Single-Cell Genetic Disorders

This list of single-cell genetic disorders serves as a preliminary guide for prospective COTA patients. If your patient has a single-cell genetic disorder that is not listed, please contact COTA to discuss potential eligibility.

ACHONDROPLASIA; ACH
ACYL-CoA DEHYDROGENASE, MEDIUM-CHAIN, DEFICIENCY
ACYL-CoA DEHYDROGENASE, VERY LONG-CHAIN; ACADVL
ADENOMATOUS POLYPOSIS OF THE COLON; APC
ADENOSINE DEAMINASE DEFICIENCY; ADA
ADRENOLEUKODYSTROPHY; ALD
AGAMMAGLOBULINEMIA, X-LINKED; XLA
AICARDI-GOUTIERES SYNDROME 1; AGS1
AICARDI-GOUTIERES SYNDROME 5; AGS5
ALBINISM, OCULAR, TYPE I; OA1
ALOPECIA UNIVERSALIS CONGENITA; ALUNC
ALPERS DIFFUSE DEGENERATION OF CEREBRAL GRAY MATTER WITH HEPATIC CIRRHOSIS
ALPHA 1 ANTITRYPSIN DEFICIENCY AAT)
ALPORT SYNDROME, X-LINKED; ATS
AMMECR1
ALZHEIMER DISEASE 4
AMYLOIDOSIS I, HEREDITARY NEUROPATHIC
AMYOTROPHIC LATERAL SCLEROSIS 1; ALS1
ANDROGEN RECEPTOR; AR (testicular feminization; spinal and bulbar muscular atrophy; Kennedy disease)
ANEUPLOIDIES BY STR GENOTYPING
ANGIOEDEMA, HEREDITARY; HAE
ARGININOSUCCINIC ACIDURIA
ASL
ARTHROGRYPOSIS, DISTAL, TYPE 2B; DA2B
ATAXIA-TELANGIECTASIA; AT
BASAL CELL NEVUS SYNDROME; BCNS (GORLIN)
BETA-HYDROXYISOBUTYRYL CoA DEACYLASE, DEFICIENCY
BLEPHAROPHIMOSIS, PTOSIS, AND EPICANTHUS INVERSUS; BPES
BLOOD GROUP–KELL-CELLANO SYSTEM
BRACHYDACTYLY, TYPE B1; BDB1
BRAIN TUMOR, POSTERIOR FOSSA OF INFANCY, FAMILIAL
BREAST-OVARIAN CANCER, FAMILIAL
BREAST-OVARIAN CANCER, FAMILIAL

CANAVAN DISEASE
CARDIOENCEPHALOMYOPATHY, FATAL INFANTILE, DUE TO CYTOCHROME c OXIDASE DEFICIENCY
CARDIOMYOPATHY, DILATED, 1A; CMD1A
CARDIOMYOPATHY, DILATED, 1DD; CMD1DD
CARDIOMYOPATHY, FAMILIAL HYPERTROPHIC, 4; CMH4
CARDIOMYOPATHY, FAMILIAL HYPERTROPHIC, 7; CMH7
CARNITINE DEFICIENCY, SYSTEMIC PRIMARY; CDSP
CARNITINE DEFICIENCY, SYSTEMIC PRIMARY; CDSP
CEROID LIPOFUSCINOSIS, NEURONAL 2, LATE INFANTILE; CLN2
CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2E
CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 1A; CMT1A
CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 1B; CMT1B
CHARCOT-MARIE-TOOTH DISEASE, X-LINKED, 1; CMTX1
CHOLESTASIS, PROGRESSIVE FAMILIAL INTRAHEPATIC 2
CHONDRODYSPLASIA PUNCTATA 1, X-LINKED RECESSIVE; CDPX1
CHOROIDEREMIA; CHM
CILIARY DYSKINESIA, PRIMARY, 3; CILD3
CITRULLINEMIA, CLASSIC
COHEN SYNDROME; COH1
COLLAGEN, TYPE IV, ALPHA-5; COL4A5
COLORECTAL CANCER, HEREDITARY NONPOLYPOSIS, TYPE 1; HNPCC1
COLORECTAL CANCER, HEREDITARY NONPOLYPOSIS, TYPE 1; HNPCC1
COLORECTAL CANCER, HEREDITARY NONPOLYPOSIS, TYPE 2; HNPCC2
CONGENITAL ADRENAL HYPERPLASIA (CAH)
CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Ia; CDG1A
CORNEAL DYSTROPHY, AVELLINO TYPE; CDA
CRANIOFACIAL DYSOSTOSIS, TYPE I; (CFD1)
CURRARINO SYNDROME
CUTIS LAXA, AUTOSOMAL RECESSIVE, TYPE I
CYSTIC FIBROSIS; CF
CYSTINOSIS, NEPHROPATHIC; CTNS
DARIER-WHITE DISEASE; DAR

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D-BIFUNCTIONAL PROTEIN DEFICIENCY
DEAFNESS, NEUROSENSORY, AUTOSOMAL RECESSIVE 1; DFNB1
DIAMOND-BLACKFAN ANEMIA; DBA
DIAMOND-BLACKFAN ANEMIA; DBA
DIHYDROXYADENINE UROLITHIASIS
DONOHUE SYNDROME
DYSKERATOSIS CONGENITA, AUTOSOMAL DOMINANT, 1; DKCA1
DYSTONIA 1, TORSION, AUTOSOMAL DOMINANT; DYT1
DYSTROPHIA MYOTONICA 1
EARLY-ONSET FAMILIAL ALZHEIMER DISEASE;
ECTODERMAL DYSPLASIA, ANHIDROTIC
ECTODERMAL DYSPLASIA, HYPOHIDROTIC, X-LINKED
ECTRODACTYLY, ECTODERMAL DYSPLASIA, AND CLEFT LIP/ PALATE SYNDROME 1; EEC1
EHLERS-DANLOS SYNDROME, TYPE I
EHLERS-DANLOS SYNDROME, TYPE IV
EHLERS-DANLOS SYNDROME, TYPE VI
EHLERS-DANLOS SYNDROME, TYPE VIIC
EMERY-DREIFUSS MUSCULAR DYSTROPHY, AUTOSOMAL RECESSIVE; EDMD3
EMERY-DREIFUSS MUSCULAR DYSTROPHY, X-LINKED; EDMD
EPIDERMOLYSIS BULLOSA DYSTROPHICA, PASINI TYPE
EPIDERMOLYSIS BULLOSA LETALIS
EPIDERMOLYSIS BULLOSA SIMPLEX WITH PYLORIC ATRESIA
EPIDERMOLYSIS BULLOSA, JUNCTIONAL, HERLITZ TYPE
EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 2
EPIPHYSEAL DYSPLASIA, MULTIPLE, 1; EDM1
EXOSTOSES, MULTIPLE, TYPE I
FABRY DISEASE
FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY 1A; FSHMD1A
FAMILIAL MEDITERRANEAN FEVER GENE; MEFV
FANCONI ANEMIA, COMPLEMENTATION GROUP C; FANCC
FANCONI ANEMIA, COMPLEMENTATION GROUP D2; FANCD2
FANCONI ANEMIA, COMPLEMENTATION GROUP E; FANCE
FANCONI ANEMIA, COMPLEMENTATION GROUP F; FANCF
FANCONI ANEMIA, COMPLEMENTATION GROUP G
FANCONI ANEMIA, COMPLEMENTATION GROUP I; FANCI
FANCONI ANEMIA, COMPLEMENTATION GROUP J
FANCONY ANEMIA, COMPLEMENTATION GROUP A; FANCA
FRAGILE SITE MENTAL RETARDATION 1

FRAGILE SITE, FOLIC ACID TYPE, RARE, FRA(X)(q28); FRAXE
FRASER SYNDROME
FRIEDREICH ATAXIA 1; FRDA
GALACTOSEMIA
GANGLIOSIDOSIS, GENERALIZED GM1, TYPE I
GASTRIC CANCER, HEREDITARY DIFFUSE; HDGC
GAUCHER DISEASE, TYPE I
GERODERMA OSTEODYSPLASTICUM; GO
GERSTMANN-STRAUSSLER DISEASE; GSD
GLAUCOMA 3, PRIMARY CONGENITAL, A; GLC3A
GLUCOSE TRANSPORT DEFECT, BLOOD-BRAIN BARRIER
GLUCOSE-6-PHOSPHATE DEHYDROGENASE; G6PD
GLUTARIC ACIDEMIA I
GLYCINE ENCEPHALOPATHY; GCE
GLYCOGEN STORAGE DISEASE I
GLYCOGEN STORAGE DISEASE II
GLYCOGEN STORAGE DISEASE TYPE VI
GRANULOMATOUS DISEASE, CHRONIC, X-LINKED; CGD
GRISCELLI SYNDROME WITH HEMOPHAGOCYTIC SYN-DROME;TYPE 2; GS2, TYPE 2; GS2
HEMOCHROMATOSIS; HFE
HEMOGLOBIN–ALPHA LOCUS 1; HBA1
HEMOGLOBIN–ALPHA LOCUS 2; HBA2
HEMOGLOBIN–BETA LOCUS; HBB
HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, FAMILIAL, 2
HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, FAMILIAL, 3; FHL3
HEMOPHILIA A
HEMOPHILIA B
HEREDITARY MOTOR AND SENSORY NEUROPATHY VI
HLA MATCHING GENOTYPING
HOLT-ORAM SYNDROME; HOS
HOMOCYSTINURIA DUE TO DEFICIENCY OF N(5,10)-METHYLENETETRAHYDROFOLATE REDUCTASE ACTIVITY
HOYERAAL-HREIDARSSON SYNDROME; HHS
HUNTINGTON DISEASE; HD
HURLER SYNDROME
HYALINOSIS, INFANTILE SYSTEMIC
HYDROCEPHALUS, X-LINKED; L1CAM
HYPERGLYCINEMIA, NONKETOTIC; NKH
HYPER-IgE RECURRENT INFECTION (JOB'S) SYNDROME

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HYPERINSULINEMIC HYPOGLYCEMIA, FAMILIAL, 1; HHF1
HYPOMAGNESEMIA, RENAL, WITH OCULAR INVOLVEMENT
HYPOPHOSPHATASIA, INFANTILE
HYPOPHOSPHATEMIC RICKETS, X-LINKED DOMINANT
ICHTHYOSIS FOLLICULARIS, ATRICHIA, AND PHOTOPHOBIA SYNDROME
ICHTHYOSIS, LAMELLAR, 1; LI1
ICHTHYOSIS, LAMELLAR, 2; LI2
IMMUNODEFICIENCY DUE TO DEFECT IN CD3-ZETA
IMMUNODEFICIENCY WITH HYPER-IgM, TYPE 1; HIGM1
IMMUNODYSREGULATION, POLYENDOCRINOPATHY, AND ENTEROPATHY, X-LINKED; IPEX
INCONTINENTIA PIGMENTI; IP
ISOVALERIC ACIDEMIA; IVA
JOUBERT SYNDROME 3; JBTS3
JOUBERT SYNDROME 6; JBTS6
KALLMANN SYNDROME 2
KRABBE DISEASE
LEBER CONGENITAL AMAUROSIS 2; LCA2
LEBER CONGENITAL AMAUROSIS 6
LEIGH SYNDROME; LS
LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER; VWM
LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER; VWM
LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER; VWM
LI-FRAUMENI SYNDROME 1; LFS1
LIPOID CONGENITAL ADRENAL HYPERPLASIA
LOEYS-DIETZ SYNDROME; LDS
LONG-CHAIN 3-HYDROXYACYL-CoA DEHYDROGENASE DEFICIENCY;HADHA
MACHADO-JOSEPH DISEASE; MJD
MARFAN SYNDROME; MFS
METACHROMATIC LUEKODYSTROPY
METAPHYSEAL CHONDRODYSPLASIA, SCHMID TYPE; MCDS
MICROCORIA-CONGENITAL NEPHROSIS SYNDROME
MICROPHTHALMIA, ISOLATED 2; MCOP2
MICROTUBULE-ASSOCIATED PROTEIN TAU; MAPT
MIGRAINE, FAMILIAL HEMIPLEGIC, 1; FHM1
MORQUIO SYNDROME, NONKERATOSULFATE-EXCRETING TYPE
MUCOPOLYSACCHARIDOSIS TYPE II (HUNTER)
HUNTER-MCALPINE CRANIOSYNOSTOSIS SYNDROME
MUCOPOLYSACCHARIDOSIS TYPE IIIA

MUCOPOLYSACCHARIDOSIS TYPE VI
MULTIPLE ACYL-CoA DEHYDROGENASE DEFICIENCY; MADD
MULTIPLE ENDOCRINE NEOPLASIA, TYPE I; MEN1
MULTIPLE ENDOCRINE NEOPLASIA, TYPE IIA; MEN2A
MUSCULAR DYSTROPHY, BECKER TYPE; BMD
MUSCULAR DYSTROPHY, CONGENITAL MEROSIN-DEFICIENT, 1A; MDC1A
MUSCULAR DYSTROPHY, DUCHENNE TYPE; DMD
MYOCLONIC EPILEPSY OF LAFORA
MYOPATHY, MYOFIBRILLAR, DESMIN-RELATED
MYOTONIA CONGENITA, AUTOSOMAL DOMINANT
MYOTUBULAR MYOPATHY 1; MTM1
N-ACETYLGLUTAMATE SYNTHASE DEFICIENCY
NAIL-PATELLA SYNDROME; NPS
NEPHROGENIC SYNDROME OF INAPPROPRIATE ANTIDIURESIS
NEPHROSIS 1, CONGENITAL, FINNISH TYPE; NPHS1
NEURAMINIDASE DEFICIENCY
NEUROFIBROMATOSIS, TYPE I; NF1
NEUROFIBROMATOSIS, TYPE II; NF2
NEUROPATHY, HEREDITARY SENSORY AND AUTONOMIC, TYPE I; HSAN1
NEUROPATHY, HEREDITARY SENSORY AND AUTONOMIC, TYPE III; HSAN3
NIEMANN-PICK DISEASE, TYPE A
NIEMANN-PICK DISEASE, TYPE C1; NPC1
NOONAN SYNDROME 1; NS1
NOONAN SYNDROME 3; NS3
NOONAN SYNDROME 4; NS4
NORRIE DISEASE; NDP
OCULOCUTANEOUS ALBINISM, TYPE I; OCA1
OCULOCUTANEOUS ALBINISM, TYPE II; OCA2
OMENN SYNDROME
OPTIC ATROPHY 1; OPA1
ORNITHINE TRANSCARBAMYLASE DEFICIENCY
OSTEOGENESIS IMPERFECTA CONGENITA; OIC
OSTEOGENESIS IMPERFECTA, TYPE IX
OSTEOPETROSIS, AUTOSOMAL RECESSIVE
PACHYGYRIA WITH MENTAL RETARDATION, SEIZURES
PANCREATITIS, HEREDITARY; PCTT
PELIZAEUS-MERZBACHER-LIKE DISEASE; PMLD
PERIODIC FEVER, FAMILIAL, AUTOSOMAL DOMINANT

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PEUTZ-JEGHERS SYNDROME; PJS
PFEIFFER SYNDROME
PHENYLKETONURIA
POLYCYSTIC KIDNEY DISEASE 1; PKD1
POLYCYSTIC KIDNEY DISEASE 2; PKD2
POLYCYSTIC KIDNEY DISEASE, AUTOSOMAL RECESSIVE; ARPKD
POLYCYSTIC LIVER DISEASE
POPLITEAL PTERYGIUM SYNDROME; PPS
PROPIONIC ACIDEMIA
PROSAPOSIN DEFICIENCY; PSAPD
PSEUDOHYPOPARATHYROIDISM, TYPE IA; PHP1A
PYRIDOXAMINE 5-PRIME-PHOSPHATE OXIDASE DEFICIENCY
PYRUVATE KINASE DEFICIENCY OF RED CELLS
RETINITIS PIGMENTOSA
RETINITIS PIGMENTOSA 3; RP3
RETINOBLASTOMA; RB1
RETINOSCHISIS 1, X-LINKED, JUVENILE; RS1
RETT SYNDROME; RTT
RHESUS BLOOD GROUP, CcEe ANTIGENS; RHCE
RHESUS BLOOD GROUP, D ANTIGEN; RHD
SAETHRE-CHOTZEN SYNDROME; SCS
SANDHOFF DISEASE
SEVERE COMBINED IMMUNODEFICIENCY
SEVERE COMBINED IMMUNODEFICIENCY, X-LINKED; SCIDX1
SHWACHMAN-DIAMOND SYNDROME; SDS
SICKLE CELL ANEMIA
SMITH-LEMLI-OPITZ SYNDROME; SLOS
SONIC HEDGEHOG; SHH
SOTOS SYNDROME
SPASTIC PARAPLEGIA 3, AUTOSOMAL DOMINANT
SPASTIC PARAPLEGIA 4, AUTOSOMAL DOMINANT; SPG4
SPINAL MUSCULAR ATROPHY, DISTAL, AUTOSOMAL RECESSIVE
SPINAL MUSCULAR ATROPHY, TYPE I; SMA1
SPINOCEREBELLAR ATAXIA 2; SCA2
SPINOCEREBELLAR ATAXIA 7; SCA7
SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 1
STICKLER SYNDROME, TYPE I; STL1
STICKLER SYNDROME, TYPE II; STL2
SUCCINIC SEMIALDEHYDE DEHYDROGENASE DEFICIENCY
SURFACTANT METABOLISM DYSFUNCTION, PULMONARY, 3; SMDP3
SYMPHALANGISM, PROXIMAL; SYM1
TAY-SACHS DISEASE; TSD
THROMBASTHENIA OF GLANZMANN AND NAEGELI
THROMBOTIC THROMBOCYTOPENIC PURPURA, CONGENITAL; TTP
TORSION DYSTONIA 1, AUTOSOMAL DOMINANT; DYT1
TREACHER COLLINS-FRANCESCHETTI SYNDROME; TCOF
TRIFUNCTIONAL PROTEIN DEFICIENCY
TUBEROUS SCLEROSIS TYPE 1
TUBEROUS SCLEROSIS TYPE 2
TYROSINEMIA, TYPE I
ULNAR-MAMMARY SYNDROME; UMS
VON HIPPEL-LINDAU SYNDROME; VHL
WAARDENBURG SYNDROME, TYPE 2A; WS2A
WISKOTT-ALDRICH SYNDROME; WAS
WOLFRAM SYNDROME 1; WFS1
WOLMAN DISEASE
ZELLWEGER SYNDROME; ZS
ZELLWEGER SYNDROME; ZS
ZELLWEGER SYNDROME; ZS

Source: Reproductive Genetics Institute

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