

# Genetic disorders in Arabs as for OMIM™

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## ABSTRACT

Arab populations are genetically a heterogeneous group that resulted from the admixture between different populations throughout history. A search for genetic disorders described in Arab populations in the online version of McKusick's "Mendelian Inheritance in Man: Catalogue of Human Genes and Genetic Disorders" (OMIM™) resulted in 374 entries. The data presented in this review emphasizes the role of non-Arab researchers in diagnosing genetic diseases in Arab populations. This observation could be due to the fact that reports presented by Arab researchers are usually published in journals not recognized by OMIM™. Thus, the number of genetic disorders in Arabs is expected to be more than 374. Reviews that covered the subject went rapidly out of date as new disorders were continuously described in Arabs. Establishing a regularly updated repository for genetic disorders diagnosed in Arabs is a necessity to unify, as much as possible, what has been carried out on the subject.

**Keywords:** Arab populations, genetic disorders, congenital malformations, Online Mendelian Inheritance in Man.

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Arab populations consist of 270 million individuals (estimates of 1996)<sup>1</sup> scattered over a vast geographical region that extends from Iraq in the East to Morocco in the West. They occupy the whole of Mesopotamia, Middle East, Arabian Gulf, North Africa, as well as some parts of East and West Africa. Arabs are not homogeneously distributed over this geographical area and tend to highly concentrate in narrow locations. Of these are: the Nile, Euphrates, and Tigris valleys, the coastal regions of North Africa, the Gulf, and Western Asia. Arab populations are distributed in 23 different countries, namely: Algeria, Bahrain, Comoros, Djibouti, Egypt, Iraq, Jordan, Saudi Arabia, Kuwait, Lebanon, Libya, Mauritania, Morocco, Oman, Palestine, Qatar, Somalia, Sudan, Syria, Tunisia, United Arab Emirates, and Yemen.

Genetically, Arabs are a large and heterogeneous group that resulted from the admixture with many other populations throughout the history. In general, Arab populations are characterized by the following: (1) presence of isolates (e.g., Armenians, Bedouins, Druzes, Jews, Kurds, Nubians, and others), (2) high

rates of inbreeding or consanguineous marriage, which is usually a common traditional practice followed within the same tribe, village, or social unit;<sup>2</sup> (3) marriage at young age (sometimes even less than 15 years of age);<sup>2</sup> (4) large family size (5-9 members);<sup>1</sup> (5) high fertility rates (3-7 children born/woman);<sup>1</sup> (6) high population growth rate (1.8-6.7%);<sup>1</sup> (7) high birth rate (20-47 births/1000 people);<sup>1</sup> (8) high infant mortality rate (11-121 deaths/1000 live births);<sup>1</sup> (9) child bearing in older maternal age;<sup>2</sup> (10) and general lack of public health measures directed at the control and prevention of genetic disorders.<sup>2</sup>

In the year 1973, the first edition of McKusick's "Mendelian Inheritance in Man: Catalogue of Human Genes and Genetic Disorders" was published. This catalogue sites for characterized genetic disorders and lists relevant references in which there is information about the genotypes and phenotypes of each disorder. This catalogue is continuously updated both in its hard printed form (1973-1994)<sup>2</sup> or its online version (1987-present), known as Online Mendelian Inheritance in Man (OMIM™; <http://>

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**Table 1** - Some of the population characteristics in Arab countries<sup>1</sup> arranged in decreasing order of frequency of genetic disorders described by OMIM™

Country	Genetic disorders described	Population size	Birth rate (births/1,000 population)	Infant mortality rate (deaths/1,000 live births)	Consanguinity (% of total marriage)
Tunisia	51	9,019,687	24.03	35.1	49.5
Saudi Arabia	50	19,409,058	38.32	46.4	54-58 <sup>6,7</sup>
Lebanon	45	3,776,317	27.93	36.7	26 <sup>8</sup>
Morocco	44	29,779,156	27.39	43.2	29-33 <sup>9,10</sup>
Algeria	34	29,183,032	28.51	48.7	23-40 <sup>11,12</sup>
Kuwait	34	1,950,047	20.28	11.1	38-54 <sup>13,14</sup>
Iraq	33	21,422,292	43.07	60.0	58 <sup>15</sup>
Palestine	27	2,351,681	44.72	28.0	39 <sup>16</sup>
Egypt	21	63,575,107	28.18	72.8	23-41 <sup>17,18</sup>
Yemen	21	13,483,178	45.22	71.5	?
Syria	15	15,608,648	39.56	40.0	33 <sup>19</sup>
Libya	13	5,445,436	44.42	59.5	?
Jordan	12	4,212,152	36.67	31.5	50 <sup>20</sup>
Sudan	8	31,547,543	41.08	76.0	?
Qatar	4	547,761	21.03	19.6	?
UAE	3	3,057,337	26.43	20.4	62 <sup>21</sup>
Mauritania	2	2,336,048	46.92	81.7	?
Oman	2	2,186,548	37.86	27.3	?
Bahrain	1	590,042	23.58	17.1	?
Somalia	1	9,639,151	44.17	121.1	?
Comoros	-	569,237	46.00	75.0	?
Djibouti	-	427,642	43.00	107.0	?

www.ncbi.nlm.nih.gov/Omim). The OMIM™ database was first supported by the Howard Hughes Medical Institute from 1987 to 1991. Since 1992, OMIM™ is supported jointly by the National Institute of Health (NIH) and the Department Of Energy (DOE) as part of the international Genome Data Base (GDB) project. The updating of OMIM™ is very frequent sometimes being once each week for files that deal with "hot topic" disorders. As a matter of fact, the OMIM™ database included 738 entries in 1993.<sup>4</sup> On September 15, 1997 the database included 8675 entries. This rapid and constant growth of OMIM™ as well as its "user friendly" interface, allowed this database to be the first choice for researchers to collect information concerning any genetic disorder in question.

**Methods of search.** A search for entries citing genetic disorders in Arab countries/populations resulted in a cumulative list of 374 different genetic disorders (Supplement I) and their references (Supplement II). The number of disorders described in each Arab country is shown in Table 1.

**Outcome.** It is usually expected that countries with the largest population size tend to contract the highest number of genetic disorders. However, Table 1 contradicts this idea, for we see Lebanon, for example, whose population is 15 times less than that of Egypt, showing 45 different genetic disorders whereas Egypt shows only 21. This fact proves that some other factors are playing role in the determination of the number of genetic disorders in each country. Of these factors we can list: (1) The role of French researchers in diagnosing the disorders described in Tunisian, Moroccan, and Algerian

patients, most of whom live in France (Supplements I and II). (2) The advanced medical care in Saudi Arabia, Lebanon, and Kuwait as well as the relative enthusiastic feeling of physicians in those countries towards publishing their findings. (3). The active role played by researchers in the Occupied Territories in diagnosing hundreds of cases of genetic disorders in Jewish communities that emigrated from Morocco, Iraq, Yemen, Libya, and others. Those researchers also contributed to the majority of cases diagnosed in Palestinians (Supplements I and II). (4) It seems that Arab physicians tend to publish their findings in journals which are not recognized by OMIM™. Thus, OMIM™ sources, mistakenly, reveal that most of the diagnosis of genetic disorders in Arabs was carried out by non-Arabs (Supplement II).

Despite the inaccuracy of the OMIM™ list, few points can be deduced: 1. Genetic disorders are relatively numerous in the Arab population (Supplement I). Although the number of these disorders is around 374 according to OMIM™, we expect that the real figure is much higher than that. One reason for this belief is our knowledge concerning genetic disorders described in Lebanon and that they add up to 121 entries<sup>22,23</sup> and not only 45 as demonstrated in Supplement I. (2) Some of the 374 disorders described, such as Familial Mediterranean Fever, Glucose-6-Phosphate Dehydrogenase Deficiency (G-6-PDD), and the Hemoglobinopathies, tend to overwhelm many Arab countries at a time. This could be explained by the exposure of these countries to common environmental factors that helped the selection for these disorders such as malaria vs. the

hemoglobinopathies/G-6-PDD, dietary traditions vs. G-6-PDD, and so on. (3) There is a relatively high number of new or Arab-specific syndromes and variants (Supplement I).

**Recommendations.** There are few comprehensive reviews that covered the subject,<sup>2,24,25</sup> and these go rapidly out of date as new disorders are continuously discovered or described in Arabs. Establishing a repository for all what was written about genetic disorders in Arabs becomes a necessity to get a unified list for genetic disorders in the Arab world. It is our intention, therefore, to publish a regular update starting with the information presented herein (Supplements I and II). Readers and researchers are encouraged to contribute to the repository by sending their comments and criticism as well as any information on the genetic disorders in Arabs provided with a complete reference.

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