Genetic Disorders in Arab Populations

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Since the 1950s, Arab countries have made progress in some health related aspects such as: infant mortality, life expectancy, and access to health care. Infectious diseases and nutritional disorders have decreased in prevalence because of significant advances in immunization and the overall improvement in hygiene. Arab scholars working in the field of biomedical sciences are giving more attention to the publication of their results especially on clinical and molecular findings for many genetic diseases *(Fig. 5).* Most of the local scientific studies on genetic disorders in the Arab World are conducted in laboratories of the Gulf Cooperation Council (e.g., Kingdom of Saudi Arabia and the United Arab Emirates), the Maghreb region (e.g., Tunisia and Morocco), Lebanon, and Jordan (Tadmouri and Bissar-Tadmouri, 2004).

Data from industrialized countries show that significant genetic diseases or birth defects that may affect approximately 3% of all pregnancies, account for up to 30% of pediatric hospital admissions, and cause about 50% of childhood deaths. In addition, recessively inherited disorders account for less than 20% of single gene disorders and less than 5% of congenital and genetic diseases (Alwan and Modell, 1997).

On the other hand, genetic and congenital disorders are responsible for a considerable proportion of perinatal and neonatal mortalities in Arab populations. At present, congenital malformations are the second leading cause of infant mortality in countries of the Gulf Cooperation Council, including Bahrain, Kuwait, Oman, and Qatar. Reports from Saudi Arabia indicate that congenital malformations account for about 30% of perinatal deaths (reviewed in Hamamy and Alwan, 1994). Additionally, in most Arab populations the birth prevalence of severe recessively inherited disorders may approach that of congenital malformations (Alwan and Modell, 1997).

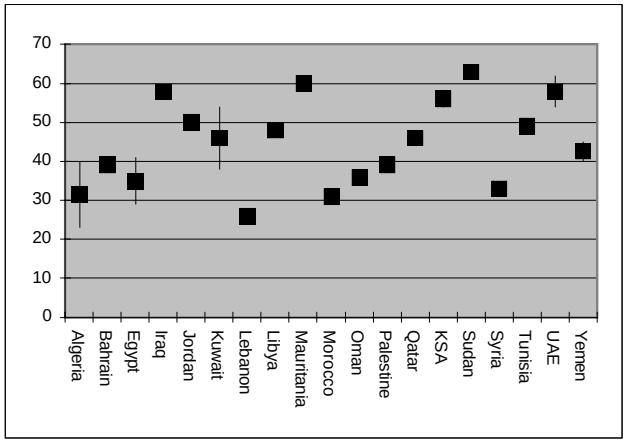


Fig. 5: Annual biomedical publication outputs in the Arab World as indexed by Medline. Data refer to articles where the first author is affiliated to an Arab Country.

In the last few years, several attempts to review different aspects of genetic diseases in Arab populations were conducted (El-Hazmi and Warsy, 1996; Alwan and Modell, 1997; Teebi and Farag, 1997). Many unique disease entries were described. Yet, data rapidly went out of date as new disorders were described in Arabs. For this reason, the Centre for Arab Genomic Studies launched a pilot project to catalogue genetic disorders described in Arab individuals. The strategy followed in this process depends on the proposal of Tadmouri and Bissar-Tadmouri (1999) to collect data at several levels. Initially, the Online Mendelian Inheritance in Man (OMIM®), a timely authoritative directory of bibliographic material and observations on inherited disorders and human genes (NCBI, 2003), was continuously mined over the last few years to collect information on genetic entities described in Arab subjects. In 1999, results of this strategy indicated the presence of 374 genetic disorders described in subjects originating from 23 Arab countries (Tadmouri and Bissar-Tadmouri, 1999). Since then, several updates on this list were conducted, using more sensitive search strategies, to exclude false positive results that are non-relevant to the topic of genetic disorders in Arab populations. In September 2004, OMIM-based search indicated the presence of 752 abnormal Mendelian characters in Arabs (Table 1).

Many of the genetic disorders in Arabs are confined to a country or region. OMIM-based results indicate that most of the genetic disorders were reported in Arabs from the Maghreb region (Tunisia, Morocco, and Algeria) as well as Lebanon and Saudi Arabia. Smaller number of genetic disorders are described in other Arab populations *(Fig. 6a).*

Table 1. Genetic disorders in Arab populations as for OMIM®

	OMIM	*Name*	*Country*
1.	100100	Abdominal Muscles, Absence of, with Urinary Tract Abnormality and Cryptorchidism	Lebanon
2.	100640	Aldehyde Dehydrogenase 1 Family, Member A1	Egypt
3.	102600	Adenine Phosphoribosyltransferase	?
4.	102610	Actin, Alpha, Skeletal Muscle 1	Oman
5.	102700	Adenosine Deaminase	KSA
6.	102730	Adenosine Deaminase, Elevated, Hemolytic Anemia due to	Libya
7.	103000	Adenylate Kinase 1	?
8.	103050	Adenylosuccinase Deficiency	Morocco
9.	103600	Albumin	Iraq
10.	104170	Alpha-Galactosidase B	Morocco
11.	104210	Alpha-2A-Adrenergic Receptor	Algeria
12.	104300	Alzheimer Disease	?
13.	105400	Amyotrophic Lateral Sclerosis 1	Tunisia
14.	106150	Angiotensin I	UAE
15.	107300	Antithrombin III Deficiency	Algeria
16.	107470	Interferon, Gamma, Receptor 1	Algeria, Tunisia, Sudan
17.	107680	Apolipoprotein A-I	UAE
18.	107730	Apolipoprotein B	?
19.	107777	Aquaporin 2	Palestine
20.	109150	Machado-Joseph Disease	Yemen
21.	109400	Basal Cell Nevus Syndrome	Egypt
22.	109535	Tumor Necrosis Factor Receptor Superfamily, Member 5	KSA
23.	110700	Blood Group--Duffy System	Yemen
24.	111000	Blood Group--Kidd System	Tunisia
25.	113100	Brachydactyly, Type C	Iraq
26.	113610	Branchial Myoclonus with Spastic Paraparesis and Cerebellar Ataxia	Kuwait
27.	113705	Breast Cancer 1 Gene	Morocco, Iraq, Yemen
28.	113900	Progressive Familial Heart Block, Type I	Lebanon
29.	114240	Calpain 3	Lebanon
30.	114830	Carbonyl Reductase 1	Sudan
31.	117000	Central Core Disease of Muscle	Algeria
32.	117550	Sotos Syndrome	?
33.	118450	Alagille Syndrome	?
34.	119100	Cleft Hand And Absent Tibia	Algeria
35.	120131	Collagen, Type IV, Alpha-4	Algeria
36.	120160	Collagen, Type I, Alpha-2	Libya, Lebanon
37.	120250	Collagen, Type VI, Alpha-3	Morocco
38.	120290	Collagen, Type XI, Alpha-2	Morocco
39.	120700	Complement Component 3	Palestine, Lebanon, Kuwait
40.	121011	Gap Junction Protein, Beta-2	Tunisia, Egypt, Palestine, Jordan

	OMIM	*Name*	*Country*
41.	122100	Corneal Dystrophy, Juvenile Epithelial, of Meesmann	KSA
42.	122470	Cornelia de Lange Syndrome	?
43.	123400	Creutzfeldt-Jakob Disease	Tunisia, Libya, Egypt
44.	124030	Cytochrome P450, Subfamily IID	KSA
45.	125630	Dermodistortive Urticaria	Lebanon
46.	125700	Diabetes Insipidus, Neurohypophyseal Type	Palestine
47.	126650	Solute Carrier Family 26, Member 3	Kuwait, KSA
48.	130070	Ehlers-Danlos Syndrome, Progeroid Form	?
49.	130500	Erythrocyte Membrane Protein Band 4.1	Algeria
50.	131244	Endothelin Receptor, Type B	Tunisia
51.	131760	Epidermolysis Bullosa Herpetiformis, Dowling-Meara Type	?
52.	133530	Excision-Repair, Complementing Defective, in Chinese Hamster, 5	Morocco
53.	134570	Factor XIII, A1 Subunit	Morocco, Syria
54.	134790	Ferritin Light Chain	Egypt
55.	135100	Fibrodysplasia Ossificans Progressiva	Tunisia, Syria
56.	136435	Follicle-Stimulating Hormone Receptor	Morocco
57.	136850	Fumarate Hydratase	Morocco
58.	137167	Gamma-Glutamyl Carboxylase	Lebanon
59.	137280	Gastritis, Familial Giant Hypertrophic	Jordan
60.	137290	Tumor-Associated Calcium Signal Transducer 2	Tunisia
61.	137800	Glioma of Brain, Familial Glioblastoma Multiforme	?
62.	138350	Glutathione S-Transferase, MU-1	KSA
63.	139090	Gray Platelet Syndrome	Palestine
64.	139191	Growth Hormone-Releasing Hormone Receptor	Morocco
65.	139250	Growth Hormone 1	KSA
66.	140300	Hashimoto Thyroiditis	Tunisia
67.	141800	Hemoglobin-Alpha Locus 1	Mauritania, Morocco, Algeria, Tunisia, KSA, Qatar, UAE
68.	141850	Hemoglobin--Alpha Locus 2	Algeria, Tunisia, Iraq, KSA, Yemen
69.	141900	Hemoglobin--Beta Locus	Morocco, Algeria, Tunisia, Egypt, Lebanon, Syria, Iraq, Kuwait, KSA, Qatar, Oman, Sudan
70.	142200	Hemoglobin, Gamma A	Tunisia
71.	142250	Hemoglobin, Gamma G	Algeria, KSA, UAE
72.	142309	Hemoglobin--Variants for which the Chain Carrying the Mutation is Unknown or Uncertain	KSA
73.	142461	Heparan Sulfate Proteoglycan of Basement Membrane	Tunisia
74.	142470	Heterocellular Hereditary Persistence of Fetal Hemoglobin	KSA
75.	142800	Major Histocompatibility Complex, Class I, A	Egypt
76.	142900	Holt-Oram Syndrome	Jordan

	OMIM	*Name*	*Country*
77.	143100	Huntington Disease	Morocco, Sudan
78.	143890	Hypercholesterolemia, Autosomal Dominant	Lebanon, Syria
79.	144200	Palmoplantar Keratoderma, Epidermolytic	Kuwait
80.	145420	Hypertelorism, Teebi Type	?
81.	145900	Hypertrophic Neuropathy of Dejerine-Sottas	Lebanon
82.	147000	IgA Constant Heavy Chain 2	Tunisia
83.	147100	IgG Heavy Chain Locus	Tunisia
84.	147120	Immunoglobulin Gm3	Tunisia
85.	147920	Kabuki Syndrome	?
86.	148820	Waardenburg Syndrome, Type III	Egypt, Yemen
87.	150330	Lamin A/C	Morocco, Algeria
88.	150590	Leg Ulcers, Familial, of Juvenile Onset	Iraq
89.	150900	Lentigines	Lebanon
90.	151443	Leukemia Inhibitory Factor Receptor	UAE, Oman
91.	151600	Leukonychia Totalis	?
92.	152200	Apolipoprotein(a)	Sudan
93.	153454	Procollagen-Lysine, 2-Oxoglutarate 5-Dioxygenase	Qatar
94.	153670	Bernard-Soulier Syndrome, Benign Autosomal Dominant	Algeria
95.	154045	Lens Intrinsic Membrane Protein 2, 19-kD	Iraq
96.	154550	Mannosephosphate Isomerase	Lebanon
97.	154570	Mannose 6-Phosphate Receptor Recognition Defect, Lebanese Type	Lebanon
98.	155255	Medulloblastoma	Morocco
99.	156225	Laminin, Alpha-2	KSA
100.	157660	Mitochondrial RNA-Processing Endoribonuclease, RNA Component of	KSA
101.	158000	Monilethrix	?
102.	159980	Myogenic Factor 4	Oman
103.	160900	Dystrophia Myotonica 1	Morocco, Egypt, Yemen
104.	161200	Nail-Patella Syndrome	Palestine
105.	161561	Interleukin 12B	KSA
106.	161900	Renal Failure, Progressive, with Hypertension	Iraq
107.	162700	Neutropenia, Chronic Familial	Yemen
108.	164500	Spinocerebellar Ataxia 7	Morocco
109.	164831	Leukemia Viral BMI-1 Oncogene, Mouse, Homolog of	Morocco
110.	166210	Osteogenesis Imperfecta Congenita	Algeria
111.	166800	Otosclerosis	Tunisia
112.	169500	Leukodystrophy, Adult-Onset, Autosomal Dominant	Sudan
113.	170200	Peptidase E	Syria
114.	170261	Transporter, ATP-Binding Cassette, Major Histocompatibility Complex, 2	Morocco
115.	173470	Integrin, Beta-3	Iraq
116.	175100	Adenomatous Polyposis of the Colon	?
117.	175200	Peutz-Jeghers Syndrome	Iraq

	OMIM	*Name*	*Country*
118.	176100	Porphyria Cutanea Tarda	Tunisia
119.	176261	Potassium Channel, Voltage-Gated, ISK-Related Subfamily, Member 1	Lebanon
120.	176640	Prion Protein	Morocco, Tunisia, Libya
121.	176670	Hutchinson-Gilford Progeria Syndrome	Libya, Egypt
122.	176860	Protein C Deficiency, Congenital Thrombotic Disease due to Protein C	?
123.	177070	Protein 4.2, Erythrocytic	Tunisia
124.	177850	Pseudoxanthoma Elasticum, Autosomal Dominant	Morocco
125.	179280	Radial-Renal Syndrome	Morocco
126.	180090	Retinaldehyde-Binding Protein	KSA
127.	180700	Robinow Syndrome	Algeria, Kuwait, KSA
128.	180860	Silver-Russell Syndrome	Lebanon
129.	180901	Ryanodine Receptor 1	Algeria
130.	180960	S-Adenosylhomocysteine Hydrolase	Tunisia
131.	181450	Ulnar-Mammary Syndrome	Yemen
132.	182380	Solute Carrier Family 5 (Sodium/Glucose Cotransporter), Member 1	Lebanon, Syria
133.	182601	Spastic Paraplegia 4, Autosomal Dominant	Tunisia
134.	182860	Spectrin, Alpha, Erythrocytic 1	Morocco, Algeria, Tunisia
135.	182870	Spectrin, Beta, Erythrocytic	Algeria
136.	183090	Spinocerebellar Ataxia 2	Tunisia
137.	184253	Spondylometaphyseal Dysplasia, Algerian Type	Algeria
138.	186970	T-Cell Antigen Receptor, Gamma Subunit	Tunisia, Lebanon
139.	187300	Telangiectasia, Hereditary Hemorrhagic, of Rendu, Osler, and Weber	?
140.	187500	Tetralogy of Fallot	Lebanon
141.	187600	Thanatophoric Dysplasia	Morocco
142.	188250	Thymidine Kinase, Mitochondrial	?
143.	188455	Thyroglossal Duct Cyst, Familial	?
144.	188540	Thyroid-Stimulating Hormone, Beta Chain	Egypt
145.	188570	Thyroid Hormone Resistance	Algeria
146.	190160	Thyroid Hormone Receptor, Beta	Algeria
147.	191315	Neurotrophic Tyrosine Kinase, Receptor, Type 1	UAE
148.	191720	5-Prime,3-Prime-@Nucleotidase, Cytosolic	Syria
149.	191740	Udp-Glycosyltransferase 1 Family, Polypeptide A1	Tunisia
150.	192340	Arginine Vasopressin	Palestine
151.	192500	Long QT Syndrome 1	Tunisia
152.	192600	Cardiomyopathy, Familial Hypertrophic	Lebanon
153.	193230	Vitreoretinal Degeneration, Snowflake Type	Algeria
154.	193700	Whistling Face-Windmill Vane Hand Syndrome	Morocco
155.	201100	Acrodermatitis Enteropathica, Zinc-Deficiency Type	Egypt, Jordan
156.	201300	Neuropathy, Hereditary Sensory and Autonomic, Type II	?
157.	201710	Lipoid Congenital Adrenal Hyperplasia	Palestine
158.	201910	Adrenal Hyperplasia, Congenital, due to 21-Hydroxylase Deficiency	Kuwait

	OMIM	*Name*	*Country*
159.	202010	Adrenal Hyperplasia, Congenital, due to 11-Beta-Hydroxylase Deficiency	Morocco, Tunisia, KSA
160.	202370	Adrenoleukodystrophy, Autosomal Neonatal Form	Egypt
161.	202400	Afibrinogenemia, Congenital	Morocco, Iraq
162.	203500	Alkaptonuria	Egypt
163.	203700	Alpers Diffuse Degeneration of Cerebral Gray Matter with Hepatic Cirrhosis	?
164.	203740	Alpha-Ketoglutarate Dehydrogenase Deficiency	Algeria, Tunisia
165.	203750	Alpha-Methylacetoaceticaciduria	Tunisia
166.	203800	Alstrom Syndrome	Algeria
167.	204200	Ceroid Lipofuscinosis, Neuronal 3, Juvenile	Morocco
168.	204500	Ceroid Lipofuscinosis, Neuronal 2, Late Infantile	Lebanon
169.	204870	Corneal Dystrophy, Gelatinous Drop-Like	Tunisia
170.	205100	Amyotrophic Lateral Sclerosis 2, Juvenile	Tunisia, Kuwait, KSA
171.	206500	Anencephaly	Iraq
172.	208050	Arterial Tortuosity	Morocco
173.	208100	Arthrogryposis Multiplex Congenita, Neurogenic Type	?
174.	208230	Arthropathy, Progressive Pseudorheumatoid, of Childhood	Jordan
175.	208250	Arthropathy-Camptodactyly Syndrome	KSA
176.	208400	Aspartylglucosaminuria Aspartylglucosaminidase	Tunisia, Palestine
177.	208850	Ataxia-Deafness-Retardation Syndrome	Kuwait
178.	208870	Ataxia-Microcephaly-Cataract Syndrome	?
179.	208900	Ataxia-Telangiectasia	Morocco
180.	209500	Atrichia with Papular Lesions	?
181.	209900	Bardet-Biedl Syndrome	Kuwait
182.	209920	Bare Lymphocyte Syndrome, Type II	Morocco, Algeria, Tunisia
183.	210000	Behr Syndrome	Iraq
184.	210200	3-@Methylcrotonyl-CoA Carboxylase 1 Deficiency	Tunisia
185.	210600	Seckel Syndrome	Iraq, Yemen
186.	210745	Blepharophimosis with Ptosis, Syndactyly, and Short Stature	Yemen
187.	211530	Bulbar Palsy, Progressive, with Sensorineural Deafness	Lebanon
188.	211770	Cahmr Syndrome	Egypt
189.	211890	Campomelia, Cumming Type	Egypt
190.	211900	Calcinosis, Tumoral, with Hyperphosphatemia	Lebanon
191.	211960	Camptodactyly with Muscular Hypoplasia, Skeletal Dysplasia, and Abnormal Palmar Creases	Morocco, Libya
192.	212110	Cardiomyopathy, Dilated, Autosomal Recessive	KSA
193.	212112	Cardiomyopathy, Congestive, with Hypergonadotropic Hypogonadism	Lebanon
194.	212135	Cardioskeletal Syndrome, Kuwaiti Type	Kuwait
195.	212138	Solute Carrier Family 25 (Carnitine/Acylcarnitine Translocase), Member 20	KSA
196.	213200	Cerebellar Ataxia 1	Lebanon
197.	213700	Cerebrotendinous Xanthomatosis	Morocco
198.	213980	Cerebrofaciothoracic Dysplasia	Morocco

	OMIM	*Name*	*Country*
199.	214100	Zellweger Syndrome	Algeria
200.	214150	Cerebrooculofacioskeletal Syndrome	Egypt
201.	214300	Cervical Vertebral Fusion, Autosomal Recessive	Iraq
202.	214400	Charcot-Marie-Tooth Disease, Type 4A	Tunisia
203.	214700	Chloride Diarrhea, Familial	Kuwait
204.	215518	Ciliary Discoordination due to Random Ciliary Orientation	Lebanon
205.	216550	Cohen Syndrome	Lebanon
206.	216900	Achromatopsia 2	Morocco, Iraq
207.	217070	Complement Component 7 Deficiency	Morocco, Tunisia, Yemen
208.	217080	Cone-Rod Dystrophy and Amelogenesis Imperfecta	?
209.	217400	Corneal Dystrophy and Perceptive Deafness	Morocco
210.	218350	Craniofacial Dyssynostosis with Short Stature	?
211.	219200	Cutis Laxa with Growth and Developmental Delay	KSA
212.	219550	Cysteine Peptiduria	Iraq
213.	219600	Cystic Disease of Lung	Yemen
214.	219700	Cystic Fibrosis	Morocco, Libya, Iraq, KSA, Yemen
215.	219721	Cystic Fibrosis with Helicobacter Pylori Gastritis, Megaloblastic Anemia, and Subnormal Mentality	?
216.	220100	Cystinuria	Libya
217.	220150	Hypouricemia, Renal	Iraq
218.	220290	Deafness, Neurosensory, Autosomal Recessive 1	Tunisia, Egypt, Palestine
219.	220500	Deafness, Congenital, and Onychodystrophy, Recessive Form	Oman
220.	220900	Deafness, Congenital, with Total Albinism	Morocco
221.	221745	Mitochondrial Deafness Modifier Gene 1	?
222.	221950	Dextrocardia with Unusual Facies and Microphthalmia	?
223.	222300	Wolfram Syndrome	Sudan
224.	222400	Diaphragm, Unilateral Agenesis of Diaphragmatic Defects, Familial Congenital	Egypt, Kuwait
225.	222448	Donnai-Barrow Syndrome	KSA
226.	222700	Lysinuric Protein Intolerance	Morocco, KSA
227.	222748	Dihydropyrimidinase	Lebanon
228.	223800	Dyggve-Melchior-Clausen Disease	Morocco, Lebanon
229.	223900	Dysautonomia, Familial	Algeria
230.	224120	Anemia, Dyserythropoietic Congenital, Type I	Kuwait, KSA
231.	224230	Dyskeratosis Congenita, Autosomal Recessive	Syria
232.	224400	Dyssegmental Dwarfism	Palestine, Lebanon, Jordan
233.	224500	Dystonia Musculorum Deformans 2	?
234.	224900	Ectodermal Dysplasia, Anhidrotic	Morocco
235.	225280	Eem Syndrome	Yemen
236.	225400	Ehlers-Danlos Syndrome, Type VI	?
237.	225750	Aicardi-Goutieres Syndrome 1	Algeria

	OMIM	*Name*	*Country*
238.	226300	Enteropathy, Protein-Losing	?
239.	226400	Epidermodysplasia Verruciformis	Algeria
240.	226980	Epiphyseal Dysplasia, Multiple, with Early-Onset Diabetes Mellitus	Oman
241.	227090	Erythroderma, Lethal Congenital	Oman
242.	227260	Facial Ectodermal Dysplasia	Oman
243.	227300	Factor V and Factor VIII, Combined Deficiency of	Algeria, Tunisia
244.	227310	Factor V and Factor VIII, Combined Deficiency of, with Normal Protein C and Protein C Inhibitor	Syria
245.	227320	Faciothoracogenital Syndrome	?
246.	227330	Faciodigitogenital Syndrome, Recessive	Kuwait
247.	227400	Factor V Deficiency	Morocco
248.	227500	Factor VII Deficiency	Morocco
249.	227645	Fanconi Anemia, Complementation Group C	Iraq
250.	228000	Farber Lipogranulomatosis	Tunisia
251.	228250	Femur, Unilateral Bifid, with Monodactylous Ectrodactyly	?
252.	228400	Fever, Familial Lifelong Persistent	Lebanon
253.	228550	Fibromatosis, Congenital Generalized	Morocco
254.	228980	Fleck Retina, Familial Benign	Palestine
255.	229200	Fragilitas Oculi with Joint Hyperextensibility	Tunisia, Syria
256.	229300	Friedreich Ataxia 1	Tunisia
257.	229800	Fructosuria Ketohexokinase	?
258.	229850	Fryns Syndrome	Sudan
259.	230740	Gapo Syndrome	Algeria, Egypt
260.	230800	Gaucher Disease, Type I	Palestine
261.	231005	Gaucher-Like Disease	?
262.	231070	Geroderma Osteodysplastica	Morocco
263.	231090	Hydatidiform Mole	Lebanon
264.	231550	Achalasia-Addisonianism-Alacrima Syndrome	KSA
265.	231670	Glutaricacidemia I Glutaryl-CoA Dehydrogenase	?
266.	232200	Glycogen Storage Disease I Glucose-6-Phosphatase, Catalytic	?
267.	232300	Glycogen Storage Disease II	Palestine
268.	232700	Glycogen Storage Disease VI Glycogen Phosphorylase, Liver	?
269.	233710	Granulomatous Disease, Chronic, Autosomal Cytochrome-b-Positive Form II	Palestine, Jordan
270.	234050	Hair-Brain Syndrome	Morocco
271.	235200	Hemochromatosis	Algeria, Egypt
272.	235510	Hennekam Lymphangiectasia-Lymphedema Syndrome	?
273.	236200	Homocystinuria	KSA
274.	236450	Hutterite Cerebroosteonephrodysplasia Syndrome	Yemen
275.	236600	Hydrocephalus	Palestine, Kuwait
276.	236700	McKusick-Kaufman Syndrome	Lebanon
277.	236730	Urofacial Syndrome	?

	OMIM	*Name*	*Country*
278.	236792	L-2-@Hydroxyglutaricacidemia	Morocco, Tunisia
279.	236800	Hydroxykynureninuria	Algeria
280.	237300	Carbamoyl Phosphate Synthetase I Deficiency, Hyperammonemia due to	?
281.	237500	Dubin-Johnson Syndrome	Morocco, Iraq
282.	237900	Hyperbilirubinemia, Transient Familial Neonatal Breastfeeding Jaundice	Yemen
283.	238310	Aminomethyltransferase	?
284.	239000	Paget Disease, Juvenile	Iraq
285.	239500	Hyperprolinemia, Type I	Algeria
286.	239710	Acrofrontofacionasal Dysostosis, Severe	Kuwait
287.	239840	Hypertrichosis, Congenital Anterior Cervical, with Peripheral Sensory and Motor Neuropathy	?
288.	241080	Hypogonadism, Diabetes Mellitus, Alopecia, Mental Retardation, and Electrocardiographic Abnormalities	KSA
289.	241410	Hypoparathyroidism-Retardation-Dysmorphism Syndrome	KSA, Qatar
290.	242300	Ichthyosis, Lamellar, 1	Egypt
291.	242870	Immunodeficiency, Partial Combined, with Absence of HLA Determinants and Beta-2-Microglobulin from Lymphocytes	Algeria
292.	243060	Infertility Associated with Multi-Tailed Spermatozoa and Excessive DNA	Libya
293.	243110	Interleukin 1, Defective T-Cell Response to	Lebanon
294.	243320	Intrinsic Factor and R Binder, Combined Congenital Deficiency of	Algeria
295.	243600	Jejunal Atresia	?
296.	243800	Johanson-Blizzard Syndrome	KSA
297.	244400	Kartagener Syndrome	?
298.	244460	Kenny-Caffey Syndrome, Type 1	Kuwait, KSA
299.	245000	Papillon-Lefevre Syndrome	Jordan
300.	245200	Krabbe Disease	Egypt, Lebanon, Syria
301.	245552	Lambotte Syndrome	Morocco
302.	245590	Laron Syndrome, Type II Growth Hormone Insensitivity with Immunodeficiency	Palestine
303.	245600	Larsen Syndrome, Recessive	UAE
304.	245800	Laurence-Moon Syndrome	Kuwait
305.	246200	Leprechaunism	Yemen
306.	246450	3-@Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency	Morocco, KSA
307.	247100	Lipoid Proteinosis of Urbach and Wiethe	Lebanon
308.	248110	Macrosomia with Microphthalmia, Lethal	Kuwait
309.	248250	Hypomagnesemia, Primary	?
310.	248300	Mal de Meleda	Algeria
311.	248500	Mannosidosis, Alpha B, Lysosomal	Palestine
312.	248600	Maple Syrup Urine Disease	Tunisia, Egypt
313.	248800	Marinesco-Sjogren Syndrome	Kuwait
314.	249100	Familial Mediterranean Fever	Morocco, Libya, Palestine, Lebanon, Syria, Jordan, Iraq, Kuwait, KSA

	OMIM	*Name*	*Country*
315.	249240	Megalencephaly with Dysmyelination	Iraq
316.	249270	Thiamine-Responsive Megaloblastic Anemia Syndrome	?
317.	249420	Ter Haar Syndrome	Syria
318.	249500	Mental Retardation, Autosomal Recessive	Algeria
319.	250100	Metachromatic Leukodystrophy Pseudoarylsulfatase A Deficiency	?
320.	250220	Metaphyseal Chondrodysplasia, Congenital Lethal	Yemen
321.	250450	Metaphyseal Dysplasia, Anetoderma, and Optic Atrophy	Egypt
322.	250790	Methemoglobinemia due to Deficiency of Cytochrome B5	Yemen
323.	250800	Methemoglobinemia due to Deficiency of Methemoglobin Reductase	Algeria
324.	250951	3-@Methylglutaconicaciduria, Type IV	Iraq
325.	251260	Nijmegen Breakage Syndrome Berlin Breakage Syndrome	?
326.	251270	Microcephaly with Chorioretinopathy	Kuwait
327.	251280	Microcephaly with Spastic Quadriplegia	Palestine
328.	251450	Desbuquois Syndrome	Morocco, UAE
329.	251600	Microphthalmos, Autosomal Recessive	?
330.	251850	Microvillus Inclusion Disease	Iraq
331.	252010	Complex I, Mitochondrial Respiratory Chain, Deficiency of Myopathy, Mitochondrial, with Deficiency of Respiratory Chain NADH-CoQ Reductase Activity	Morocco
332.	252350	Moyamoya Disease	?
333.	252650	Mucolipidosis IV	Sudan
334.	252800	Alpha-L-Iduronidase	Morocco, Libya, Egypt, Lebanon, Syria
335.	252920	Mucopolysaccharidosis Type IIIb N-Acetylglucosaminidase, Alpha-	?
336.	253220	Mucopolysaccharidosis Type VII	Algeria
337.	253250	Mulibrey Nanism	Egypt
338.	253270	Multiple Carboxylase Deficiency	Jordan
339.	253290	Multiple Pterygium Syndrome, Lethal Type	Morocco
340.	253300	Spinal Muscular Atrophy, Type I	Kuwait
341.	253550	Spinal Muscular Atrophy, Type II	Kuwait
342.	253600	Muscular Dystrophy, Limb-Girdle, Type 2A	?
343.	253601	Muscular Dystrophy, Limb-Girdle, Type 2B	Yemen
344.	253700	Muscular Dystrophy, Limb-Girdle, Type 2C	Morocco, Algeria, Tunisia, Libya, Egypt
345.	254130	Miyoshi Myopathy	Tunisia
346.	254210	Myasthenia Gravis, Familial Infantile	Iraq
347.	254780	Myoclonic Epilepsy of Lafora Epilepsy	Palestine
348.	254800	Myoclonic Epilepsy of Unverricht and Lundborg	?
349.	255800	Schwartz-Jampel Syndrome, Type 1	Algeria, Tunisia, UAE
350.	256000	Leigh Syndrome	Mauritania
351.	256020	Nail-Patella-Like Renal Disease	Palestine
352.	256370	Nephrotic Syndrome, Early-Onset, with Diffuse Mesangial Sclerosis	?

	OMIM	*Name*	*Country*
353.	256450	Nesidioblastosis of Pancreas Persistent Hyperinsulinemic Hypoglycemia of Infancy due to Focal Adenomatous Hyperplasia	KSA
354.	256520	Neu-Laxova Syndrome	Egypt
355.	256800	Insensitivity to Pain, Congenital, with Anhidrosis	Kuwait
356.	256850	Giant Axonal Neuropathy 1	Tunisia
357.	256851	Neuropathy, Giant Axonal, Tunisian Form	Tunisia
358.	257300	Nondisjunction	Kuwait
359.	257320	Lissencephaly Syndrome, Norman-Roberts Type	KSA
360.	257980	Odontoonychodermal Dysplasia	Lebanon
361.	258501	3-@Methylglutaconicaciduria, Type III	Iraq
362.	258860	Oral-Facial-Digital Syndrome, Type IV	Lebanon
363.	258870	Ornithine Aminotransferase Deficiency	Algeria, Lebanon, Iraq
364.	259700	Osteopetrosis, Autosomal Recessive	Palestine
365.	259730	Osteopetrosis with Renal Tubular Acidosis	Tunisia, Kuwait, KSA
366.	259775	Osteoclerotic Bone Dysplasia, Lethal	?
367.	260600	Pelizaeus-Merzbacher Disease, Acute Infantile Type	Yemen
368.	260650	Pellagra-Like Syndrome	Sudan
369.	260800	Pentosuria	Lebanon
370.	260920	Hyper-IgD Syndrome	?
371.	261100	Megaloblastic Anemia 1	KSA
372.	261500	Peroxidase and Phospholipid Deficiency in Eosinophils	Yemen, Sudan
373.	261550	Persistent Mullerian Duct Syndrome, Types I and II	?
374.	261600	Phenylketonuria	Kuwait, Yemen
375.	261630	Phenylketonuria II	Tunisia
376.	261750	Phosphorylase Kinase Deficiency of Liver and Muscle, Autosomal Recessive	?
377.	262400	Pituitary Dwarfism I	Iraq, Yemen
378.	262500	Pituitary Dwarfism II	?
379.	263630	Polysyndactyly with Cardiac Malformation	Oman
380.	263650	Popliteal Pterygium Syndrome, Lethal Type	Qatar, UAE
381.	263700	Porphyria, Congenital Erythropoietic	Algeria
382.	264300	17-@Beta Hydroxysteroid Dehydrogenase III Deficiency Polycystic Ovarian Disease due to 17-Ketosteroid Reductase Deficiency	?
383.	264600	Pseudovaginal Perineoscrotal Hypospadias	Jordan
384.	264900	Pta Deficiency Coagulation Factor XI	Iraq
385.	265000	Pterygium Syndrome	Kuwait
386.	265100	Pulmonary Alveolar Microlithiasis	Lebanon
387.	265380	Pulmonary Hypertension, Familial Persistent, of the Newborn	Tunisia
388.	265800	Pycnodysostosis	?
389.	265950	Pyloric Atresia	Iraq
390.	266140	Pyropoikilocytosis, Hereditary	KSA
391.	266150	Pyruvate Carboxylase Deficiency	Egypt

	OMIM	*Name*	*Country*
392.	266200	Pyruvate Kinase Deficiency of Erythrocyte	Lebanon
393.	266265	Congenital Disorder of Glycosylation, Type IIc	?
394.	267000	Renal Hamartomas, Nephroblastomatosis, and Fetal Gigantism	Yemen
395.	267430	Renal Tubular Dysgenesis	Palestine
396.	267700	Reticulosis, Familial Histiocytic	Iraq
397.	268020	Retinitis Pigmentosa, Deafness, Mental Retardation, and Hypogonadism	Morocco
398.	268050	Retinopathy, Pigmentary, and Mental Retardation	Lebanon
399.	268130	Revesz Syndrome	Sudan
400.	268200	Rhabdomyolysis, Acute Recurrent	Kuwait
401.	268250	Rhizomelic Syndrome	?
402.	268310	Robinow Syndrome, Autosomal Recessive	Kuwait, Oman
403.	268800	Sandhoff Disease	Lebanon
404.	269000	SC Phocomelia Syndrome	Lebanon
405.	269150	Schinzel-Giedion Midface-Retraction Syndrome	Egypt
406.	269700	Lipodystrophy, Congenital Generalized, Type 2	Lebanon, Oman
407.	269950	Sideroblastic Anemia, Autosomal	Libya
408.	270200	Sjogren-Larsson Syndrome	Egypt
409.	270300	Skin Peeling, Familial Continuous	Kuwait
410.	270550	Spastic Ataxia, Charlevoix-Saguenay Type	Tunisia
411.	270750	Spastic Paraplegia 23	Jordan
412.	270800	Spastic Paraplegia 5A, Autosomal Recessive	Tunisia
413.	271322	Spinocerebellar Degeneration with Slow Eye Movements	Palestine, Kuwait
414.	271550	Spondyloenchondrodysplasia	Iraq
415.	271640	Spondyloepimetaphyseal Dysplasia with Joint Laxity	?
416.	271665	Spondylometaepiphyseal Dysplasia, Short Limb-Hand Type	Egypt
417.	271900	Canavan Disease	KSA
418.	272300	Sulfocysteinuria	Algeria
419.	272440	Syndactyly, Type I, with Microcephaly and Mental Retardation	Tunisia
420.	272450	Syndesmodysplasic Dwarfism	Algeria
421.	272460	Spondylocarpotarsal Synostosis Syndrome	Lebanon
422.	272750	Tay-Sachs Disease, AB Variant	KSA
423.	272800	Tay-Sachs Disease	Morocco, Lebanon, Syria
424.	272950	Teebi-Shaltout Syndrome	Tunisia
425.	273150	Testes, Rudimentary	Lebanon
426.	273250	Testicular Regression Syndrome	Tunisia
427.	273395	Tetra-Amelia with Pulmonary Hypoplasia	Palestine
428.	273800	Thrombasthenia of Glanzmann and Naegeli	Jordan, Iraq
429.	275000	Graves Disease	Tunisia
430.	275200	Thyrotropin, Unresponsiveness to	Egypt, Syria
431.	275210	Tight Skin Contracture Syndrome, Lethal	Algeria
432.	275350	Transcobalamin II Deficiency	Morocco

	OMIM	*Name*	*Countr*
433.	275595	Trigonobrachycephaly, Bulbous Bifid Nose, Micrognathia, and Abnormalities of the Hands and Feet	Palestine
434.	275630	Triglyceride Storage Disease with Impaired Long-Chain Fatty Acid Oxidation	Egypt
435.	275900	Spastic Paraplegia 20, Autosomal Recessive	Kuwait
436.	276600	Tyrosine Transaminase Deficiency	KSA
437.	276820	Ulna And Fibula, Absence of, with Severe Limb Deficiency	Jordan
438.	276821	Ulnar Hypoplasia with Mental Retardation	?
439.	276901	Usher Syndrome, Type IIA	Tunisia
440.	276902	Usher Syndrome, Type III	Yemen
441.	276903	Myosin VIIA	Tunisia, Yemen
442.	276905	Usher Syndrome, Type IIB	Tunisia
443.	277300	Spondylocostal Dysostosis, Autosomal Recessive, 1	?
444.	277320	Visceral Myopathy, Familial, with External Ophthalmoplegia	Iraq
445.	277350	Vitamin A Metabolic Defect	Lebanon
446.	277440	Vitamin D-Resistant Rickets with End-Organ Unresponsiveness to 1,25-Dihydroxycholecalciferol	?
447.	277450	Vitamin K-Dependent Clotting Factors, Combined Deficiency of, 1 Chondrodysplasia Punctata with Coagulation Factor Deficiency	?
448.	277460	Vitamin E, Familial Isolated Deficiency of	Tunisia
449.	277465	Vitiligo, Progressive, with Mental Retardation and Urethral Duplication	Algeria
450.	277580	Waardenburg-Shah Syndrome	Yemen
451.	277600	Weill-Marchesani Syndrome, Autosomal Recessive	Lebanon
452.	277900	Wilson Disease	?
453.	278250	Wrinkly Skin Syndrome	Iraq, KSA
454.	278300	Xanthinuria, Type I	Lebanon, Kuwait
455.	278700	Xeroderma Pigmentosum, Complementation Group A	Tunisia, Egypt, Palestine
456.	279000	Young Syndrome	Yemen
457.	300265	Zinc Finger Protein of Cerebellum, 3	Lebanon
458.	300331	Thrombocytosis, Familial X-Linked	?
459.	300383	Properdin P Factor, Complement	Tunisia
460.	300392	WAS Gene	Lebanon, Syria
461.	300419	Mental Retardation, X-Linked 54	Tunisia
462.	300463	Polyglutamine-Binding Protein 1	Morocco
463.	301090	Amelia, X-Linked	?
464.	301900	Borjeson-Forssman-Lehmann Syndrome	KSA
465.	304790	Immunodysregulation, Polyendocrinopathy, and Enteropathy, X-Linked	Morocco
466.	305900	Glucose-6-Phosphate Dehydrogenase	Algeria, Egypt, Lebanon, Syria, Jordan, Iraq, KSA, Sudan
467.	308050	Congenital Hemidysplasia with Ichthyosiform Erythroderma and Limb Defects	Egypt
468.	309200	Major Affective Disorder 2	Iraq, Yemen

	OMIM	*Name*	*Country*
469.	309550	Fragile Site Mental Retardation 1 Gene	Tunisia
470.	309900	Mucopolysaccharidosis Type II	Morocco
471.	310400	Myotubular Myopathy 1	Yemen
472.	312060	Properdin Deficiency, X-Linked	Tunisia
473.	312550	Retinal Dysplasia, Primary	Iraq
474.	313700	Androgen Receptor	Kuwait
475.	313900	Thrombocytopenia 1	KSA
476.	561000	Ribosomal RNA, Mitochondrial, 12S	?
477.	580000	Streptomycin Ototoxicity	?
478.	600060	Deafness, Neurosensory, Autosomal Recessive 2	Tunisia
479.	600116	Parkinson Disease, Juvenile, Autosomal Recessive	Algeria
480.	600118	Warburg Micro Syndrome 1	Lebanon
481.	600146	Spastic Paraplegia 5B, Autosomal Recessive	Tunisia
482.	600160	Cyclin-Dependent Kinase Inhibitor 2A	Tunisia
483.	600179	Guanylate Cyclase 2D, Membrane	Algeria, Tunisia
484.	600185	Breast Cancer 2 Gene	Yemen
485.	600252	Lowry-Maclean Syndrome	Kuwait
486.	600262	Prostaglandin-Endoperoxide Synthase 2	Algeria
487.	600266	Solute Carrier Family 11 (Proton-Coupled Divalent Metal Ion Transporter), Member 1	Sudan
488.	600301	Acyl-CoA Dehydrogenase, Short/Branched Chain	Eritrea
489.	600360	Aplasia Cutis Congenita of Limbs, Recessive	Yemen
490.	600374	BBS4 Gene	KSA
491.	600502	Immunoglobulin MU Binding Protein 2	Lebanon
492.	600509	ATP-Binding Cassette, Subfamily C, Member 8	KSA
493.	600514	Reelin	KSA
494.	600529	AU-Specific RNA-Binding Protein	Morocco, Lebanon
495.	600617	Steroidogenic Acute Regulatory Protein	Palestine, Jordan, Kuwait
496.	600650	Carnitine Palmitoyltransferase II	Morocco
497.	600662	Mads Box Transcription Enhancer Factor 2, Polypeptide C	Oman
498.	600737	Inclusion Body Myopathy 2, Autosomal Recessive	Palestine
499.	600760	Sodium Channel, Nonvoltage-Gated 1, Beta Subunit	?
500.	600794	Spinal Muscular Atrophy, Distal, Type V	Algeria
501.	600805	Laminin, Alpha-3	KSA
502.	600818	Transgelin	Algeria
503.	600850	Schizophrenia 4	Algeria
504.	600863	Casein Kinase I, Epsilon	Syria
505.	600900	Sarcoglycan, Beta	Tunisia
506.	600918	Cystinuria, Type III	Libya
507.	600923	Protoporphyrinogen Oxidase	Lebanon
508.	600957	Anti-Mullerian Hormone	Morocco

	OMIM	*Name*	*Country*
509.	601007	Leptin Receptor	Algeria
510.	601015	NPC2 Gene	Algeria
511.	601067	Usher Syndrome, Type ID	Morocco
512.	601071	Deafness, Autosomal Recessive 9	Lebanon
513.	601105	Cathepsin K	Morocco
514.	601107	ATP-Binding Cassette, Subfamily C, Member 2	Morocco
515.	601145	Cystatin B	?
516.	601146	Growth/Differentiation Factor 5	Oman
517.	601170	Muscular Dystrophy, Congenital, with Severe Central Nervous System Atrophy and Absence of Large Myelinated Fibers	?
518.	601214	Naxos Disease	?
519.	601277	Ichthyosis, Lamellar, 2	Morocco
520.	601386	Deafness, Autosomal Recessive 12	Syria
521.	601441	Diacylglycerol Kinase, Zeta, 104-kD	Jordan
522.	601451	Nevo Syndrome	?
523.	601537	Microcephaly, Retinitis Pigmentosa, and Sutural Cataract	Morocco
524.	601549	Alacrima	Jordan
525.	601552	Ectopia Lentis, Spontaneous Filtering Blebs, and Craniofacial Dysmorphism	Lebanon
526.	601553	Hypotrichosis, Congenital, with Juvenile Macular Dystrophy	Egypt
527.	601559	Stuve-Wiedemann Syndrome/Schwartz-Jampel Syndrome, Type 2	UAE, Oman
528.	601596	Charcot-Marie-Tooth Disease, Type 4C	Algeria
529.	601601	Transcription Factor AP2-Beta	Palestine
530.	601604	Interleukin 12 Receptor, Beta-1	Morocco
531.	601618	SRY-Box 18	Oman
532.	601623	Ubiquitin-Protein Ligase E3A	Iraq
533.	601691	ATP-Binding Cassette, Subfamily A, Member 4	KSA
534.	601706	Yemenite Deaf-Blind Hypopigmentation Syndrome	Yemen
535.	601769	Vitamin D Receptor	KSA
536.	601771	Cytochrome P450, Subfamily I, Polypeptide 1	Morocco, Algeria, KSA
537.	601843	Solute Carrier Family 5 (Sodium Iodide Symporter), Member 5	Algeria
538.	601993	Nuclear Receptor Coactivator 2	Oman
539.	602078	Fibrosis of Extraocular Muscles, Congenital, 2	KSA
540.	602097	Usher Syndrome, Type IE	Morocco
541.	602099	Amyotrophic Lateral Sclerosis 5	Tunisia
542.	602109	Matrilin 3	?
543.	602201	Extracellular Matrix Protein 1	Kuwait
544.	602229	SRY-Box 10	Yemen
545.	602247	Xanthomatosis, Susceptibility to	Syria
546.	602302	Hairless, Mouse, Homolog of	Palestine, Oman
547.	602337	Receptor Tyrosine Kinase-Like Orphan Receptor 2	Oman
548.	602400	Ichthyosis, Follicular Atrophoderma, Hypotrichosis, and Hypohidrosis	UAE

	OMIM	*Name*	*Country*
549.	602401	Ectodermal Dysplasia, Hidrotic, Autosomal Recessive	Lebanon
550.	602419	Early Growth Response 3	Syria
551.	602421	Cystic Fibrosis Transmembrane Conductance Regulator	?
552.	602459	Deafness, Autosomal Dominant Nonsyndromic Sensorineural 15	Tunisia, Libya, Egypt
553.	602544	Parkin	Algeria
554.	602557	Spondyloepimetaphyseal Dysplasia, Shohat Type	Iraq
555.	602574	Tectorin, Alpha	Lebanon
556.	602593	Corneodesmosin	Yemen
557.	602643	Tumor Necrosis Factor Receptor Superfamily, Member 11B	Iraq
558.	602753	Aristaless Homeo Box, Drosophila, Homolog of	KSA
559.	602768	Delta-Like 3	?
560.	602956	X-Ray Repair, Complementing Defective, in Chinese Hamster, 9	Lebanon
561.	602976	Max-Like Protein X	Egypt
562.	603009	Dysferlin Libya, Palestine,	Yemen
563.	603033	Collagenic Tail of Endplate Acetylcholinesterase	Palestine, Iraq
564.	603034	Endplate Acetylcholinesterase Deficiency	Palestine, Iraq
565.	603098	Deafness, Autosomal Recessive 13	Lebanon
566.	603133	Dislocated Elbows, Bowed Tibias, Scoliosis, Deafness, Cataract, Microcephaly, and Mental Retardation	Lebanon
567.	603194	Meckel Syndrome, Type 2	Tunisia
568.	603266	Diabetes Mellitus, Insulin-Dependent, 17	?
569.	603335	Dynein, Axonemal, Heavy Chain 5	Lebanon
570.	603400	WNT1-Inducible Signaling Pathway Protein 3	Jordan, KSA
571.	603438	Radioulnar Synostosis with Short Stature, Microcephaly, Scoliosis, and Mental Retardation	?
572.	603552	Hemophagocytic Lymphohistiocytosis, Familial, 1	KSA
573.	603554	Omenn Syndrome	Morocco
574.	603557	Myotubularin-Related Protein 2	KSA
575.	603593	Solute Carrier Family 7, Member 7	Tunisia
576.	603629	Deafness, Autosomal Recessive 21	Lebanon
577.	603642	Atrial Septal Defect, Secundum, with Various Cardiac and Noncardiac Defects	Lebanon
578.	603650	Bardet-Biedl Syndrome 5	Kuwait, KSA
579.	603678	Deafness, Autosomal Recessive 14	Lebanon
580.	603681	Otoferlin	Lebanon
581.	603707	Molybdenum Cofactor Synthesis 1	?
582.	603708	Molybdenum Cofactor Synthesis 2	Egypt
583.	603720	Deafness, Autosomal Recessive 16	Palestine, Syria
584.	603813	Hypercholesterolemia, Autosomal Recessive	Syria
585.	603824	UDP-N-Acetylglucosamine 2-Epimerase/N-Acetylmannosamine Kinase	Palestine
586.	603868	Ras-Associated Protein RAB27A	?
587.	603896	Leukoencephalopathy with Vanishing White Matter	Algeria

	OMIM	*Name*	*Country*
588.	603903	Sickle Cell Anemia	Oman
589.	603968	Polymerase, DNA, ETA	Lebanon
590.	604032	Eukaryotic Translation Initiation Factor 2-Alpha Kinase 3	Tunisia
591.	604110	G Protein-Coupled Receptor 56	Palestine, Qatar
592.	604144	Solute Carrier Family 7, Member 9	Libya
593.	604161	G Protein-Coupled Receptor 54	KSA
594.	604201	Hepatic Fibrosis, Severe, Susceptibility to, due to Schistosoma Mansoni Infection	Sudan
595.	604228	Partial Albinism and Immunodeficiency Syndrome	KSA
596.	604232	Leber Congenital Amaurosis, Type III	KSA
597.	604320	Spinal Muscular Atrophy with Respiratory Distress 1	Lebanon
598.	604321	Microcephaly, Primary Autosomal Recessive, 4	Morocco
599.	604327	Xylosylprotein 4-Beta-Galactosyltransferase, Polypeptide 7	?
600.	604490	Sacsin	Tunisia
601.	604559	Progressive Familial Heart Block, Type I, Locus 1	Lebanon
602.	604563	Charcot-Marie-Tooth Disease, Type 4B2	Tunisia
603.	604571	Bare Lymphocyte Syndrome, Type I	Morocco
604.	604595	Cholesterol-Lowering Factor	Syria
605.	604611	RECQ Protein-Like 2	Syria
606.	604780	Comparative Gene Identification 58	Morocco, Algeria, Tunisia
607.	604801	Muscular Dystrophy, Congenital, 1B	UAE
608.	604934	Tubulin-Specific Chaperone E	?
609.	605156	Nodulosis-Arthropathy-Osteolysis Syndrome	KSA
610.	605195	Mesoderm Posterior 2	Lebanon
611.	605203	Multiple Pterygium Syndrome, Aslan Type	Qatar
612.	605225	Inflammatory Bowel Disease 7	Iraq
613.	605239	ATPase, H+ Transporting, Lysosomal, V0 Subunit A, Isoform 4	KSA
614.	605242	USH1C Gene	Lebanon
615.	605248	Mucolipin 1	?
616.	605316	Deafness, Congenital Neurosensory, Autosomal Recessive 10	Palestine
617.	605378	Aladin	Algeria
618.	605379	Gigaxonin	Tunisia
619.	605511	Transmembrane Protease, Serine 3	Tunisia, Palestine
620.	605573	17-@Beta Hydroxysteroid Dehydrogenase III	Syria
621.	605588	Charcot-Marie-Tooth Disease, Axonal, Type 2B1	Morocco, Algeria
622.	605597	Forkhead Transcription Factor FOXL2	Algeria
623.	605646	Solute Carrier Family 26, Member 4	Tunisia
624.	605678	Williams-Beuren Syndrome Chromosome Region 14	Egypt
625.	605685	Cutis Verticis Gyrata, Retinitis Pigmentosa, and Sensorineural Deafness	Lebanon
626.	605725	Periaxin	Lebanon
627.	605726	Neuropathy, Distal Hereditary Motor, Jerash Type	Jordan
628.	605747	Autosomal Recessive Hypercholesterolemia Gene	Lebanon, Syria

	OMIM	*Name*	*Country*
629.	605799	Amnionless, Mouse, Homolog of	Tunisia
630.	605818	Deafness, Autosomal Recessive 27	UAE
631.	605822	Spondyloocular Syndrome, Autosomal Recessive	Iraq
632.	605828	Epidermodysplasia Verruciformis Gene 1	Algeria
633.	605829	Epidermodysplasia Verruciformis Gene 2	Algeria
634.	605881	GDP-Fucose Transporter 1	?
635.	605899	Glycine Encephalopathy	?
636.	605908	Megalencephalic Leukoencephalopathy with Subcortical Cysts Gene 1	Libya
637.	606054	Propionicacidemia	KSA
638.	606119	Secreted Ly6/uPAR-Related Protein 1	Algeria, Tunisia, Palestine, UAE
639.	606125	Tripartite Motif-Containing Protein 8	Egypt
640.	606158	BerardinellI-Seip Congenital Lipodystrophy Gene 2	Lebanon
641.	606201	Wolfram Syndrome Gene 1	KSA
642.	606220	Mental Retardation, Short Stature, Facial Anomalies, and Joint Dislocations	Lebanon
643.	606324	Parkinson Disease, Type 7, Autosomal Recessive Early-Onset	Lebanon
644.	606352	Alsin	Tunisia, Kuwait
645.	606397	USH3A Gene	Yemen
646.	606412	BSND Gene	Lebanon
647.	606416	CIAS1 Gene	Algeria
648.	606438	Huntington Disease-Like 2	Morocco, KSA
649.	606463	Glucosidase, Acid Beta	Lebanon, KSA
650.	606527	Megarbane Syndrome	Iraq
651.	606530	Cytochrome P450, Subfamily XXVIIa, Polypeptide 1	Morocco
652.	606545	Ichthyosis, Lamellar, 5	?
653.	606555	Tripartite Motif-Containing Protein 9	Egypt
654.	606556	Tripartite Motif-Containing Protein 14	Egypt
655.	606559	Tripartite Motif-Containing Protein 22	Egypt
656.	606580	Optic Atrophy 3 Gene	Iraq
657.	606596	Fukutin-Related Protein	Algeria, Tunisia, Libya
658.	606597	Paired Box Gene 3	Yemen
659.	606598	Ganglioside-Induced Differentiation-Associated Protein 1	Morocco, Tunisia
660.	606612	Muscular Dystrophy, Congenital, 1C	Algeria, Tunisia
661.	606693	Parkinson Disease 9	Jordan
662.	606709	Protease, Serine, 12	Algeria
663.	606744	Seckel Syndrome 2	Iraq
664.	606808	Myosin IIIA	Iraq
665.	606810	Proline Dehydrogenase	Algeria
666.	606824	Glucose/Galactose Malabsorption	Iraq
667.	606844	Alstrom Syndrome Gene	Algeria
668.	606854	Polymicrogyria, Bilateral Frontoparietal	Palestine

	OMIM	*Name*	*Country*
669.	606869	Hexosaminidase A	Morocco, Algeria, Lebanon, Iraq
670.	606873	Hexosaminidase B	?
671.	606879	Phosphoglycerate Dehydrogenase	Morocco
672.	606883	Interleukin 1 Receptor-Associated Kinase 4	KSA
673.	606897	Lysosomal Trafficking Regulator	Kuwait
674.	606937	Cerebellar Ataxia with Mental Retardation, Optic Atrophy, and Skin Abnormalities	Lebanon
675.	606943	Usher Syndrome, Type IG	Tunisia, Palestine, Jordan
676.	606945	Low Density Lipoprotein Receptor	Algeria, Lebanon, Syria, Kuwait, Bahrain
677.	607009	Transient Receptor Potential Cation Channel, Subfamily M, Member 6	?
678.	607038	Otoancorin	Palestine, Lebanon, Jordan, Yemen
679.	607039	Deafness, Autosomal Recessive 22	Palestine
680.	607042	CLN3 Gene	Morocco
681.	607059	Solute Carrier Family 39 (Zinc Transporter), Member 4	Tunisia, Egypt
682.	607067	Saitohin	Algeria
683.	607084	Deafness, Autosomal Recessive 31	Palestine, Jordan
684.	607088	Spinal Muscular Atrophy, Chronic Distal, Autosomal Recessive	Lebanon
685.	607095	Anauxetic Dysplasia	Jordan
686.	607101	Deafness, Autosomal Recessive 30	Iraq
687.	607131	Macrocephaly with Multiple Epiphyseal Dysplasia and Distinctive Facies	Oman
688.	607139	Fanconi Anemia Complementation Group A Gene	Morocco, Tunisia
689.	607155	Muscular Dystrophy, Limb-Girdle, Type 2I	Tunisia
690.	607198	Tyrosyl-DNA Phosphodiesterase 1	KSA
691.	607239	Deafness, Autosomal Recessive 33	Jordan
692.	607250	Spinocerebellar Ataxia, Autosomal Recessive, with Axonal Neuropathy	KSA
693.	607306	Steroid 5-Alpha-Reductase 2	Lebanon
694.	607358	Autoimmune Regulator	Egypt
695.	607473	Vitamin K-Dependent Clotting Factors, Combined Deficiency of, 2	Lebanon
696.	607483	Basal Ganglia Disease, Biotin-Responsive	Syria, Yemen
697.	607564	Tripartite Motif-Containing Protein 6	Egypt
698.	607574	Arylsulfatase A	Lebanon, KSA
699.	607584	Spastic Paraplegia 24, Autosomal Recessive	KSA
700.	607585	Ataxia-Telangiectasia Mutated Gene	Morocco, Tunisia, Palestine
701.	607590	BBS7 Gene	KSA
702.	607608	Sphingomyelin Phosphodiesterase 1, Acid Lysosomal	Morocco, Algeria, Tunisia, KSA
703.	607624	Griscelli Syndrome, Type 2	?
704.	607625	Niemann-Pick Disease, Type C2	Algeria

	OMIM	*Name*	*Country*
705.	607626	Ichthyosis, Leukocyte Vacuoles, Alopecia, and Sclerosing Cholangitis	Morocco
706.	607690	Sar1a, S. Cerevisiae, Homolog 2	Algeria
707.	607694	Leukodystrophy With Oligodontia	Syria
708.	607696	USH1G Gene	Tunisia, Jordan
709.	607697	Set-Binding Factor 2	Morocco, Tunisia
710.	607731	Charcot-Marie-Tooth Disease, Axonal, Type 2H	Tunisia
711.	607739	Charcot-Marie-Tooth Disease, Type 4B2, With Early-Onset Glaucoma	Morocco, Tunisia
712.	607759	Integrin, Alpha-2B	Iraq
713.	607764	3-@Beta-Hydroxy-Delta-5-C27-Steroid Oxidoreductase	KSA
714.	607765	Cholestasis, Progressive Familial Intrahepatic 4	KSA
715.	607800	ATP-Binding Cassette, Subfamily A, Member 12	Morocco, Algeria
716.	607812	Craniolenticulosutural Dysplasia	KSA
717.	607817	COH1 Gene	Lebanon, Oman
718.	607831	Charcot-Marie-Tooth Disease, Axonal, Type 2K	Morocco
719.	607868	Tripartite Motif-Containing Protein 11	Egypt
720.	607900	Kindlin 1	Oman
721.	607928	Whirlin	Jordan
722.	608027	Cerebellar Atrophy with Progressive Microcephaly	Oman
723.	608034	Aspartoacylase	?
724.	608041	Anthrax Toxin Receptor 2	Morocco
725.	608091	Cerebellooculorenal Syndrome 2	UAE
726.	608097	Heterotopia, Periventricular, Autosomal Recessive	Yemen
727.	608107	Familial Mediterranean Fever Gene	Morocco, Lebanon, Jordan, Iraq
728.	608115	Ovarian Hyperstimulation Syndrome	Morocco
729.	608132	Tetratricopeptide Repeat Domain 8	KSA
730.	608154	Lipodystrophy, Generalized, with Mental Retardation, Deafness, Short Stature, and Slender Bones	Oman
731.	608156	Nablus Mask-Like Facial Syndrome	Palestine
732.	608207	Kala-Azar, Susceptibility to	Sudan
733.	608222	Adenylosuccinate Lyase	Morocco
734.	608358	Myopathy, Myosin Storage	KSA
735.	608367	Myopia 4	Algeria
736.	608395	Karak Syndrome	Jordan
737.	608443	Mental Retardation, Nonsyndromic, Autosomal Recessive, 3	?
738.	608465	Senataxin	Algeria
739.	608487	Tripartite Motif-Containing Protein 5	Egypt
740.	608509	Alopecia Universalis Congenita, XY Gonadal Dysgenesis, and Laryngomalacia	Jordan
741.	608515	Neutrophil Cytosolic Factor 2	Palestine, Jordan
742.	608547	Vitamin K Epoxide Reductase Complex, Subunit 1	Lebanon
743.	608585	Brachial Palsy, Familial Congenital	Egypt

	OMIM	*Name*	*Country*
744.	608630	Roundabout, Drosophila, Homolog of, 3	?
745.	608637	Spondyloepiphyseal Dysplasia, Omani Type	Oman
746.	608653	Deafness, Autosomal Recessive 32	Tunisia
747.	608681	Spondylocostal Dysostosis, Autosomal Recessive 2	Lebanon
748.	608728	Spondyloepimetaphyseal Dysplasia, Matrilin-3 Related	?
749.	608801	Glutaryl-CoA Dehydrogenase	?
750.	608845	ADP-Ribosylation Factor-Like 6	?
751.	608911	Choanal Atresia, Posterior	Yemen
752.	608931	Myasthenic Syndrome, Congenital, Associated with Acetylcholine Receptor Deficiency	Iraq

To investigate the reason for the peculiar geographic distribution of genetic disorders in our data, we conducted comparative analyses using different country indicators such as: population size, median age of population, birth rate, death rate, infant mortality rate, life expectancy at birth, and total fertility rate in corresponding Arab countries *(Fig. 6b-h).* No clear correlation could be concluded from such an analysis. However, a careful investigation of the references citing genetic disorders in Arab populations emphasizes:

- The role of French researchers in diagnosing disorders in Tunisian, Moroccan, and Algerian patients living in France (Tadmouri and Bissar-Tadmouri, 1999).
- The established culture of extended collaborations between scientific groups in the Maghreb countries and Central Europe (Tadmouri, 2004).
- The advanced medical care in Lebanon and Saudi Arabia as well as the relative enthusiasm of physicians in those countries towards publishing their findings whether alone or in collaboration with European or American research groups (Tadmouri and Tadmouri, 2002; Tadmouri and Bissar-Tadmouri, 2003; Tadmouri and Bissar-Tadmouri, 2004; Tadmouri, 2004).
- The role of researchers in the Occupied Territories in diagnosing many genetic disorders in Jewish communities that emigrated mainly from Morocco, Iraq, Yemen, and Libya. Those researchers also contributed to the description of many disorders in non-Jewish communities in Palestine (Zlotogora, 2004).

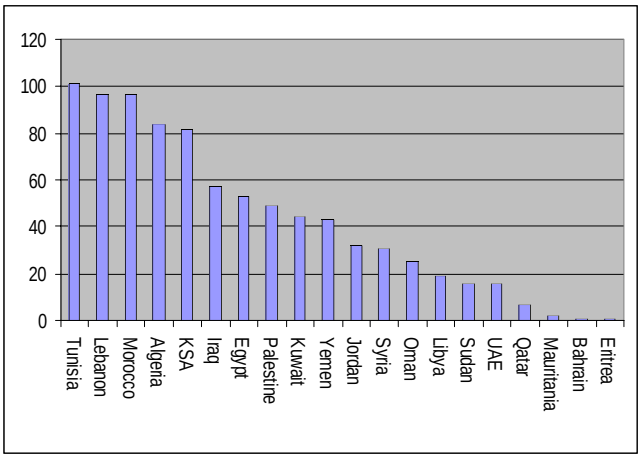
On the other hand, references citing Mendelian disorders in Arabs can generally be classified into two main groups:

1. Papers that characterize the clinical manifestations of an inherited disease: A paper in this category is usually authored by a small number of local scientists and mostly contains detailed information on the medical presentation of a disease case with appropriate literature review.
2. Papers that depict the molecular pathology of a genetic disease: In this case, a paper is usually the result of an extended collaboration among scholars from local Arab institutes and others from international laboratories. The content of the paper usually emphasizes the concepts of gene discovery, mutation characterization, and population genetics through the analysis of multiple subjects of different geographical origins.

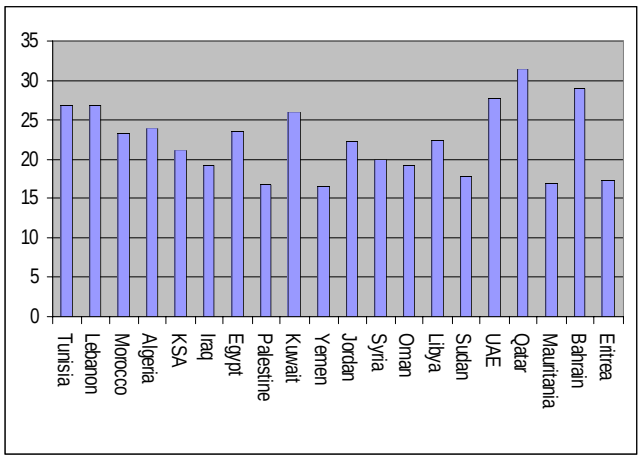
Approximately 35% of genetic diseases in Arabs do not have a defined molecular etiology. This is a clear indication that exhaustive research is required in the area to depict the molecular pathologies causing diseases in Arab populations. On the other hand, the chromosomal distribution of known causative disease genes in Arabs does not show any bias. A comparison of our data with the total gene loci cited in OMIM® does not reveal any abnormal pattern *(Fig. 7)*.

A detailed analysis of the molecular basis of defined genetic diseases indicates that the majority of genetic diseases in Arabs result from single-gene alterations whereas complex genetic diseases account only for less than 1% of genetic abnormalities in Arabs (*Fig. 8).* Hence, if the proper infrastructure is available, diagnostic services for many of these genetic disorders may be offered to people at risk.

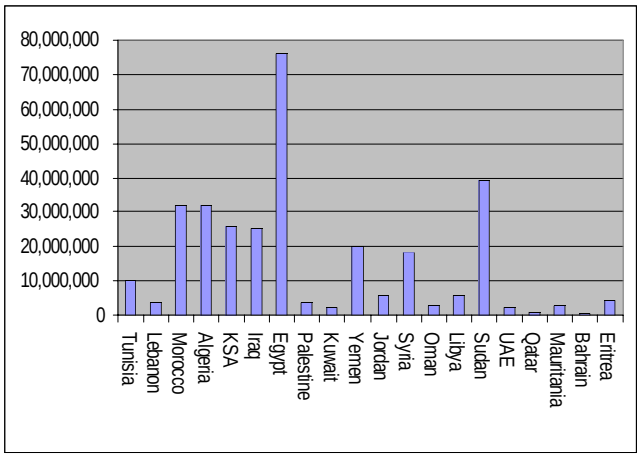
A striking observation, however, is the presence of an overwhelming number of recessively inherited genet-



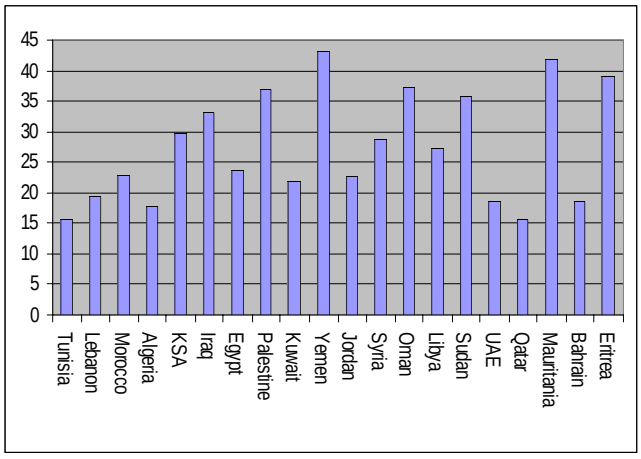
a.: Genetic disorders in Arabs.



c.: Median age of population (years).



b.: Population size.



d.: Birth rate (births/1000 population).

ic traits in Arab patients *(Fig. 9).* This is in agreement with many other reports on the subject (Teebi, 1994; Al-Gazali *et al.*, 1995). High rates of consanguinity among Arabs as well as the structure of Arab families could be major factors to explain this observation.

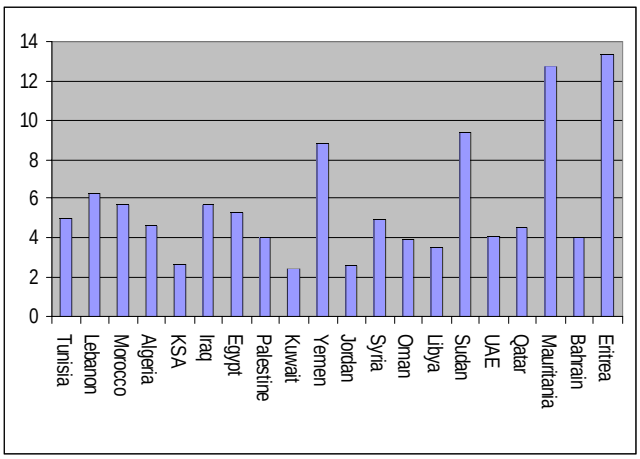
The Spectrum of Genetic Disorders in Arabs

In the last few decades, a relatively large number of new syndromes and variants were delineated in Arab people. Incidentally, Arab scholars were first to describe a number of genetic disorders, such as: The Teebi-Shaltout syndrome (Teebi and Shaltout, 1989) and Megarbane syndrome (Megarbane *et al.*, 2001).

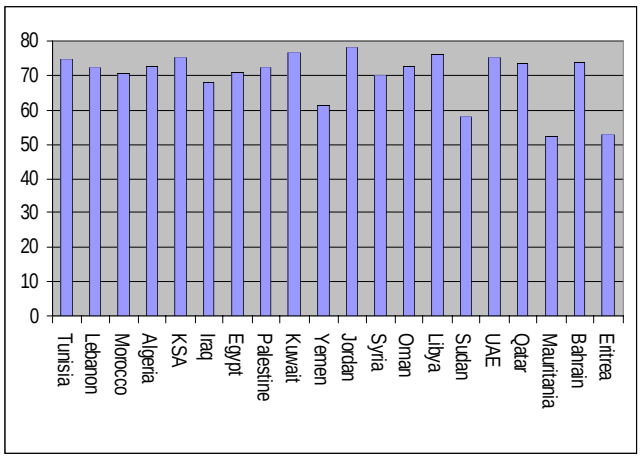
Some of the 752 genetic disorders in Arabs are Arab-specific syndromes, such as: The Algerian type of spondylometaphyseal dysplasia (Kozlowski *et al.*, 1988), the Kuwaiti type of the cardioskeletal syndrome (Reardon *et al.*, 1990), the Nablus mask-like facial syndrome (Teebi, 2000), and the Omani type of spondyloepiphyseal dysplasia (Rajab *et al.*, 2004). However, many other disorders do exist in other parts of the world, thus, negating the concept of the private syndrome.

On the other hand, many genetic diseases exhibit specific geographical distributions. Examples include:

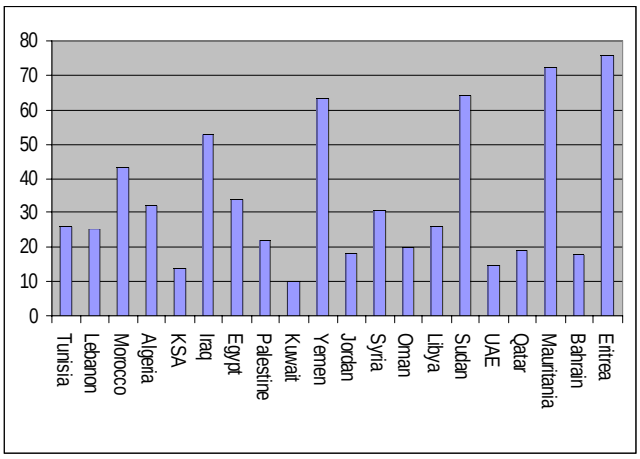
1. The Maghreb Region: Type II bare lymphocyte syndrome and mutations in the erythrocytic 1 alpha-spectrin gene *(Fig. 10a,b).*
2. North Africa: Creutzfeldt-Jakob disease and limb-girdle muscular dystrophy type 2c *(Fig. 10c,d).*
3. The Middle East: Krabbe disease and dyssegmental dwarfism *(Fig. 10e,f).*



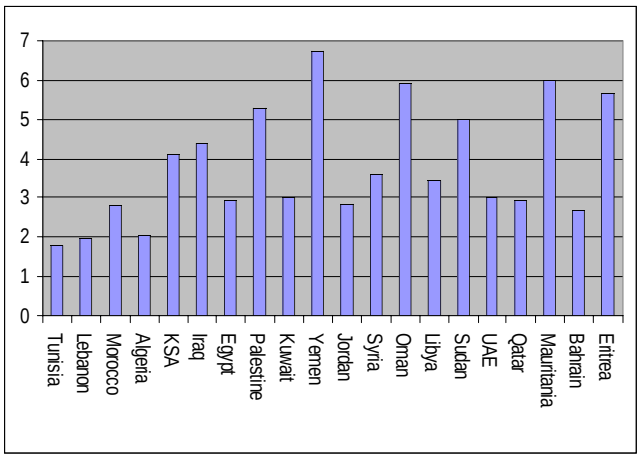
e.: Death rate (deaths/1000 population).



g.: Life expectancy at birth (years).



f.: Infant mortality rate (deaths/1000 live births).



h.: Total fertility rate (children born/woman).

Figure 6.: Genetic disorders in Arabs (a) and major health indicators for Arab countries (b-h).

4. The Arabian Gulf: Hypoparathyroidism-retardation-dysmorphism syndrome and the lethal type of popliteal pterygium syndrome *(Fig. 10g,h).*

However, the sporadic distribution of many genetic diseases in geographically separated Arab populations hint for a possible wider presence in Arabs. For this reason, more research is needed on these disorders to complete the picture. Candidate disorders for future research may include: Gγ and Aγ globin gene mutations, cystic fibrosis, low-density lipoprotein receptor gene mutations, and α-thalassemia *(Fig. 10i-l).*

On the other hand, extensive published research available for certain genetic disorders reflects their wide presence in Arab populations. Examples of such disorders include: Familial Mediterranean fever, β-thalassemia, and glucose-6-phosphate dehydrogenase deficiency *(Fig. 10m-o).* The overwhelming distribution of these diseases in Arabs is best explained by the exposure of Arab countries to common environmental factors that helped the natural selection for these disorders such as malaria in the case of hemoglobinopathies and dietary traditions in the case of glucose-6-phosphate dehydrogenase deficiency.

Recommendations

Data from OMIM® clearly indicate that genetic disorders are relatively numerous in Arab people. However, results on the occurrence of genetic disorders as mined from OMIM® should only be considered as preliminary and only represent a fraction of the

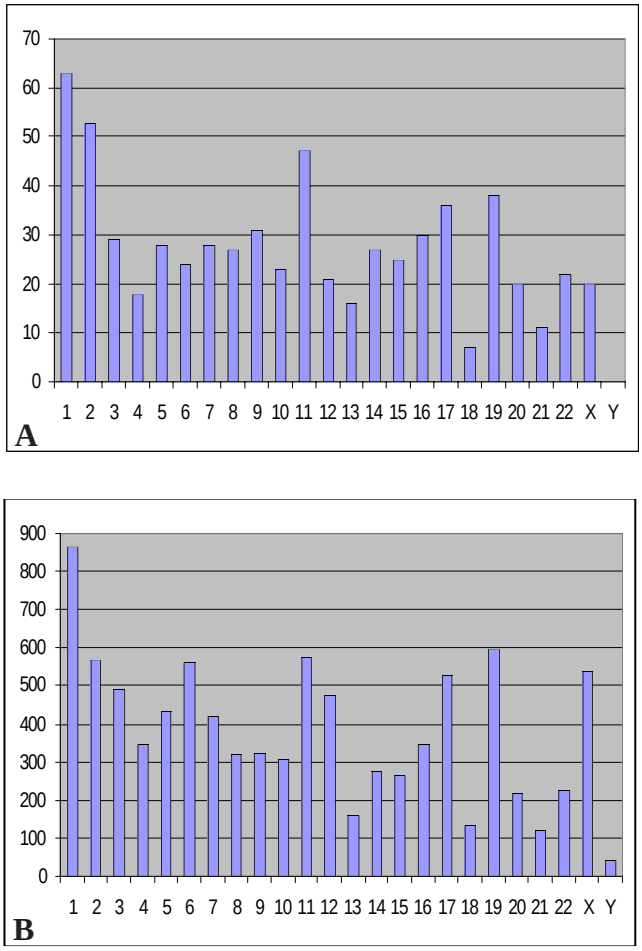


Fig. 7: Chromosomal distributions of gene abnormalities in Arab populations (a) and the chromosomal distribution of total gene loci in OMIM (b).

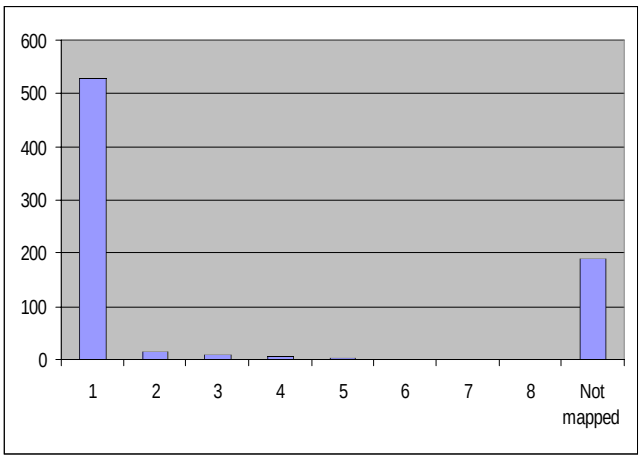


Fig. 8: Distribution of genetic disorders in Arabs according to the number of causative gene loci.

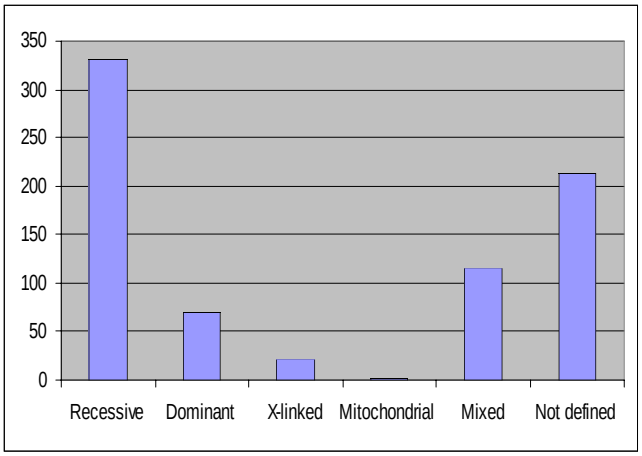
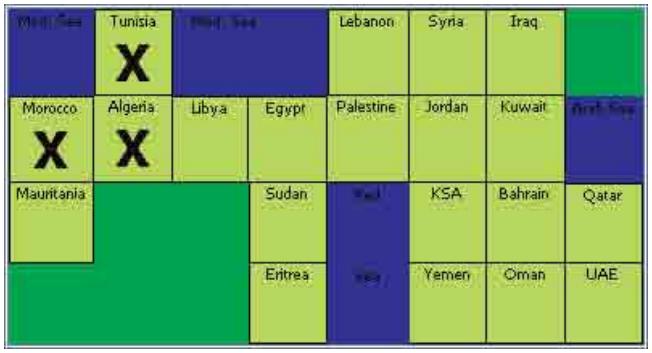


Fig. 9: Mode of inheritance of genetic disorders in Arabs.

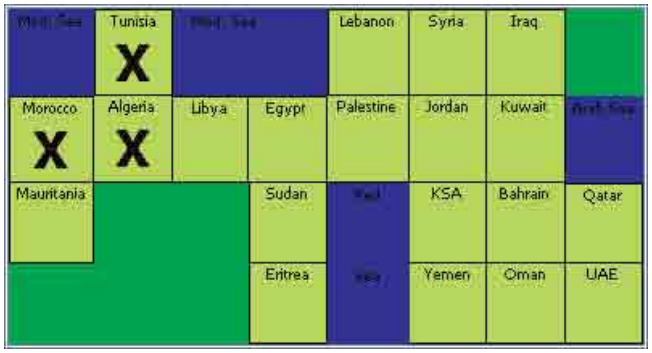
actual figure. According to results from OMIM®, the numbers of genetic disorders described in Maghreb countries and Lebanon, with a total population of approximately 78 million, far exceed those described in Egyptians, with similar population size. In addition, we accounted for more than 121 genetic disease entities in the Lebanese people (Tadmouri and Bissar-Tadmouri, 1997) while OMIM® describes 97 disorders only. Furthermore, OMIM® indicates the presence of 49 genetic diseases in the non-Jewish population of Palestine while a review of the published literature revealed the presence of 179 genetic abnormalities (Zlotogora, 2004).

OMIM® editors continuously monitor published results in internationally peer-reviewed publications to update the content of the database. Since many Arab physicians tend to publish their results in journals not recognized by OMIM® editors, OMIM® results inadvertently give the impression that most of the diagnosis of genetic disorders in Arabs were carried out by non-Arab researchers. Accordingly, investigating the details of many genetic diseases in Arab populations should extend to incorporate other sources of information. These include:

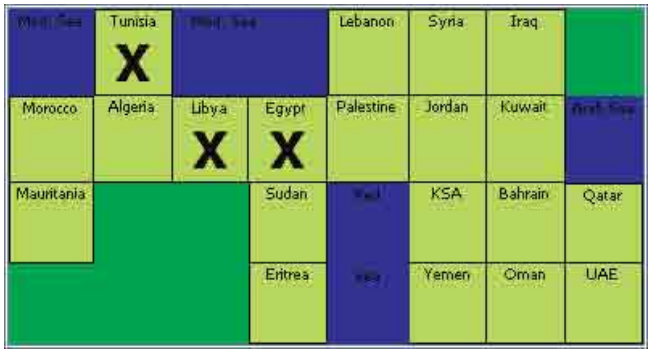
1. Careful monitoring, editing, and archiving of international peer-reviewed publications. For example, a dictionary of highly sensitive search strategies may be used to search for articles cited in major biomedical bibliography indices such as PubMed.
2. Extensive assessment of national peer-reviewed



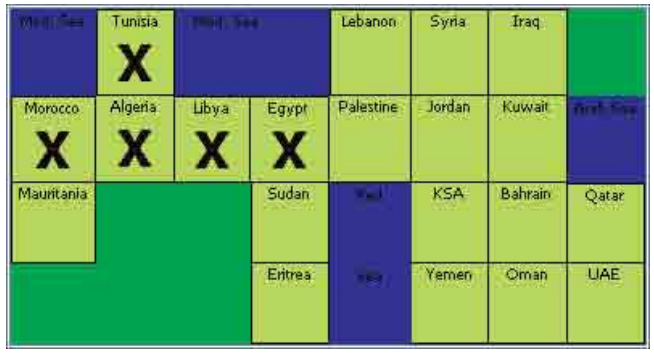
a.: Type II bare lymphocyte syndrome (OMIM: 209920)



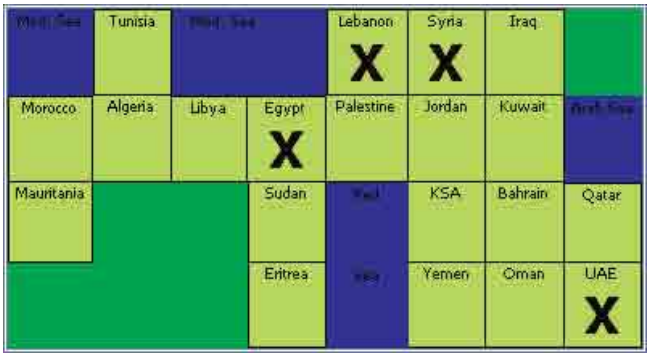
b.: Erythrocytic 1 alpha-spectrin (OMIM: 182860)



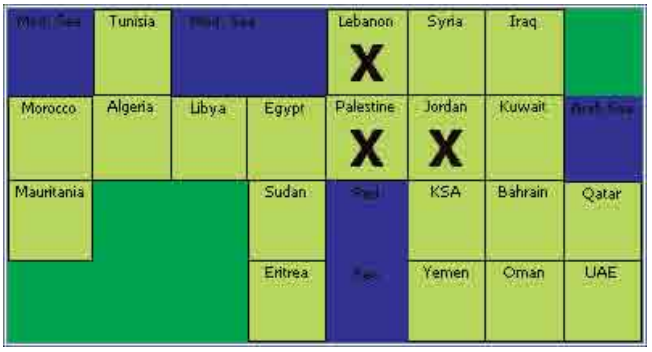
c.: Creutzfeldt-Jakob disease (OMIM: 123400)



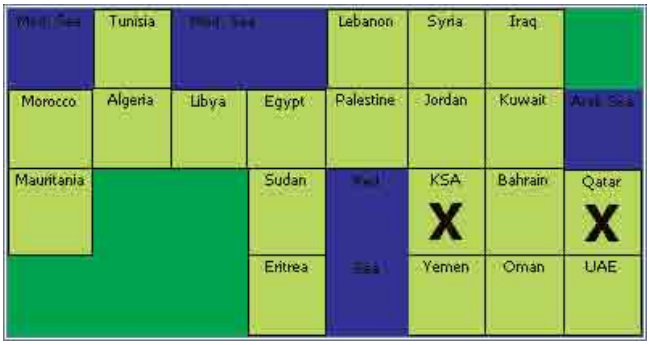
d.: Limb-girdle muscular dystrophy type 2c (OMIM: 253700)



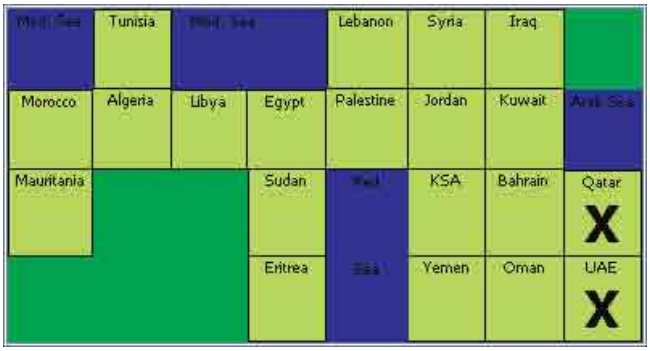
e.: Krabbe disease (OMIM: 245200)



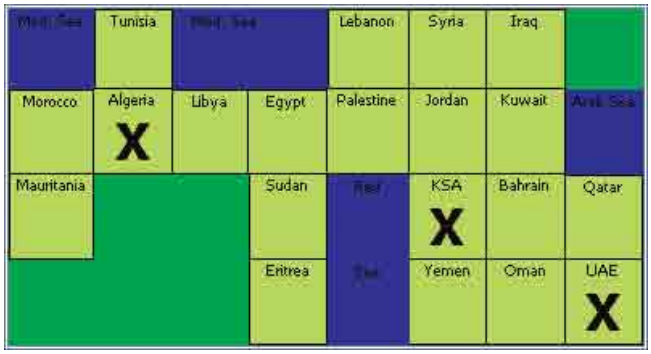
f.: Dyssegmental dwarfism (OMIM: 224400)



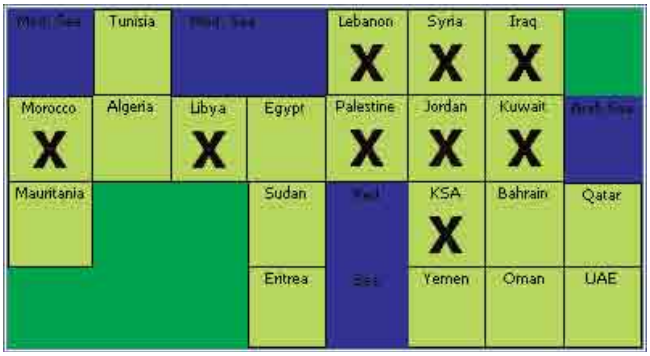
g.: Hypoparathyroidism-retardation-dysmorphism syndrome (OMIM: 241410)



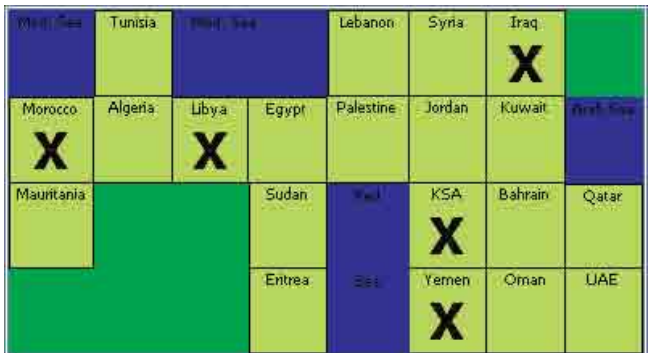
h.: The lethal type of popliteal pterygium syndrome (OMIM: 263650)



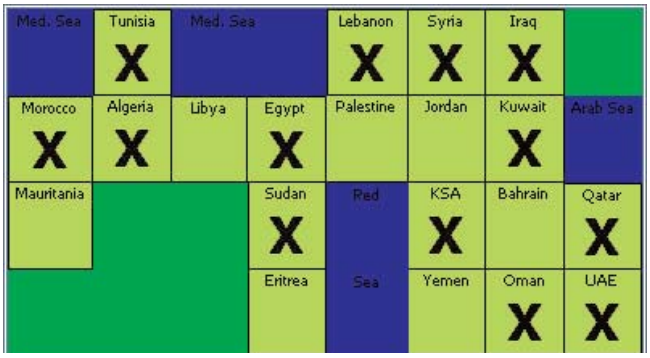
i.: Gamma G Hemoglobin (OMIM: 142250)



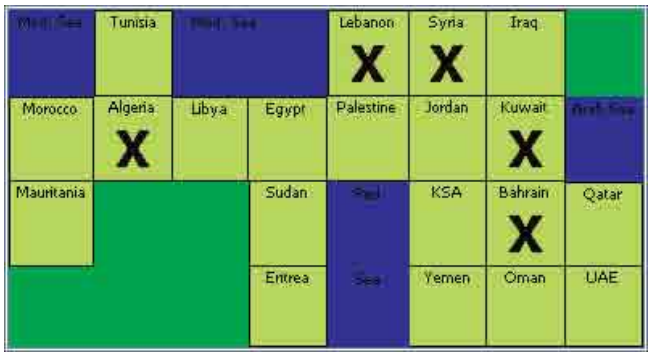
m.: Familial Mediterranean fever (OMIM: 249100)



j.: Cystic fibrosis (OMIM: 219700)

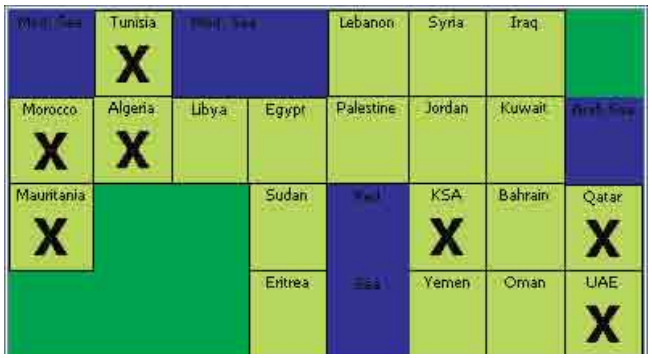


n.: Beta-globin gene locus (OMIM: 141900)



k.: Low density lipoprotein receptor (OMIM: 606945)

o.: Glucose-6-phosphate dehydrogenase deficiency (OMIM: 305900)



l.: Alpha 1 Globin (OMIM: 141800)

Fig. 10: Computer generated synthetic maps indicating the relative geographic distribution of some genetic disorders in the Arab World.

communications. The search for such information may start from local indices such as the Saudi MedBase, which is a digital index for nine of the most prominent medical journals published in Saudi Arabia between years 1979 and 1999. In addition, the awaited "Index Medicus of the Eastern Mediterranean Region" may also be an invaluable source of local information since it

ultimately aims at the public release of the full text of more than 400 national biomedical journals published in the Eastern Mediterranean region.

3. Whenever feasible, the access of hospital records could offer important insight on many genetic abnormalities that were not previously published or their incidence is unknown in certain localities.
4. Concurrently, personal contacts with local practitioners and the contribution of scholars in the region would be an important factor for the continued maintenance and development of the repository; thus, allowing our knowledge to grow as a result of a community effort.