Appendix 1

Appendix 1: Alphabetical listing of genetic disorders in the Arab population of Saudi Arabia as indexed in the CTGA Database (November 2018).

OMIM#	Genetic Disorder
	Autosomal recessive
246450	3-@Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency
210200	3-@Methylcrotonyl-CoA Carboxylase 1 Deficiency
250950	3-@Methylglutaconic Aciduria, Type I
201810	3-Beta-Hydroxysteroid Dehydrogenase, Type II, Deficiency of
210210	3-Methylcrotonyl-CoA carboxylase 2 deficiency*
258501	3-Methylglutaconic Aciduria, Type III
200100	Abetalipoproteinemia*
231550	Achalasia-Addisonianism-Alacrima Syndrome
201100	Acrodermatitis Enteropathica, Zinc-Deficiency Type
201450	Acyl-CoA Dehydrogenase, Medium-Chain, Deficiency of
201470	Acyl-CoA Dehydrogenase, Short-Chain, Deficiency of*
201475	Acyl-CoA Dehydrogenase, Very Long-Chain, Deficiency of*
614219	Adams-Oliver Syndrome 2*
202010	Adrenal Hyperplasia, Congenital, due to 11-Beta-Hydroxylase Deficiency
201910	Adrenal Hyperplasia, Congenital, due to 21-Hydroxylase Deficiency
617126	Alazami-Yuan syndrome*
203100	Albinism, oculocutaneous, type IA
203290	Albinism, Oculocutaneous, Type III*
203500	Alkaptonuria
617822	Alkuraya-Kucinskas syndrome*
203655	Alopecia Universalis Congenita
203800	Alstrom Syndrome
604498	Amegakaryocytic Thrombocytopenia, Congenital
612529	Amelogenesis Imperfecta, Hypomaturation Type, IIA2*
206500	Anencephaly
269400	Anterior segment dysgenesis 7, with sclerocornea*
207800	Argininemia
207900	Argininosuccinic Aciduria
208050	Arterial Tortuosity Syndrome
617146	Arthrogryposis, Distal, with Impaired Proprioception and Touch*
614262	Arthrogryposis, Perthes Disease, and Upward Gaze Palsy*
208085	Arthrogryposis, Renal Dysfunction, and Cholestasis 1
208230	Arthropathy, Progressive Pseudorheumatoid, of Childhood
208300	Ascites, Chylous
615574	Asparagine Synthetase Deficiency*
615217	Ataxia-Oculomotor Apraxia 3*
604391	Ataxia-Telangiectasia-Like Disorder*

OMIM#	Genetic Disorder
601536	Athabaskan Brainstem Dysgenesis Syndrome*
192132	ATPase, H+ Transporting, Lysosomal, 56/58-Kd, V1 Subunit B, Isoform 1
601071	Auditory neuropathy, autosomal recessive, 1; Deafness, autosomal recessive 9
602271	Axial Spondylometaphyseal Dysplasia*
251290	Band-Like Calcification with Simplified Gyration and Polymicrogyria
209900	Bardet-Biedl Syndrome
615981	Bardet-Biedl syndrome 2*
607364	Bartter Syndrome, Type 3
607483	Basal Ganglia Disease, Biotin-Responsive
231200	Bernard-Soulier Syndrome*
607765	Bile Acid Synthesis Defect, Congenital, 1*
253260	Biotinidase Deficiency
607475	Bothnia Retinal Dystrophy*
229200	Brittle Cornea Syndrome 1
259450	Bruck Syndrome 1*
208250	Camptodactyly-Arthropathy-Coxa Vara-Pericarditis Syndrome
271900	Canavan Disease
611880	Cardiomyopathy, Dilated, 2A*
212110	Cardiomyopathy, Dilated, Autosomal Recessive *
212140	Carnitine Deficiency, Systemic Primary
255120	Carnitine Palmitoyltransferase I Deficiency
608836	Carnitine Palmitoyltransferase II Deficiency, Lethal Neonatal
212138	Carnitine-Acylcarnitine Translocase Deficiency*
116700	Cataract 13 with adult i phenotype*
611544	Cataract 17, Multiple Types*
610019	Cataract 18, autosomal recessive*
212750	Celiac Disease
613227	Cerebellar Ataxia, Mental Retardation, and Dysequilibrium Syndrome 3
616875	Cerebellar Atrophy, Visual Impairment, And Psychomotor Retardation*
214150	Cerebrooculofacioskeletal Syndrome 1
213700	Cerebrotendinous Xanthomatosis*
204200	Ceroid Lipofuscinosis, Neuronal, 3
601780	Ceroid lipofuscinosis, neuronal, 6*
615284	Charcot-Marie-Tooth Disease, Type 4B3*
214500	Chediak-Higashi Syndrome
605479	Cholestasis, benign recurrent intrahepatic, 2*
211600	Cholestasis, progressive familial intrahepatic 1

OMIM#	Genetic Disorder
602347	Cholestasis, progressive familial intrahepatic 3*
615878	Cholestasis, progressive familial intrahepatic 4*
200700	Chondrodysplasia, Grebe Type
200150	Choreoacanthocytosis
215480	Choroid Plexus Calcification and Mental Retardation*
244400	Ciliary Dyskinesia, Primary, 1
215700	Citrullinemia, Classic
600373	CODAS syndrome
216550	Cohen Syndrome
616294	Cole-Carpenter syndrome 2*
610498	Combined oxidative phosphorylation deficiency 2*
610427	Cone-Rod Synaptic Disorder, Congenital Nonprogressive*
212065	Congenital Disorder of Glycosylation, Type 1a
603147	Congenital Disorder of Glycosylation, Type 1c*
607906	Congenital Disorder of Glycosylation, Type 1i*
608093	Congenital Disorder of Glycosylation, Type 1j*
614921	Congenital Disorder of Glycosylation, type 1t*
614576	Congenital Disorder of Glycosylation, Type 2l
601110	Congenital disorder of glycosylation, type Id*
608104	Congenital disorder of glycosylation, type Ih*
212066	Congenital Disorder of Glycosylation, Type IIa*
613612	Congenital Disorder of Glycosylation, Type IIi*
608776	Congenital Disorder of Glycosylation, Type II*
612015	Congenital Disorder of Glycosylation, Type In
217095	Conotruncal Heart Malformations
217300	Cornea Plana 2*
217700	Corneal Endothelial Dystrophy 2, Autosomal Recessive
217990	Corpus Callosum, Agenesis of
614115	Cortical malformations, occipital*
607812	Craniolenticulosutural Dysplasia*
218800	Crigler-Najjar Syndrome
219150	Custis Laxa, Autosomal Recessive, Type IIIA
219200	Cutis Laxa, Autosomal Recessive, Type IIA
614438	Cutis Laxa, Autosomal Recessive, Type IIIB*
219700	Cystic Fibrosis
219800	Cystinosis, Nephropathic
220100	Cystinuria
220200	Dandy-Walker Syndrome
220210	Dandy-Walker-Like Malformation with Atrioventricular Septal Defect
610706	Deafness, Congenital, with Inner Ear Agenesis, Microtia, and Microdontia*
251450	Desbuquois Syndrome
616901	Developmental delay with short stature, dysmorphic features, and sparse hair*

OMIM#	Genetic Disorder
222100	Diabetes Mellitus, Insulin-Dependent
610199	Diabetes Mellitus, Neonatal, with Congenital Hypothyroidism*
222400	Diaphragmatic Hernia 2
142340	Diaphragmatic Hernia, Congenital
214700	Diarrhea 1, Secretory Chloride, Congenital
270420	Diarrhea 3, Secretory Sodium, Congenital, with or without Other Congenital Anomalies*
613217	Diarrhea 5, with tufting enteropathy, congenital
246900	Dihydrolipoamide dehydrogenase deficiency
274270	Dihydropyrimidine Dehydrogenase Deficiency*
222448	Donnai-Barrow Syndrome
609535	Drug Metabolism, Poor, CYP2C19-Related
608902	Drug Metabolism, Poor, CYP2D6-Related
237500	Dubin-Johnson syndrome
612715	Dyschromatosis Universalis Hereditaria 2*
225400	Ehlers-Danlos Syndrome, Type VI
225500	Ellis-van Creveld Syndrome
268100	Enhanced S-Cone Syndrome
226600	Epidermolysis Bullosa Dystrophica, Autosomal Recessive
226730	Epidermolysis Bullosa Junctionalis with Pyloric Atresia
226670	Epidermolysis Bullosa Simplex with Muscular Dystrophy*
226650	Epidermolysis Bullosa, Junctional, Non-Herlitz Type
617020	Epileptic Encephalopathy, Early Infantile, 38
606369	Epileptic Encephalopathy, Lennox-Gastaut Type
226980	Epiphyseal Dysplasia, Multiple, with Early-Onset Diabetes Mellitus
249100	Familial Mediterranean Fever
227650	Fanconi Anemia
227810	Fanconi-Bickel Syndrome*
228000	Farber Lipogranulomatosis
602078	Fibrosis of Extraocular Muscles, Congenital, 2*
219000	Fraser Syndrome
229300	Friedreich Ataxia 1
229700	Fructose-1,6-Bisphophatase Deficiency
230400	Galactosemia
256540	Galactosialidosis
608013	Gaucher disease, perinatal lethal*
607313	Gaze Palsy, Familial Horizontal, with Progressive Scoliosis
231070	Geroderma Osteodysplasticum
273800	Glanzmann Thrombasthenia
231300	Glaucoma 3, Primary Congenital, A
202200	Glucocorticoid Deficiency 1
231670	Glutaric Acidemia I
138300	Glutathione Reductase*

OMIM#	Genetic Disorder
266130	Glutathione Synthetase Deficiency
605899	Glycine Encephalopathy
232200	Glycogen Storage Disease I*
232220	Glycogen Storage Disease Ib; GSD1B
232300	Glycogen Storage Disease II
232400	Glycogen Storage Disease III
232800	Glycogen Storage Disease VII*
230500	GM1-Gangliosidosis, Type I
230600	GM1-Gangliosidosis, Type II*
275000	Graves Disease
607624	Griscelli Syndrome, Type 2
616920	Heart and Brain Malformation Syndrome
267700	Hemophagocytic Lymphohistiocytosis, Familial, 1
603553	Hemophagocytic Lymphohistiocytosis, Familial, 2
603552	Hemophagocytic Lymphohistiocytosis, Familial, 4*
236000	Hodgkin Lymphoma
253270	Holocarboxylase Synthetase Deficiency
236100	Holoprosencephaly
236200	Homocystinuria Due to Cystathionine Beta-Synthase Deficiency
236250	Homocystinuria due to Deficiency of N(5,10)-Methylenetetrahydrofolate Reductase Activity
236410	Humeroradial Synostosis with Craniofacial Anomalies*
604802	Huntington Disease-Like 3*
607014	Hurler Syndrome
228600	Hyaline Fibromatosis Syndrome
236600	Hydrocephalus
615219	Hydrocephalus, congenital, 2, with or without brain or eye anomalies
236750	Hydrops Fetalis, Idiopathic
237450	Hyperbilirubinemia, Rotor Type*
614619	Hyperekplexia 2*
243700	Hyperimmunoglobulin E-Recurrent Infection Syndrome, Autosomal Recessive
238970	Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome
259900	Hyperoxaluria, Primary, Type I
248250	Hypomagnesemia 3, Renal
241410	Hypoparathyroidism-Retardation-Dysmorphism Syndrome
616900	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3*
278150	Hypotrichosis 8*
242100	Ichthyosiform Erythroderma, Congenital, Nonbullous, 1
601277	Ichthyosis, Congenital, Autosomal Recessive 4A*
242300	Ichthyosis, Lamellar, 1
614457	Ichthyosis, Spastic Quadriplegia, and Mental Retardation*

OMIM#	Genetic Disorder
209950	Immunodeficiency 27A*
614890	Immunodeficiency 29*
614700	Immunodeficiency, common variable, 8, with autoimmunity
242860	Immunodeficiency-Centromeric Instability-Facial Anomalies Syndrome 1
256800	Insensitivity to Pain, Congenital, with Anhidrosis
243500	Isovaleric Acidemia
217080	Jalili Syndrome
243600	Jejunal Atresia
220400	Jervell and Lange-Nielsen Syndrome 1
243800	Johanson-Blizzard Syndrome*
213300	Joubert Syndrome
615636	Joubert Syndrome 21*
616654	Joubert Syndrome 24*
608629	Joubert Syndrome 3
610188	Joubert Syndrome 5
610688	Joubert Syndrome 6*
611560	Joubert Syndrome 7*
612285	Joubert Syndrome 9*
244460	Kenny-Caffey Syndrome, Type 1
173650	Kindler Syndrome
616549	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism*
267750	Knobloch Syndrome 1*
245200	Krabbe Disease
236792	L-2-Hydroxyglutaric Aciduria
245570	Landau-Kleffner Syndrome
617182	Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia*
204000	Leber Congenital Amaurosis 1*
612712	Leber Congenital Amaurosis 13*
613843	Leber Congenital Amaurosis 15
204100	Leber Congenital Amaurosis 2
613835	Leber Congenital Amaurosis 8
256000	Leigh Syndrome
116920	Leukocyte Adhesion Deficiency, Type I
608804	Leukodystrophy, Hypomyelinating, 2*
611105	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation*
603896	Leukoencephalopathy with Vanishing White Matter
608594	Lipodystrophy, congenital generalized, type 1
201710	Lipoid Congenital Adrenal Hyperplasia
247100	Lipoid Proteinosis of Urbach and Wiethe
614019	Lissencephaly 4*
257320	Lissencephaly Syndrome, Norman-Roberts Type*

OMIM#	Genetic Disorder
211980	Lung Cancer
222700	Lysinuric Protein Intolerance
278000	Lysosomal Acid Lipase Deficiency
248360	Malonyl-CoA Decarboxylase Deficiency
248500	Mannosidosis, Alpha B, Lysosomal
248600	Maple Syrup Urine Disease
248800	Marinesco-Sjogren Syndrome
613885	Meckel Syndrome 8*
249000	Meckel Syndrome, Type 1
615397	Meckel Syndrome, Type 11*
612284	Meckel Syndrome, Type 6*
614209	Meckel Syndrome, Type 9*
261100	Megaloblastic Anemia 1
224690	Meier-Gorlin Syndrome 1
615162	Mental Retardation, Autosomal Recessive 35*
615286	Mental Retardation, Autosomal Recessive 36
615942	Mental Retardation, Autosomal Recessive 44
617028	Mental Retardation, Autosomal Recessive 54*
616878	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration*
250100	Metachromatic Leukodystrophy
249900	Metachromatic Leukodystrophy due to Saposin B Deficiency*
251000	Methylmalonic Aciduria due to Methylmalonyl-CoA Mutase Deficiency
251100	Methylmalonic Aciduria, cblA Type*
251110	Methylmalonic Aciduria, cblB Type
616486	Microcephaly 15, primary, autosomal recessive*
617090	Microcephaly 17, Primary, Autosomal Recessive*
604317	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations*
604804	Microcephaly 3, primary, autosomal recessive*
608716	Microcephaly 5, primary, autosomal recessive
608393	Microcephaly 6, primary, autosomal recessive*
612703	Microcephaly 7, primary, autosomal recessive*
614673	Microcephaly 8, primary, autosomal recessive*
614852	Microcephaly 9, primary, autosomal recessive*
251200	Microcephaly, Primary Autosomal Recessive, 1
614833	Microcephaly, Short Stature, and Polymicrogyria with or without Seizures*
611040	Microphthalmia, Isolated 5*
613517	Microphthalmia, Isolated 6*
615113	Microphthalmia, isolated 8*
601186	Microphthalmia, isolated, with coloboma 8*
615145	Microphthalmia, Isolated, With Coloboma 9*
276300	Mismatch Repair Cancer Syndrome

OMIM#	Genetic Disorder
124000	Mitochondrial Complex III Deficiency, Nuclear Type 1*
251880	Mitochondrial DNA Depletion Syndrome 3 (Hepatocerebral Type)
256810	Mitochondrial DNA Depletion Syndrome 6 (Hepatocerebral Type)*
616277	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency
253000	Morquio Syndrome A
252350	Moyamoya Disease 1
252500	Mucolipidosis II Alpha/Beta
252900	Mucopolysaccharidosis Type IIIA
253200	Mucopolysaccharidosis Type VI
253220	Mucopolysaccharidosis VII
259600	Multicentric Osteolysis, Nodulosis, and Arthropathy
616370	Multiple mitochondrial dysfunctions syndrome 4*
265000	Multiple Pterygium Syndrome, Escobar Variant
253600	Muscular Dystrophy, Limb-Girdle, Type 2A
253700	Muscular Dystrophy, Limb-Girdle, Type 2C
236670	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1
607155	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5
608931	Myasthenic Syndrome, Congenital, 4C, Associated with Acetylcholine Receptor Deficiency
615511	Myopathy due to myoadenylate deaminase deficiency*
206800	Nail Disorder, Nonsyndromic Congenital, 4*
256100	Nephronophthisis 1
614377	Nephronophthisis 13; Senior-Loken syndrome 8*
602088	Nephronophthisis 2
606966	Nephronophthisis 4; Senior-Loken syndrome 4*
600995	Nephrotic Syndrome, Steroid-Resistant, Autosomal Recessive
256300	Nephrotic Syndrome, Type 1
616730	Nephrotic syndrome, type 11*
610725	Nephrotic syndrome, type 3*
256500	Netherton Syndrome
256520	Neu-Laxova Syndrome 1
601634	Neural Tube Defects, Folate-Sensitive
256550	Neuraminidase Deficiency
256600	Neuroaxonal Dystrophy, Infantile
617913	Neurodevelopmental disorder with microcephaly, cataracts, and renal abnormalities*
616263	Neurologic, Endocrine, and Pancreatic Disease, Multisystem, Infantile-Onset
613115	Neuropathy, Hereditary Sensory and Autonomic, Type IIB*
608654	Neuropathy, Hereditary Sensory and Autonomic, Type V
610738	Neutropenia, Severe Congenital, 3, Autosomal Recessive*
257200	Niemann-Pick Disease, Type A
607616	Niemann-Pick Disease, Type B

OMIM#	Genetic Disorder
609016	Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency
211980	Lung Cancer
222700	Lysinuric Protein Intolerance
278000	Lysosomal Acid Lipase Deficiency
248360	Malonyl-CoA Decarboxylase Deficiency
248500	Mannosidosis, Alpha B, Lysosomal
248600	Maple Syrup Urine Disease
248800	Marinesco-Sjogren Syndrome
613885	Meckel Syndrome 8*
249000	Meckel Syndrome, Type 1
615397	Meckel Syndrome, Type 11*
612284	Meckel Syndrome, Type 6*
614209	Meckel Syndrome, Type 9*
261100	Megaloblastic Anemia 1
224690	Meier-Gorlin Syndrome 1
615162	Mental Retardation, Autosomal Recessive 35*
615286	Mental Retardation, Autosomal Recessive 36
615942	Mental Retardation, Autosomal Recessive 44
617028	Mental Retardation, Autosomal Recessive 54*
616878	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration*
250100	Metachromatic Leukodystrophy
249900	Metachromatic Leukodystrophy due to Saposin B Deficiency*
251000	Methylmalonic Aciduria due to Methylmalonyl-CoA Mutase Deficiency
251100	Methylmalonic Aciduria, cblA Type*
251110	Methylmalonic Aciduria, cblB Type
616486	Microcephaly 15, primary, autosomal recessive*
617090	Microcephaly 17, Primary, Autosomal Recessive*
604317	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations*
604804	Microcephaly 3, primary, autosomal recessive*
608716	Microcephaly 5, primary, autosomal recessive
608393	Microcephaly 6, primary, autosomal recessive*
612703	Microcephaly 7, primary, autosomal recessive*
614673	Microcephaly 8, primary, autosomal recessive*
614852	Microcephaly 9, primary, autosomal recessive*
251200	Microcephaly, Primary Autosomal Recessive, 1
614833	Microcephaly, Short Stature, and Polymicrogyria with or without Seizures*
611040	Microphthalmia, Isolated 5*
613517	Microphthalmia, Isolated 6*
615113	Microphthalmia, isolated 8*
601186	Microphthalmia, isolated, with coloboma 8*
615145	Microphthalmia, Isolated, With Coloboma 9*

OMIM#	Genetic Disorder
276300	Mismatch Repair Cancer Syndrome
124000	Mitochondrial Complex III Deficiency, Nuclear Type 1*
251880	Mitochondrial DNA Depletion Syndrome 3 (Hepatocerebral Type)
256810	Mitochondrial DNA Depletion Syndrome 6 (Hepatocerebral Type)*
616277	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency
253000	Morquio Syndrome A
252350	Moyamoya Disease 1
252500	Mucolipidosis II Alpha/Beta
252900	Mucopolysaccharidosis Type IIIA
253200	Mucopolysaccharidosis Type VI
253220	Mucopolysaccharidosis VII
259600	Multicentric Osteolysis, Nodulosis, and Arthropathy
616370	Multiple mitochondrial dysfunctions syndrome 4*
265000	Multiple Pterygium Syndrome, Escobar Variant
253600	Muscular Dystrophy, Limb-Girdle, Type 2A
253700	Muscular Dystrophy, Limb-Girdle, Type 2C
236670	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1
607155	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5
608931	Myasthenic Syndrome, Congenital, 4C, Associated with Acetylcholine Receptor Deficiency
615511	Myopathy due to myoadenylate deaminase deficiency*
206800	Nail Disorder, Nonsyndromic Congenital, 4*
256100	Nephronophthisis 1
614377	Nephronophthisis 13; Senior-Loken syndrome 8*
602088	Nephronophthisis 2
606966	Nephronophthisis 4; Senior-Loken syndrome 4*
600995	Nephrotic Syndrome, Steroid-Resistant, Autosomal Recessive
256300	Nephrotic Syndrome, Type 1
616730	Nephrotic syndrome, type 11*
610725	Nephrotic syndrome, type 3*
256500	Netherton Syndrome
256520	Neu-Laxova Syndrome 1
601634	Neural Tube Defects, Folate-Sensitive
256550	Neuraminidase Deficiency
256600	Neuroaxonal Dystrophy, Infantile
617913	Neurodevelopmental disorder with microcephaly, cataracts, and renal abnormalities*
616263	Neurologic, Endocrine, and Pancreatic Disease, Multisystem, Infantile-Onset
613115	Neuropathy, Hereditary Sensory and Autonomic, Type IIB*
608654	Neuropathy, Hereditary Sensory and Autonomic, Type V

OMIM#	Genetic Disorder
610738	Neutropenia, Severe Congenital, 3, Autosomal Recessive*
257200	Niemann-Pick Disease, Type A
607616	Niemann-Pick Disease, Type B
257220	Niemann-Pick Disease, Type C1
614565	Night blindness, congenital stationary (complete), 1E, autosomal recessive*
613216	Night Blindness, Congenital Stationary, Type 1C*
258660	Nonarteritic Anterior Ischemic Optic Neuropathy, Susceptibility to*
212550	Optic Disc Anomalies with Retinal and/or Macular Dystrophy
277170	Orofaciodigital Syndrome VI
610968	Osteogenesis Imperfecta, Type XI*
259700	Osteopetrosis, Autosomal Recessive 1
259730	Osteopetrosis, Autosomal Recessive 3
245000	Papillon-Lefevre Syndrome
605909	Parkinson Disease 6, Autosomal Recessive Early-Onset
604228	Partial Albinism and Immunodeficiency Syndrome*
607411	Patent Ductus Arteriosus
270300	Peeling Skin Syndrome
614886	Peroxisome Biogenesis Disorder 12A (Zellweger)*
202370	Peroxisome biogenesis disorder 2B
614872	Peroxisome biogenesis disorder 7A (Zellweger)*
615300	Perrault Syndrome 4*
261550	Persistent Mullerian Duct Syndrome, Types I and II
261600	Phenylketonuria
263200	Polycystic Kidney Disease, Autosomal Recessive
617026	Pontocerebellar hypoplasia, type 2F*
601815	Phosphoglycerate Dehydrogenase Deficiency*
606353	Primary Lateral Sclerosis, Juvenile
606054	Propionic Acidemia
264600	Pseudovaginal Perineoscrotal Hypospadias
265430	Pulmonary Hypoplasia, Primary
613179	Purine Nucleoside Phosphorylase Deficiency*
265950	Pyloric Atresia
266140	Pyropoikilocytosis, Hereditary*
259775	Raine Syndrome
602722	Renal Tubular Acidosis, Distal, Autosomal Recessive
611555	Renal Tubular Acidosis, Distal, with Nephrocalcinosis, Short Stature, Mental Retardation, and Distinctive Facies
267300	Renal Tubular Acidosis, Distal, with Progressive Nerve Deafness
208540	Renal-Hepatic-Pancreatic Dysplasia 1*
615415	Renal-Hepatic-Pancreatic Dysplasia 2*
614224	Retinal Arterial Macroaneurysm with Supravalvular Pulmonic Stenosis*

OMIM#	Genetic Disorder
610356	Retinal cone dystrophy 3B*
600105	Retinitis Pigmentosa 12*
613862	Retinitis Pigmentosa 38*
613581	Retinitis pigmentosa 56*
617023	Retinitis pigmentosa 75*
268240	Rheumatic Fever-Related Antigen
222765	Rhizomelic Chondrodysplasia Punctata, Type 2
268310	Robinow Syndrome, Autosomal Recessive
268800	Sandhoff Disease
242900	Schimke Immunoosseous Dysplasia*
266900	Senior-Loken Syndrome 1
102700	Severe Combined Immunodeficiency, Autosomal Recessive, T Cell-Negative, B Cell-Negative, Nk Cell-Negative, due to Adenosine Deaminase Deficiency
615328	Shaheen Syndrome*
613091	Short-rib thoracic dysplasia 3 with or without polydactyly*
266920	Short-Rib Thoracic Dysplasia 9 with or without Polydactyly*
603903	Sickle Cell Anemia
607655	Skin Fragility-Woolly Hair Syndrome*
270400	Smith-Lemli-Opitz Syndrome
611225	Spastic Paraplegia 18, Autosomal Recessive
607584	Spastic Paraplegia 24, Autosomal Recessive*
601608	Spastic Paraplegia and Evans Syndrome*
607608	Sphingomyelin Phosphodiesterase 1, Acid Lysosomal
604320	Spinal Muscular Atrophy with Respiratory Distress 1
253300	Spinal Muscular Atrophy, Type I
253550	Spinal Muscular Atrophy, Type II
253400	Spinal Muscular Atrophy, Type III
606002	Spinocerebellar Ataxia, Autosomal Recessive 1*
614322	Spinocerebellar Ataxia, Autosomal Recessive 12*
607250	Spinocerebellar Ataxia, Autosomal Recessive, with Axonal Neuropathy*
220600	Split-Hand/Foot Malformation 1 with Sensorineural Hearing Loss, Autosomal Recessive
225300	Split-Hand/Foot Malformation 6*
275355	Squamous Cell Carcinoma, Head and Neck
272300	Sulfocysteinuria
610921	Surfactant Metabolism Dysfunction, Pulmonary, 3*
614420	Systemic Lupus Erythematosus 16*
272800	Tay-Sachs Disease
272750	Tay-Sachs Disease, AB Variant*
218340	Temtamy Syndrome
273750	Three M Syndrome 1
612921	Three M Syndrome 2

OMIM#	Genetic Disorder
612304	Thrombophilia Due to Protein C Deficiency, Autosomal Recessive*
606003	Transaldolase Deficiency
276600	Tyrosine Transaminase Deficiency
276700	Tyrosinemia, Type I
276820	Ulna and Fibula, Absence of, with Severe Limb Deficiency
236730	Urofacial Syndrome
276904	Usher Syndrome, Type IC
276902	Usher Syndrome, Type IIIA*
611938	Ventricular Tachycardia, Catecholaminergic Polymorphic, 2*
242840	Vici Syndrome
264700	Vitamin D Hydroxylation-Deficient Rickets, Type 1A
277440	Vitamin D-Dependent Rickets, Type II
277480	Von Willebrand Disease, Type 3
600118	Warburg micro syndrome 1
277600	Weill-Marchesani Syndrome, Autosomal Recessive*
613195	Weill-Marchesani-like Syndrome
277900	Wilson Disease
241080	Woodhouse-Sakati Syndrome
278250	Wrinkly Skin Syndrome
279000	Young Syndrome
214100	Zellweger Syndrome

Autosomal dominant

105250	Amyloidosis, Primary Localized Cutaneous, 1
106210	Aniridia
107320	Antiphospholipid Syndrome, Familial
101200	Apert Syndrome
107650	Apnea, Obstructive Sleep
108010	Arteriovenous Malformations of the Brain
193700	Arthrogryposis, Distal, Type 2A
600807	Asthma, Susceptibility To
108800	Atrial Septal Defect 1
606215	Atrioventricular Septal Defect
143465	Attention Deficit-Hyperactivity Disorder
615032	Autism, Susceptibility to, 18*
269200	Autoimmune Polyendocrine Syndrome, Type II*
109400	Basal Cell Nevus Syndrome
110100	Blepharophimosis, Ptosis, and Epicanthus Inversus*
114480	Breast Cancer
114000	Caffey Disease
604307	Cataract, Coppock-Like
116600	Cataract 6, multiple types*
604219	Cataract 9, Multiple Types*

OMIM#	Genetic Disorder
125310	Cerebral Arteriopathy, Autosomal Dominant, with Subcortical Infarcts and Leukoencephalopathy
118420	Chiari Malformation Type I*
155145	Cleft, Median, of Upper Lip with Polyps of Facial Skin and Nasal Mucosa
120000	Coarctation of Aorta
114500	Colorectal Cancer
122100	Corneal Dystrophy, Juvenile Epithelial, of Meesmann*
122000	Corneal Dystrophy, Posterior Polymorphous, 1*
121700	Corneal Endothelial Dystrophy 1, Autosomal Dominant*
614701	Cornelia de Lange Syndrome 4*
608320	Coronary Artery Disease, Autosomal Dominant, 1
158350	Cowden Syndrome 1*
123500	Crouzon Syndrome
124200	Darier-White Disease
603165	Dermatitis, Atopic
125853	Diabetes Mellitus, Noninsulin-Dependent
188400	DiGeorge Syndrome
126800	Duane Retraction Syndrome 1*
130020	Ehlers-Danlos Syndrome, Type III*
130710	Emphysema, Congenital Lobar
113800	Epidermolytic Hyperkeratosis*
605375	Epilepsy, Nocturnal Frontal Lobe, 3*
616366	Epileptic Encephalopathy, Early Infantile, 32
133450	Ewing Sarcoma Breakpoint Region 1
177650	Exfoliation Syndrome*
125350	Failure of Tooth Eruption, Primary*
135700	Fibrosis of Extraocular Muscles, Congenital, 1*
600638	Fibrosis of Extraocular Muscles, Congenital, 3A, with or without Extraocular Involvement
137580	Gilles De La Tourette Syndrome
137750	Glaucoma 1, Open Angle, A
102500	Hajdu-Cheney Syndrome*
141000	Hemangioma-Thrombocytopenia Syndrome
141300	Hemifacial Atrophy, Progressive
164210	Hemifacial Microsomia
141800	Hemoglobin - Alpha Locus 1
142250	Hemoglobin, Gamma G
142309	Hemoglobin--Variants for which the Chain Carrying the Mutation is Unknown or Uncertain*
142470	Heterocellular Hereditary Persistence of Fetal Hemoglobin*
142623	Hirschsprung Disease, Susceptibility to, 1
142946	Holoprosencephaly 4*
142900	Holt-Oram Syndrome
143100	Huntington Disease

OMIM#	Genetic Disorder
606438	Huntington Disease-Like 2
143870	Hypercalciuria, Absorptive, 2
143890	Hypercholesterolemia, Familial
218700	Hypothyroidism, Congenital, Nongoitrous, 2
146700	Ichthyosis Vulgaris*
146750	Ichthyosis, Lamellar, Autosomal Dominant*
614892	Immunodeficiency 31A*
147920	Kabuki Syndrome
148100	Keloid Formation*
148300	Keratoconus 1*
610253	Kleefstra Syndrome*
148840	Kleine-Levin Hibernation Syndrome*
150250	Larsen Syndrome, Autosomal Dominant
601626	Leukemia, Acute Myeloid
151620	Lichen Planus, Familial
192500	Long QT Syndrome 1
613688	Long QT Syndrome 2*
153400	Lymphedema-Distichiasis Syndrome*
145600	Malignant Hyperthermia, Susceptibility To, 1
125851	Maturity-Onset Diabetes of the Young, Type II
277000	Mayer-Rokitansky-Kuster-Hauser Syndrome
152950	Microcephaly with or without Chorioretinopathy, Lymphedema, or Mental Retardation*
610125	Microphthalmia, syndromic 5*
157600	Mirror Movements 1*
157900	Moebius Syndrome
131100	Multiple Endocrine Neoplasia, Type I*
171400	Multiple Endocrine Neoplasia, Type IIA
162300	Multiple Endocrine Neoplasia, Type IIB
600080	Myelocytic Leukemia-Like Syndrome, Familial, Chronic*
608358	Myopathy, Myosin Storage*
107000	Nail Disorder, Nonsyndromic Congenital, 6*
182940	Neural tube defects, susceptibility to
162200	Neurofibromatosis, Type I
604169	Noncompaction of Left Ventricular Myocardium, Familial Isolated, Autosomal Dominant 1
164230	Obsessive-Compulsive Disorder*
613587	Occult macular dystrophy*
164185	Ocular Cicatricial Pemphigoid
164300	Oculopharyngeal Muscular Dystrophy*
119530	Orofacial Cleft 1
166710	Osteoporosis
167000	Ovarian Cancer
148600	Palmoplantar Keratoderma, Punctate Type IA*
175860	Palmoplantar Keratoderma, Punctate Type II
168600	Parkinson Disease
169610	Pemphigus Vulgaris, Familial
173000	Pilonidal Sinus
173800	Poland Syndrome
613095	Polycystic Kidney Disease 2*
174050	Polycystic Liver Disease 1*
174200	Polydactyly, Postaxial, Type A1*
174400	Polydactyly, Preaxial I
176880	Protein S
178550	Pulmonary Hemosiderosis
179010	Pyloric Stenosis, Infantile Hypertrophic 1
180200	Retinoblastoma
180700	Robinow Syndrome, Autosomal Dominant
181500	Schizophrenia
121800	Schnyder Corneal Dystrophy*
181800	Scoliosis, Isolated, Susceptibility to, 1*
601764	Seizures, Benign Familial Infantile, 1*
156610	Skin Creases, Congenital Symmetric Circumferential, 1*
108600	Spastic Ataxia 1, Autosomal Dominant*
603563	Spastic Paraplegia 8, Autosomal Dominant*
606658	Spinocerebellar Ataxia 15
607136	Spinocerebellar Ataxia 17*
119100	Split-Hand/Foot Malformation with Long Bone Deficiency 1
106300	Spondyloarthropathy, Susceptibility to, 1
186000	Synpolydactyly 1*
152700	Systemic Lupus Erythematosus
600376	Telangiectasia, Hereditary Hemorrhagic, Type 2*
187500	Tetralogy of Fallot
188030	Thrombocytopenic Purpura, Autoimmune
188050	Thrombophilia
176860	Thrombophilia, Hereditary due to Protein C Deficiency, Autosomal Dominant
616535	Thyroid Cancer, Nonmedullary, 5*
155240	Thyroid Carcinoma, Familial Medullary*
188550	Thyroid Carcinoma, Papillary
604625	Tooth Agenesis, Selective, 3*
106700	Total Anomalous Pulmonary Venous Return 1
608808	Transposition of the Great Arteries, Dextro-Looped
601606	Trichoepithelioma, Multiple Familial, 1*
191100	Tuberous Sclerosis
119300	Van der Woude Syndrome
193000	Vesicoureteral Reflux 1

OMIM#	Genetic Disorder
193200	Vitiligo
193400	Von Willebrand Disease, Type 1
193510	Waardenburg Syndrome, Type 2A*
277590	Weaver Syndrome
194070	Wilms Tumor 1
	Autosomal dominant / Autosomal recessive
100100	Abdominal Muscles, Absence of, with Urinary Tract Abnormality and Cryptorchidism
107600	Aplasia Cutis Congenita, Nonsyndromic
115200	Cardiomyopathy, Dilated, 1A
613763	Cataract 16, Multiple Types*
209880	Central Hypoventilation Syndrome, Congenital
125800	Diabetes Insipidus, Nephrogenic, Autosomal
136520	Foveal Hypoplasia 1
136880	Fundus Albipunctatus
139090	Gray Platelet Syndrome
141900	Hemoglobin - Beta Locus
235400	Hemolytic Uremic Syndrome, Atypical, Susceptibility to, 1
614850	Herpes Simplex Encephalitis, Susceptibility to, 4*
256450	Hyperinsulinemic Hypoglycemia, Familial, 1
601820	Hyperinsulinemic Hypoglycemia, Familial, 2*
605389	Hypotrichosis 1
605809	Myasthenic Syndrome, Congenital, 4A, Slow-Channel*
161800	Nemaline Myopathy 3*
613038	Pituitary Hormone Deficiency, Combined, 1
180100	Retinitis Pigmentosa 1*
608133	Retinitis Pigmentosa 7*
248200	Stargardt Disease 1
613554	Von Willebrand Disease, Type 2
	X-linked recessive
300068	Androgen Insensitivity Syndrome
301800	Anus, Imperforate
301900	Borjeson-Forssman-Lehmann Syndrome*
302950	Chondrodysplasia punctata, X-linked recessive*
308350	Epileptic Encephalopathy, Early Infantile, 1
300559	Glycogen Storage Disease, Type Ixd*
306700	Hemophilia A
306900	Hemophilia B
307000	Hydrocephalus due to Congenital Stenosis of Aqueduct of Sylvius
308100	Ichthyosis, X-Linked
308240	Lymphoproliferative Syndrome, X-Linked
300486	Mental Retardation, X-Linked, with Cerebellar Hypoplasia and Distinctive Facial Appearance*

OMIM#	Genetic Disorder
310200	Muscular Dystrophy, Duchenne Type
310600	Norrie Disease
300770	Surfactant Metabolism Dysfunction, Pulmonary, 4*
313900	Thrombocytopenia 1*
300331	Thrombocytosis, Familial X-Linked*
301000	Wiskott-Aldrich Syndrome
	X-linked dominant
300908	Anemia, Nonspherocytic Hemolytic, due to G6PD Deficiency*
300896	Congenital Disorder of Glycosylation, Type 2m*
305600	Focal Dermal Hypoplasia
300624	Fragile X Mental Retardation Syndrome
308300	Incontinentia Pigmenti
300867	Kabuki Syndrome 2*
302350	Nance-Horan Syndrome
311200	Orofaciodigital Syndrome I*
312170	Pyruvate Dehydrogenase E1-Alpha Deficiency
312750	Rett Syndrome
313500	Tooth Agenesis, Selective, X-Linked, 1*
	X-linked unspecified
300100	Adrenoleukodystrophy
303800	Colorblindness, Partial, Deutan Series*
303900	Colorblindness, Partial, Protan Series*
301500	Fabry Disease
305900	Glucose-6-Phosphate Dehydrogenase
305800	Membranoproliferative Glomerulonephritis, X-Linked
	Isolated cases
108110	Arthrogryposis Multiplex Congenita
113970	Burkitt Lymphoma
190685	Down Syndrome
136760	Frontonasal Dysplasia
612776	Hypoglossia with Situs Inversus*
149000	Klippel-Trenaunay-Weber Syndrome
607432	Lissencephaly I
176270	Prader-Willi Syndrome
181000	Sarcoidosis, Susceptibility to, 1
117550	Sotos Syndrome
185300	Sturge-Weber Syndrome
188580	Thyrotoxic Periodic Paralysis
192350	VATER/VACTERL Association
	Multifactorial
605552	Abdominal Obesity-Metabolic Syndrome
603003	Bile Duct Cysts

OMIM#	Genetic Disorder
207950	Chiari Malformation Type II*
608911	Choanal Atresia, Posterior
613806	Cholangitis, Primary Sclerosing
145500	Hypertension, Essential
266600	Inflammatory Bowel Disease 1
126200	Multiple Sclerosis
177900	Psoriasis Susceptibility 1
189960	Tracheoesophageal Fistula with or without Esophageal Atresia

Unknown

250951	3-Methylglutaconic Aciduria, Type IV*
607154	Allergic Rhinitis
604919	Becker Nevus Syndrome*
109650	Behcet Syndrome
608415	Bradyopsia*
607339	Coronary Heart Disease, Susceptibility to, 1*
607907	Dermatofibrosarcoma Protuberans
613101	Hemophagocytic lymphohistiocytosis, familial, 5*
604232	Leber Congenital Amaurosis 3
613065	Leukemia, Acute Lymphoblastic
605027	Lymphoma, Non-Hodgkin, Familial
603933	Microvascular Complications of Diabetes, Susceptibility to, 1
608446	Myocardial Infarction, Susceptibility to, 1
161550	Nasopharyngeal Carcinoma
606963	Pulmonary Disease, Chronic Obstructive
613575	Retinitis Pigmentosa 55*
180300	Rheumatoid Arthritis

OMIM#	Genetic Disorder
604302	Rheumatoid Arthritis, Systemic Juvenile
269160	Schizencephaly
245050	Succinyl CoA:3-oxoacid CoA transferase deficiency*
608446	Myocardial Infarction, Susceptibility to, 1
161550	Nasopharyngeal Carcinoma
606963	Pulmonary Disease, Chronic Obstructive
613575	Retinitis Pigmentosa 55*
180300	Rheumatoid Arthritis
604302	Rheumatoid Arthritis, Systemic Juvenile
269160	Schizencephaly
245050	Succinyl CoA:3-oxoacid CoA transferase deficiency*

With Isolated Cases
With Multifactorial inheritance
With Digenic inheritance
With Somatic Mosaicism
With Mitochondrial inheritance

*CTGA disease record exclusive to KSA

Other

209850	Autism	Isolated cases; Multifactorial
516000	Complex I, Subunit ND1	Mitochondrial
133780	Exudative Vitreoretinopathy 1	Autosomal Dominant; Autosomal Recessive; X-linked
114550	Hepatocellular Carcinoma	Sporadic
146110	Hypogonadotropic Hypogonadism	Autosomal Dominant; Autosomal Recessive; X-linked
540000	Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, and Stroke-Like Episodes	Mitochondrial
253290	Multiple Pterygium Syndrome, Lethal Type	Autosomal Recessive; X-linked
254200	Myasthenia Gravis	Sporadic
254450	Myelofibrosis	Sporadic
601665	Obesity	Autosomal Dominant; Autosomal Recessive; Multifactorial
261800	Pierre Robin Syndrome	Autosomal Recessive; X-linked
176920	Proteus Syndrome	Somatic Mosaicism
187400	Testicular Torsion	Y-linked