The Egyptian Journal of Medical Human Genetics (2012) **13**, 45–62

Ain Shams University

The Egyptian Journal of Medical Human Genetics

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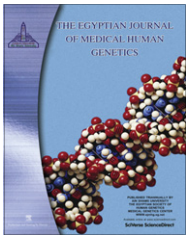

ORIGINAL ARTICLE

Profile of genetic disorders prevalent in northeast region of Cairo, Egypt

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Received 29 May 2011; accepted 20 October 2011
Available online 4 February 2012

KEYWORDS

Genetic disorders;
Congenital malformations;
Inbreeding profile;
Northeast region;
Cairo;
Egypt

Abstract As clinical geneticists, we recently reviewed our 43 years experience in an attempt to represent the frequency of genetic disorders in the Division of Genetics at Pediatric Hospital, Faculty of Medicine, Ain-Shams University, Cairo, Egypt, during the period from 1966 to 2009.

All patients (from birth up to 18 years) suspected of having a genetic disorder were referred to the Genetics Clinic in the same hospital. 28,689 Patients were proved to have genetic disorders after full investigations among 660,280 children attending the Pediatrics Hospital which constituted 4.35% or 43.5/1000. Neurologic disorders were the most common (31.38%) followed by hematologic disorders (18.48%), chromosomal abnormalities (11.51%), fetal, neonatal and infant deaths (6.56%), special senses (5.82%), inborn errors of metabolism (4.24%), endocrine disorders (3.87%), skeletal disorders (3.17%), genito-gonadal anomalies (3.10%), neuromuscular disorders (2.86%), syndromes (2.08%), genodermatoses (1.92%), cardiac disorders (1.47%), gastrointestinal tract anomalies (1.37%), renal anomalies (0.26%), connective tissue disorders (0.26%), respiratory defects (0.22%), vascular anomalies (0.21%), and immunologic disorders were the least common (0.19%).

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Peer review under responsibility of Ain Shams University.
doi:10.1016/j.ejmhg.2011.10.002

Our study showed a high prevalence of genetic diseases among Egyptians which are nearly the same in the other studies in Egypt and are rapidly becoming a major public health concern. Establishment of national or hospital based registers for genetic disorders is very important to know the magnitude of the problem so that the national program for the prevention of genetic disorders can be implemented.

1. Introduction

Hereditary diseases and congenital malformations have been reported to affect 2–5% of all livebirths [1]. The British Columbia Health Surveillance Registry shows that before the age of 25, at least 53 out of 1000 liveborn individuals can be expected to have a disease with an important genetic component. This comprises 3.6 single gene disorders per 1000, 1.8 chromosomal disorders per 1000, 46.4 multifactorial or part-genetic disorders per 1000 and 1.2 genetic type of uncertain disorders per 1000. In this analysis, congenital anomalies were included only if a multifactorial or part-genetic etiology had been established, but if all congenital abnormalities were included the cumulative figure rose to more than 79 per 1000 livebirths [2]. Available evidence suggests that congenital and genetic disorders are responsible for a major proportion of infant mortality, morbidity, and handicap in Arab countries [3].

Egypt was classified as one of the more developed Arab countries according to categorization of the Committee of Development Policy (CDP) [4]. Egypt, one of the main civilisations of the ancient world, has a history that goes back more than 5000 years. It enjoys a distinguished geographical location at the juncture of the ancient world continents of Africa, Asia and Europe. It has always been a place of inter-civilization and reactivation between the East and West as well as the North and South. It was also the crossing road of the heavenly religious of the World. It is about 1 million (m) km^2 and is located in the north-eastern corner of Africa and Southwestern Asia [5].

Egypt is divided into three main geographical regions: the Nile Vally, the Eastern desert and the Western desert. The Nile Vally represents 4% from the area of Egypt and is divided into Upper Egypt region, Lower Egypt region, Suez Canal region and Northern coast lakes region. It is also divided into 27 governorates.

Cairo, the most populous Arab country, is the glorious capital of Egypt. Cairo's population rose to more than 18 millions (the highest population density in Egypt). Egyptians are mainly descended from ancient Egyptian Society (94%). Ethnic minorities in Egypt include, Nubians, Berbers, Bedouin Arabs, Beja and Dome (4%) and others (2%) [5].

1.1. Demographic features of Egypt

Egyptian population according to census 2006 hit 76.5 million, around 3.9 million are living abroad. One Egyptian baby is born every 23 s, birth rate 22.94 births/1000.

In Egypt, birth rates among women over 35 years have been almost twice (65/1000) as often as those occurring among women of the same age in USA (33.7/1000) [6]. Old maternal age is associated with aberrant genetic recombination or similar genetic mechanisms together with one or more environmental factors [7]. Moreover, risks to the fetus could have been modulated by the effects of aging on the mother's general or reproductive health [8]. Other investigators have suggested that physiologic changes associated with aging and or environmental factors may increase birth defects [9,10]. In Egypt advanced maternal age has significant role in causation of repeated abortions [11].

1.2. Consanguinity among Egyptians

While ancient Egyptians of the reigning dynasty (1580–1350 BC) favored marriages of brother and sisters among the royal family, current laws of the Egyptian society emphasize the value of outbreeding and prohibit marriages between relatives closer than first cousins.

Consanguineous marriage is still high in Egypt (35.3%) especially among first cousins (86%). However, the frequency varies by region. It is higher in Sohag (42.2%) and Cairo (36.1%) than in Assuit (21.7%). Also it was higher in rural areas (59.9%) than in semiurban and urban areas (23.5%) and (17.7%), respectively [12]. This increase in consanguinity rate is due to the fact that many families prefer marriage among first cousins to preserve family structure, links and provide social, economics and cultural benefits. Many Egyptians believe that there may be more compatibility and less tendency to divorce between husband and wife from a family. This favored the appearance of complex phenotypes of genetic disorders which result in difficulties in phenotype classification.

1.3. Effect of inbreeding on the genetic morbidity

This high consanguinity rate is reflected on higher risk of infant and child mortality in Egypt. There was 30% and 19% higher risk of infant mortality among close and remote consanguineous couples, respectively. Similarly the risk of child mortality is found higher among close consanguineous couples by more than 50% and among remote consanguineous couples by 27% as compared to non-consanguineous marriage [12,13].

Consanguinity also increases birth prevalence of severe recessive disorders, appearance of new autosomal recessive syndromes, multiple genetic disorders in the same individual or same family and homozygosity for autosomal dominant disorders. Consanguinity also increases the risk of birth of a child with a malformation [14].

Several genetic disorders have been reported to be frequent among Egyptians [4,15]. This observation could be the result of an ascertainment bias resulting from a possible higher awareness of the hereditary disorders in Egypt.

2. Aim of the study

As clinical geneticists, we recently reviewed our 43 years experience in an attempt to represent the frequency of genetic