Rare Disease Conditions and Phenotypes currently included in the 100,000 Genomes Project

Categories	Sub-Category	Disease
Cardiovascular disorders	**Connective tissue disorders and aortopathies**	Familial Thoracic Aortic Aneurysm Disease
	Cardiac arrhythmia	Brugada syndrome Long QT syndrome Catecholaminergic Polymorphic Ventricular Tachycardia
	Cardiomyopathy	Arrhythmogenic Right Ventricular Cardiomyopathy Left Ventricular Noncompaction Cardiomyopathy Dilated Cardiomyopathy (DCM) Dilated Cardiomyopathy and conduction defects Hypertrophic Cardiomyopathy
	Congenital heart disease	Fallots tetralogy Hypoplastic Left Heart Syndrome Pulmonary atresia Transposition of the great vessels Left Ventricular Outflow Tract obstruction disorders Isomerism and laterality disorders
Dermatological disorders	**Sun-exposure related conditions**	Erythropoietic protoporphyria, mild variant Hydroa vacciniforme

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	Atopy	Severe multi-system atopic disease with high IgE
Dysmorphic and congenital abnormality syndromes	**Kabuki syndrome**	Kabuki syndrome
	RASopathies	Noonan syndrome Noonan syndrome plus other features Cardio-facio-cutaneous syndrome LEOPARD syndrome Costello syndrome Legius syndrome
	Balanced translocations	Balanced translocations with an unusual phenotype
	Limb disorders	VACTERL-like phenotype
	DNA repair disorders	Non-Fanconi anaemia XP-like disorders
	Autophagy disorders	Vici Syndrome and other autophagy disorders
Ciliopathies	**Congenital malformations caused by ciliopathies**	Bardet-Biedl syndrome
	Respiratory ciliopathy phenotypes	Primary ciliary disorders Non-CF bronchiectasis
Endocrine disorders	**Adrenal disorders**	Congenital adrenal hypoplasia
	Disorders of calcium homeostasis	Familial or syndromic hypoparathyroidism
	Growth hormone disorders	IUGR and IGF abnormalities
	Disorders of unusual phenotypes	Familial diabetes Hyperinsulinism
	Obesity Syndromes	Significant early-onset obesity +/- other endocrine features and short stature

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Growth disorders	**Beckwith-Wiedemann syndrome (BWS) and other congenital overgrowth disorders**	Classical Beckwith-Wiedemann syndrome Atypical Beckwith-Wiedemann syndrome Simpson-Golabi-Behmel syndrome Sotos syndrome Weaver syndrome
Haematological disorders	**Primary immunodeficiency disorders**	A- or hypo-gammaglobulinaemia Agranulocytosis Congenital neutropaenia SCID Combined B and T cell defect
	Anaemias and red cell disorders	Early onset pancytopenia and red cell disorders Congenital anaemias
Hearing and ear disorders	**Non-syndromic hearing loss**	Congenital hearing impairment (profound/severe)
	Deafness and congenital structural abnormalities	Bilateral microtia Choanal atresia
Metabolic disorders	**Specific metabolic abnormalities**	Ketotic hypoglycaemia Lactic acidosis Cerebral folate deficiency
	Urea Cycle disorders	Hyperammonaemia
	Lysosomal storage disorders	Mucopolysaccharideosis, Gaucher, Fabry
	Mitochondrial disorders	All recognised syndromes and those with suggestive features

Categories	Sub-Category	Disease
	Peroxisomal disorders	Peroxisomal biogenesis disorders Other peroxisomal disorders
	Disorders of extremely low weight, severe familial anorexia	Severe familial anorexia
Neurology and neurodevelopmental disorders	**Motor Disorders of the CNS**	Hereditary ataxia Early onset dystonia Hereditary spastic paraplegia
	Inherited Epilepsy Syndromes	Genetic Epilepsies with Febrile Seizures Plus (GEFS+) Familial Genetic Generalised Epilepsies Familial Focal Epilepsies Epileptic encephalopathy
	Motor and Sensory Disorders of the PNS	Charcot-Marie-Tooth disease Paediatric motor neuronopathies
	Neurodegenerative disorders	Early onset and familial Parkinson's Disease Complex Parkinsonism (includes pallido-pyramidal syndromes) Early onset dementia (encompassing fronto-temporal dementia and prion disease) Amyotrophic lateral sclerosis/motor neuron disease
	Neurodevelopmental disorders	Classical tuberous sclerosis Intellectual disability
	Neuromuscular disorders	Congenital muscular dystrophy Congenital myopathy Congenital myaesthenia

Categories	Sub-Category	Disease
		Rhabdomyolysis and metabolic muscle disorders Distal myopathies Limb girdle muscular dystrophy Arthrogrythsis
	Channelopathies	Skeletal Muscle Channelopathies Brain channelopathy
	Sleep disorders	Kleine-Levin syndrome
Ophthalmological disorders	**Anterior segment abnormalities**	Corneal abnormalities Glaucoma (developmental) Cataracts
	Posterior segment abnormalities	Inherited optic neuropathies Rod-cone dystrophy Rod Dysfunction Syndrome Cone Dysfunction Syndrome Inherited macular dystrophy Leber Congenital Amaurosis / Early-Onset Severe Retinal Dystrophy Developmental macular and foveal dystrophy
	Ocular malformations	Anophthalmia/microphthamia Ocular coloboma

Categories	Sub-Category	Disease
Renal and urinary tract disorders	**Syndromes with prominent renal abnormalities**	Alport syndrome Imerslund - Grasbeck syndrome
	Structural renal and urinary tract disease	Cystic kidney disease CAKUT
	Disorders of function	Renal tubular acidosis Renal tract calcification (or Nephrolithiasis/nephrocalcinosis) Extreme early-onset hypertension
Skeletal disorders	**Craniosynostosis syndromes**	Craniosynostosis syndromes
	Skeletal dysplasias	Multiple Epiphyseal Dysplasia Chondrodysplasia punctata Thoracic dystrophies Stickler syndrome
	Metabolic bone disease	
Rheumatological disorders	**Multi-system inflammatory/ autoimmune disorders**	Juvenile dermatomyositis Periodic fever syndromes
Tumour predisposition syndromes	**Breast and endocrine**	Familial breast cancer Multiple endocrine tumours Neuro-endocrine Tumours- PCC and PGL

Categories	Sub-Category	Disease
	GI tract	Familial colon cancer
	Muscle and nerve	Familial rhabdomyosarcoma Familial schwannomatosis
	Skin	Gorlin syndrome