

LIST OF DISEASES ASSESSED BY G-MENDELIOME CES PANEL

Category	Related Diseases
Cardiology	Aortopathy and connective tissue disorders
	Arrhythmia
	Cardiomyopathy
	Congenital heart defect
	Dyslipidemia
	Other cardiovascular disease
	Pulmonary hypertension
Dermatology	Adams-Oliver syndrome
	Albinism
	Cardiofaciocutaneous syndrome
	Cutis laxa
	Dyskeratosis congenita
	Ectodermal dysplasia
	Ehlers-Danlos syndrome
	Epidermolysis bullosa
	Hereditary acrodermatitis enteropathica
	Hermansky-Pudlak syndrome
	Hypotrichosis
	Ichthyosis
	Neurofibromatosis
	Pachyonychia congenita
	Palmoplantar keratoderma
	Progeria and Progeroid Syndromes
	Skin cancer
	Tuberous sclerosis
	Waardenburg syndrome
Xeroderma pigmentosum	
Endocrinology	Adrenal hyperplasia
	Diabetes
	Hyperinsulinism
	Hyperparathyroidism
	Hypothyroidism
	Kallmann syndrome
	Multiple endocrine neoplasia
	Obesity
	Pancreatitis
	Premature ovarian failure
ENT	Hearing loss
GI/Hepatology	Cholelithiasis
	Congenital diarrhea
	Congenital hepatic fibrosis
	Gastrointestinal atresia
	Hirschsprung disease
Hematology	Polycystic liver disease
	Anemia
	Bleeding&Thrombotic disorder
	Bone marrow failure
	Congenital neutropenia
	Hemochromatosis
Immunology	RBC membrane disorder
	Antibody deficiencies
	Autoinflammatory disorders
	Combined T/B cell deficiencies
	Complement deficiencies
	Defects in intrinsic and innate immunity
	Immune dysregulation
	Phagocytic defects

Category	Related Diseases
Metabolism	Aminoacidopathies
	Carbohydrate disorders
	Congenital disorders of glycosylation
	Creatine biosynthesis disorders
	Fatty acid oxidation defects
	Lipodystrophy
	Lysosomal storage disorders
	Organic acidemias
	Peroxisomal disorders
	Porphyria
	Purine/Pyrimidine metabolism disorders
	Pyruvate metabolism and tricarboxylic acid cycle defects
	Urea cycle disorders
Nephrology	Bartter syndrome
	Ciliopathies
	Diabetes insipidus
	Hemolytic uremic syndrome
	Hypokalemia
	Hypomagnesemia
	Hypophosphatemic rickets
	Nephrolithiasis
	Nephrotic syndrome/Focal glomerulonephrosis
	Pseudohypoadosteronism
	Renal malformation
Renal tubular acidosis	
Neurology	Autism
	Movement disorders
	Neurodegenerative disorders
	Neuromuscular disorders
	Neuropathies and related disorders
	Seizures and Brain abnormalities
	Breast and gynecological cancer
	Colorectal cancer
	Endocrine cancer
	Gastrointestinal cancer
Oncology	Hematologic malignancy
	Lung cancer
	Nervous system/brain cancer
	Pancreatic cancer
	Prostate cancer
	Renal cancer
	Sarcoma
	Skin cancer
	Albinism
	Cataract/Ectopia lentis
	Corneal dystrophy
Ophthalmology	Glaucoma
	Microphthalmia/Anophthalmia
	Nystagmus
	Ophthalmoplegia/Oculomotor apraxia
	Optic atrophy
Pulmonology	Retinal dystrophy
	Retinoblastoma
	Bronchiectasis
	Central hypoventilation/Apnea
	Cystic fibrosis
	Cystic lung disease
	Hermansky-Pudlak syndrome
	Interstitial lung disease
	Primary ciliary dyskinesia
	Surfactant dysfunction
Skeletal disorders	Amelogenesis imperfecta
	Arthrogyposes
	Cleft lip palate
	Craniosynostosis
	Exostosis
	Facial dysostosis
	Macrocephaly/Overgrowth syndrome
	Osteopetrosis
	Short stature syndrome
	Skeletal dysplasia

