaminidase A Deficiency) HEXA
- ◯ TCAP-Related Disorders
- ◯ T-cell Immunodeficiency, Congenital Alopecia, and Nail Dystrophy FOXN1
- ◯ TMEM43-Related Disorders
- ◯ TMEM67-Related Disorders
- ◯ TMLHE Deficiency
- ◯ TPM2-Related Disorders
- ◯ TPM3-Related Disorders
- ◯ TTN-Related Disorders
- ◯ Transcobalamin II Deficiency TCN2
- ◯ TRIM32-Related Disorders
- ◯ TSHR-Related Disorders TSHR
- ◯ TUSC3-Related Disorders
- ◯ Tyrosine Hydroxylase Deficiency TH
- ◯ Tyrosinemia Type I FAH
- ◯ Tyrosinemia Type II TAT
- ◯ Usher Syndrome 1C USH1C
- ◯ Usher Syndrome 1F PCDH15
- ◯ USH2A-Related Disorders (Usher Syndrome 2A; Retinitis Pigmentosa)
- ◯ Usher Syndrome 2C GPR98
- ◯ Usher Syndrome 2D DFNB31
- ◯ VCP-Related Disorders
- ◯ VLCAD Deficiency ACADVL
- ◯ Von Hippel-Lindau Syndrome VHL
- ◯ VSX1-Related Disorders
- ◯ Welander Distal Myopathy TIA1
- ◯ WFS1-Related Disorders
- ◯ Wolfram Syndrome 2 CISD2
- ◯ Wolman Disease LIPA
- ◯ Wilson Disease ATP7B
- ◯ WT1-Related Disorders

BAYLOR GENETICS

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CONNECT

INFORMED CONSENT FOR PRENATAL COMPREHENSIVE TESTING

Fetus of: _____ (Patient Last Name) _____ (Patient First Name) _____ (MI) _____ / _____ / _____ (Date of Birth MM / DD / YYYY) _____ (Biological Sex)

INFORMED CONSENT FOR GENETIC TESTING

TEST INFORMATION

This consent form will provide you with information regarding genetic testing, which you should discuss with your healthcare provider or a genetic counselor. In order to ensure that you have understood the purpose and significance of genetic testing, we have provided information about the testing process and potential results below.

The purpose of genetic testing is to identify the cause of a suspected disease in you or your family. The testing analyzes your genetic material (DNA) for an abnormal change (variant) that could explain the disease you or members of your family are experiencing. Genetic testing can be a diagnostic test, which is used to identify or rule out a specific genetic condition. Genetic screening tests are used to assess the chance for a person to develop or have a child with a genetic condition. Genetic screening tests are not typically diagnostic, and results may require additional testing.

The purpose of this test is to see if you or your child may have a genetic variant or chromosome rearrangement. This may cause a genetic disorder or may determine the chance that you or your child will develop or pass on a genetic disorder in the future. "Your child" can also mean your unborn child, for the purposes of this consent.

In a genetic test, depending on the case, you can be tested for:

- A single gene/variant responsible for a specific, suspected genetic disease.
- Multiple genes in parallel.

The sample/specimen that is needed to perform the genetic test is stated in the test order form and is typically blood or purified DNA, but may also be tissue, saliva or buccal swab.

RESULTS

There are several categories of test results that may be reported including:

- **Positive:** Positive or "abnormal" results mean there is a change in the genetic material found that is related to your/your child's medical issues or that you/your child are at an increased risk of developing the disorder in the future. It is possible to test positive for more than one genetic variant.
- **Negative:** Negative or "normal" results mean no relevant genetic change related to your/your child's medical issues was detected. This does not mean there is no genetic change, but it may mean that the type of testing performed could not detect it.
- **Results of Unclear Significance:** Testing can detect change(s) in DNA which we do not yet fully understand. These alterations are also referred to as variants of uncertain significance (VUS). Additional studies may be recommended if a VUS is identified in a gene that may be associated with your/your child's medical concerns.
- **Secondary / Incidental Findings:** Testing can sometimes detect a change in a person's DNA unrelated to the reason for testing. If this change has medical or reproductive significance, it is called a secondary or incidental finding.

CONSIDERATIONS AND LIMITATIONS

- Results may indicate affected status, increased risk to someday be affected with, and/or reproductive risk for a genetic disorder. It is important to understand that genetic tests, even if negative, are not exhaustive. It is not possible to exclude risks for all possible genetic diseases for yourself and your family members.
- A positive test result is an indication that the individual(s) being tested may be predisposed to or have the specific disease or condition which prompted testing. You might consider additional independent testing, consult a personal physician, or pursue genetic counseling.
- It is possible that the knowledge of the test results may result in psychological stress for you and your family. It is always recommended to discuss the results with your healthcare provider or genetic counselor.
- If several family members are tested, the correct interpretation of the results depends on the provided relationships between family members. In rare cases, genetic testing can reveal that the true biological relationships in a family are not as they were reported. If a discrepancy is identified, it may be necessary to report this to the physician who ordered the testing.
- Genetic testing is highly accurate. Rarely, inaccurate results may occur for various reasons. These reasons include, but are not limited to, mislabeled samples, inaccurate reporting of clinical/medical information, or rare technical errors.
- If you sign this consent form, but you no longer wish to have your sample(s) tested, you can contact your physician to cancel the test. If testing is complete, but you have not received your results yet, you can inform your physician that you no longer wish to receive the results. If you withdraw consent for testing after 5pm CST the next business day following sample receipt by the laboratory, you will be charged for the full cost of the test.

PATIENT CONFIDENTIALITY AND SPECIMEN RETENTION

- Results will only be released to a licensed healthcare provider, to those allowed access to test results by law, and to those authorized in writing.
- In rare cases, persons with genetic diagnoses have experienced problems with insurance coverage and employment. The U.S. Federal Government has enacted several laws that prohibit discrimination based on genetic test results by health insurance companies and employers. In addition, these laws prohibit unauthorized disclosure of this information. For more information, you can visit www.genome.gov/10002077.
- Samples will be retained in the laboratory in accordance with the laboratory retention policy.
- After testing is complete, the de-identified submitted specimen may be used for test development and improvement, internal validation, quality assurance, and training purposes. DNA specimens are not returned to individuals or to referring heath care providers unless specific prior arrangements have been made.

INFORMED CONSENT FOR PRENATAL COMPREHENSIVE TESTING

Fetus of: _____ (Patient Last Name) _____ (Patient First Name) _____ (MI) _____ / _____ / _____ (Date of Birth (MM / DD / YYYY)) _____ (Biological Sex)

INFORMED CONSENT FOR GENETIC TESTING

PATIENT CONFIDENTIALITY AND SPECIMEN RETENTION (CONT.)

- Samples from residents of New York State will not be included in the de-identified research studies described in this authorization and will not be retained for more than 60 days after test completion, unless specifically authorized by your selection. No tests other than those authorized shall be performed on the biological sample.
- Information including results, indications for testing and clinical status obtained from this testing may be shared with healthcare providers, scientists and healthcare databases or used in scientific publications or presentations, but the personal identifying information of all persons studied will not be revealed in such data sharing or publications/presentations.

RESEARCH & RECONTACT CONSENT

For more information on research at Baylor Genetics, please visit baylorgenetics.com. Please read the below statements carefully and check the appropriate box.

Note: If left blank, consent is interpreted as "NO."

☐ I agree to use of my de-identified specimen for research to improve genetic testing for all patients and contribute to scientific research.

☐ I am a New York State Resident, and I give Baylor Genetics permission to store my specimen in accordance to the laboratory retention policy for internal quality assurance and possible research studies.

☐ In addition to agreeing above, I agree to be contacted by Baylor Genetics regarding research opportunities.

PATIENT AUTHORIZATION

By signing this statement of consent, I acknowledge that I have read and understand the informed consent for genetic testing. I have received appropriate explanations from my physician regarding the purpose, scope, type and significance of the planned genetic testing and achievable results. All my questions have been answered and I have had the necessary time to make an informed decision about the genetic test.

I give permission to Baylor Genetics to conduct genetic testing as recommended by my physician.

_____ (Patient Signature) _____ (Date (DD/MM/YYYY))

_____ (Printed Name)