The 100,000 Genomes Project – Current Rare Disease List

v1.8.1, November 2017

Summary:

Diseases live for recruitment 216

Category	Subcategory	Disease	Status
Cardiovascular disorders (10950)	Arteriopathies (33332)	Familial cerebral small vessel disease (36469)	Live
		Familial Hypercholesterolaemia (33666)	Live
		Severe hypertriglyceridaemia (42185)	Live
	Connective Tissues Disorders and Aortopathies (10951)	Familial Thoracic Aortic Aneurysm Disease (11021)	Live
	Cardiac arrhythmia (10952)	Brugada syndrome (11022)	Live
		Long QT syndrome (11023)	Live
		Catecholaminergic Polymorphic Ventricular Tachycardia (11024)	Live
		Unexplained sudden death in the young (38566)	Live
		Idiopathic ventricular fibrillation (42161)	Live
		Short QT syndrome (55487)	Live
	Cardiomyopathy (10953)	Arrhythmogenic Right Ventricular Cardiomyopathy (11025)	Live
		Left Ventricular Noncompaction Cardiomyopathy (15044)	Live
		Dilated Cardiomyopathy (31340)	Live
		Dilated Cardiomyopathy and conduction defects (11027)	Live
		Hypertrophic Cardiomyopathy (11028)	Live
	Congenital heart disease (10954)	Familial congenital heart disease (42212)	Live
		Syndromic congenital heart disease (42213)	Live
	Lymphatic disorders (33334)	Meige disease (34328)	Live
		Milroy disease (37604)	Live
		Lymphoedema distichiasis (37612)	Live
		Lipoedema disease (55456)	Live
		Primary lymphoedema (55517)	Live
	Pulmonary heart disease (55662)	Pulmonary arterial hypertension (55499)	Live
Ciliopathies (10963)	Congenital malformations caused by ciliopathies (15091)	Bardet-Biedl Syndrome (11046)	Live
		Joubert syndrome (36478)	Live
		Rare multisystem ciliopathy disorders (36488)	Live

	Respiratory ciliopathies (15092)	Primary ciliary dyskinesia (11047)	Live
		Non-CF bronchiectasis (11048)	Live
Dermatological disorders (10956)	Atopy (15084)	Severe multi-system atopic disease with high IgE (15085)	Live
	Autoimmune skin disorders (33336)	Generalised pustular psoriasis (33646)	Live
	Ectodermal dysplasias (33338)	Ectodermal dysplasia without a known gene mutation (33699)	Live
	Ichthyoses (33340)	Autosomal recessive congenital ichthyosis (33700)	Live
	Keratodermas (33342)	Palmoplantar keratoderma and erythrokeratodermas (33701)	Live
		Familial disseminated superficial actinic porokeratosis (37644)	Live
	Neurocutaneous disorders (33344)	Undiagnosed neurocutaneous disorders (33686)	Live
	Skin adnexa disorders (36587)	Familial cicatricial alopecia (36588)	Live
		Familial hidradenitis suppurativa (41844)	Live
		Non-syndromic hypotrichosis (36849)	Live
	Skin fragility disorders (33346)	Epidermolysis bullosa (33684)	Live
		Peeling skin syndrome (36540)	Live
	Sun-exposure related conditions (10958)	Erythropoietic protoporphyria, mild variant (11037)	Live
		Hydroa vacciniforme (15083)	Live
Dysmorphic and congenital abnormality syndromes (10959)	Kabuki (28664)	Kabuki syndrome (10960)	Live
	RASopathies (10961)	Noonan syndrome (11039)	Live
		Noonan syndrome plus other features (11040)	Live
		Cardio-facio-cutaneous syndrome (11041)	Live
		LEOPARD syndrome (11042)	Live
		Costello syndrome (11043)	Live
		Legius syndrome (11044)	Live
	Balanced translocations (10962)	Balanced translocations with an unusual phenotype (11045)	Live
	Limb disorders (15087)	VACTERL-like phenotypes (10964)	Live
	DNA repair disorders (10965)	Cockayne syndrome (36497)	Live
		Non-Fanconi anaemia (11050)	Live
		Xeroderma Pigmentosum-like disorders (15089)	Live
		Primary Microcephaly - Microcephalic Dwarfism Spectrum (36505)	Live
	Autophagy disorders (10966)	Vici Syndrome and other autophagy disorders (11051)	Live
	Dysmorphic disorders (36595)	Coarse facial features including Coffin-Siris-like disorders (36596)	Live

		Familial non-syndromic cleft lip and or familial cleft palate (37565)	Live
		Syndromic cleft lip and or cleft palate (37573)	Live
		PHACE(S) syndrome (37578)	Live
		Radial dysplasia (37636)	Live
	Fetal disorders (38586)	Fetal hydrops (37586)	Live
		Unexplained monogenic fetal disorders (38665)	Live
Endocrine disorders (10967)	Adrenal disorders (10969)	Congenital adrenal hypoplasia (11053)	Live
	Disorders of calcium homeostasis (10970)	Familial or syndromic hypoparathyroidism (11054)	Live
	Gonadal and sex development disorders (36923)	Disorders of sex development (36852)	Live
		Early onset familial premature ovarian insufficiency (36851)	Live
	Growth hormone disorders (10971)	IUGR and IGF abnormalities (11057)	Live
	Hypothalamic and pituitary disorders (42204)	Idiopathic hypogonadotropic hypogonadism (41827)	Live
	Obesity syndromes (10973)	Significant early-onset obesity with or without other endocrine features and short stature (11060)	Live
	Rare subtypes of diabetes (15099)	Familial young-onset non-insulin-dependent diabetes (15103)	Live
		Hyperinsulinism (15105)	Live
		Neonatal diabetes (diagnosed less than 6 months) (30553)	Live
		Diabetes with additional phenotypes suggestive of a monogenic aetiology (30559)	Live
		Insulin resistance (including lipodystrophy) (30561)	Live
		Multi-organ autoimmune diabetes (30563)	Live
	Thyroid disorders (42208)	Congenital hypothyroidism (41908)	Live
		Resistance to thyroid hormone (41916)	Live
Gastroenterological disorders (38581)	Gastrointestinal disorders (38582)	Infantile enterocolitis and monogenic inflammatory bowel disease (37490)	Live
		Gastrointestinal epithelial barrier disorders (37772)	Live
		Non-syndromic familial congenital anorectal malformations (41868)	Live
		Early onset or familial intestinal pseudo obstruction (41876)	Live
		Familial Hirschsprung Disease (55463)	Live
	Liver disease (55663)	Ductal plate malformation (55469)	Live
		Neonatal cholestasis (71744)	Live

Growth disorders (10974)	Beckwith-Wiedemann syndrome (BWS) and other congenital overgrowth disorders (10975)	Classical Beckwith-Wiedemann syndrome (11063)	Live
		Atypical Beckwith-Wiedemann syndrome (11064)	Live
		Simpson-Golabi-Behmel syndrome (11065)	Live
		Sotos syndrome (11066)	Live
		Weaver syndrome (11067)	Live
	Growth restriction (38585)	Silver Russell syndrome (37553)	Live
Haematological and immunological disorders (10977)	Anaemias and red cell disorders (10979)	Congenital anaemias (11075)	Live
		Hereditary erythrocytosis (55505)	Live
	Primary immunodeficiency disorders (10978)	Primary immunodeficiency (55674)	Live
	Haemostasis disorders (55664)	Inherited bleeding and or platelet disorders (55475)	Live
		Monogenic venous thrombosis (55523)	Live
	Myeloid and marrow failure disorders (71739)	Cytopenia and pancytopenia (71752)	Live
Hearing and ear disorders (10980)	Non-syndromic hearing loss (10981)	Congenital hearing impairment (11076)	Live
		Auditory Neuropathy Spectrum Disorder (30607)	Live
		Autosomal dominant deafness (36848)	Live
	Deafness and congenital structural abnormalities (10982)	Bilateral microtia (11077)	Live
		Familial hemifacial microsomia (37649)	Live
		Ear malformations with hearing impairment (37657)	Live
	Other hearing and ear disorders (71738)	Familial Meniere Disease (71748)	Live
Infectious diseases (42209)	Bacterial disorders (42210)	Disseminated non-tuberculous mycobacterial infection (41932)	Live
	Sepsis (55671)	GAinS study (55665)	Live
Metabolic disorders (10983)	Specific metabolic abnormalities (10984)	Ketotic hypoglycaemia (11080)	Live
		Lactic acidosis (11081)	Live
		Cerebral folate deficiency (11083)	Live
		Undiagnosed metabolic disorders (37620)	Live
		Congenital disorders of glycosylation (37628)	Live
	Urea Cycle disorders (15108)	Hyperammonaemia (11079)	Live
	Lysosomal storage disorders (10985)	Mucopolysaccharideosis, Gaucher, Fabry (11084)	Live
	Mitochondrial (10986)	Mitochondrial disorders (11085)	Live
	Peroxisomal disorders (10987)	Peroxisomal biogenesis disorders (11086)	Live

		Other peroxisomal disorders (15109)	Live
Neurology and neurodevelopmental disorders (10988)	Motor Disorders of the CNS (10989)	Cerebellar hypoplasia (36512)	Live
		Hereditary ataxia (11087)	Live
		Early onset dystonia (11088)	Live
		Hereditary spastic paraplegia (11089)	Live
		Neurotransmitter disorders (37779)	Live
		Structural basal ganglia disorders (37786)	Live
	Inherited Epilepsy Syndromes (10990)	Genetic Epilepsies with Febrile Seizures Plus (11091)	Live
		Familial Genetic Generalised Epilepsies (11092)	Live
		Familial Focal Epilepsies (11093)	Live
		Epileptic encephalopathy (11094)	Live
		Epilepsy plus other features (41924)	Live
	Motor and Sensory Disorders of the PNS (10991)	Charcot-Marie-Tooth disease (15111)	Live
		Paediatric motor neuronopathies (11099)	Live
	Neurodegenerative disorders (10992)	Early onset and familial Parkinson's Disease (11100)	Live
		Complex Parkinsonism (includes pallido-pyramidal syndromes) (15112)	Live
		Early onset dementia (15113)	Live
		Amyotrophic lateral sclerosis or motor neuron disease (15114)	Live
	Neurodevelopmental disorders (10993)	Classical tuberous sclerosis (11101)	Live
		Intellectual disability (11102)	Live
		Holoprosencephaly (36519)	Live
		Rhomboencephalosynapsis (36603)	Live
		Malformations of cortical development (36526)	Live
		Fetal structural CNS abnormalities (36850)	Live
		Pontine tegmental cap dysplasia (55493)	Live
	Neuromuscular disorders (10994)	Congenital muscular dystrophy (15135)	Live
		Congenital myopathy (11103)	Live
		Congenital myaesthenia (15136)	Live
		Rhabdomyolysis and metabolic muscle disorders (15137)	Live
		Distal myopathies (11104)	Live
		Arthrogryposis (15138)	Live
		Limb girdle muscular dystrophy (11106)	Live
	Channelopathies (11097)	Skeletal Muscle Channelopathies (15139)	Live
		Brain channelopathy (15140)	Live

	Sleep disorders (10995)	Kleine-Levin syndrome and other inherited sleep disorders (11108)	Live
	Cerebrovascular disorders (36610)	Moyamoya disease (36611)	Live
		Vein of Galen malformation (42174)	Live
	Parenchymal brain disorders (36618)	Intracerebral calcification disorders (36619)	Live
	White matter disorders (36626)	Inherited white matter disorders (36627)	Live
Ophthalmological disorders (10996)	Anterior segment abnormalities (10997)	Corneal abnormalities (11110)	Live
		Glaucoma (developmental) (11111)	Live
		Cataracts (11112)	Live
	Posterior segment abnormalities (10998)	Inherited optic neuropathies (11114)	Live
		Rod-cone dystrophy (29268)	Live
		Rod Dysfunction Syndrome (29269)	Live
		Cone Dysfunction Syndrome (29270)	Live
		Inherited macular dystrophy (29271)	Live
		Leber Congenital Amaurosis or Early-Onset Severe Retinal Dystrophy (29272)	Live
		Developmental macular and foveal dystrophy (29273)	Live
		Familial exudative vitreoretinopathy (41900)	Live
	Ocular malformations (10999)	Anophthalmia or microphthamia (11115)	Live
		Ocular coloboma (15141)	Live
	Ocular movement disorders (33350)	Infantile nystagmus (33662)	Live
Psychiatric disorders (71735)	Schizophrenia and other psychotic disorders (71736)	Schizophrenia plus additional features (71740)	Live
	Feeding and eating disorders (71737)	Severe familial anorexia (29278)	Live
Renal and urinary tract disorders (11000)	Syndromes with prominent renal abnormalities (11001)	Proteinuric renal disease (30732)	Live
		Familial haematuria (30733)	Live
		Atypical haemolytic uraemic syndrome (33489)	Live
		Primary membranoproliferative glomerulonephritis (55481)	Live
	Structural renal and urinary tract disease (11003)	Cystic kidney disease (11120)	Live
		Congenital Anomaly of the Kidneys and Urinary Tract (CAKUT) (29277)	Live
	Disorders of function (11004)	Renal tubular acidosis (11123)	Live
		Renal tract calcification (or Nephrolithiasis or nephrocalcinosis) (11124)	Live
		Extreme early-onset hypertension (15142)	Live

		Unexplained kidney failure in young people (36855)	Live
Respiratory disorders (33353)	Interstitial lung disorders (33354)	Familial pulmonary fibrosis (33671)	Live
	Vascular lung disorders (33355)	Hereditary haemorrhagic telangiectasia (33674)	Live
		Familial and multiple pulmonary arteriovenous malformations (33677)	Live
	Structural lung disorders (42203)	Familial primary spontaneous pneumothorax (41819)	Live
Rheumatological disorders (11009)	Multi-system inflammatory or autoimmune disorders (11008)	Periodic fever syndromes and amyloidosis (11127)	Live
		Juvenile dermatomyositis (29219)	Live
	Connective tissues disorders (36930)	Kyphoscoliotic Ehlers-Danlos syndrome (36853)	Live
		Classical Ehlers-Danlos Syndrome (41860)	Live
Skeletal disorders (11005)	Skeletal dysplasias (11007)	Multiple Epiphyseal Dysplasia (11125)	Live
		Chondrodysplasia punctata (15143)	Live
		Thoracic dystrophies (11126)	Live
		Stickler syndrome (11129)	Live
		Osteogenesis imperfecta (30627)	Live
		Unexplained skeletal dysplasia (36854)	Live
		Amelogenesis imperfecta (55449)	Live
	Craniosynostosis (30775)	Craniosynostosis syndromes (11006)	Live
	Choanal anomalies (31500)	Choanal atresia (11078)	Live
Tumour syndromes (11012)	Breast and endocrine (11013)	Familial breast and or ovarian cancer (11131)	Live
		Multiple endocrine tumours (11132)	Live
		Neuro-endocrine Tumours- PCC and PGL (11133)	Live
		Parathyroid cancer (30611)	Live
		Inherited non-medullary thyroid cancer (41884)	Live
	GI tract (11014)	Familial colon cancer (11135)	Live
		Multiple bowel polyps (30615)	Live
		Peutz-Jeghers syndrome (36533)	Live
	Muscle and nerve (11015)	Familial rhabdomyosarcoma or sarcoma (11138)	Live
		Familial tumour syndromes of the central and peripheral nervous system (30619)	Live
		Neurofibromatosis Type 1 (38874)	Live
	Skin (11016)	Genodermatoses with malignancies (30623)	Live
	Young onset tumour syndromes (30781)	Paediatric congenital malformation-dysmorphism-tumour syndromes (30686)	Live

		Exceptionally young adult onset cancer (41892)	Live
	Multiple Primaries (30782)	Multiple Tumours (30685)	Live
Ultra-rare disorders (30783)	Undescribed disorders (30784)	Ultra-rare undescribed monogenic disorders (30785)	Live
	Multi-system groups (38589)	Neonatal or paediatric intensive care admission with a likely monogenic disease (38558)	Live
		Single autosomal recessive mutation in rare disease (38672)	Live
		Undiagnosed monogenic disorder seen in a specialist genetics clinic (42193)	Live

*please refer to relevant Eligibility Statement