Epidemiology of neuro-genetic disorders in Northwestern Iran

Saeed Dastgiri, *MPH, PhD,* ***Mortaza J. Bonyadi,*** *MSc, PhD,* ***Tuba Mizani,*** *MD, MSc.*

Neuro-genetic disorders are responsible for a major proportion of mortalities, morbidities, and handicaps across the world varying by racial, ethnic, and cultural differences. These disorders are common among developed and developing countries. Genetic disorders and congenital abnormalities occur in around 2-5% of live births accounting for approximately 30% of pediatric hospital admissions in many places in the region.[1,2] The objective of this study was to estimate some epidemiological features of the neuro-genetic disorders in the northwest of Iran.

In this cross-sectional study, 1613 cases (855 males and 758 females, age range: 6 months-67 years) with confirmed neuro-genetic disorders or carriers of the traits, diagnosed by molecular methods were identified in the Departments of Molecular Genetics and Epidemiology of Tabriz University of Medical Sciences, Tabriz, Iran, between January 2005 and December 2009. The data also included place of birth, year of diagnosis, and demographic information for the subjects. The area population was estimated according to the official reports for 2005-2009 from the regional Statistics Bureau. Data were considered confidential and the Ethical Committee of the Tabriz University of Medical Sciences approved the study protocol. The prevalence rate was calculated by dividing the numerator (number of cases in each disorder) by the relevant denominator (total population in the study area) for the same period of time at the same place. For statistical analysis, we calculated 95% confidence intervals (CI) for each prevalence rate.

The study subjects included patients (n=889), carriers (n=614), suspected (n=109), and unknown cases (n=1). The 5-year frequency and prevalence of neuro-genetic disorders by the type of disease are presented in **Table 1**. The most prevalent (for 5 years) disorders were identified as inherited deafness, spinal-muscular atrophy, Duchenne muscular dystrophy, and Down syndrome. Charcot-Marie-Tooth syndrome, Turner syndrome, spinocerebellar ataxia, microcephaly, ataxia telangiectasia, Anderson-Fabry syndrome, gyrate atrophy, Maple syrup urine syndrome, Sanfilippo syndrome, and inherited myopathy accounted proportionally for less than one (per 100000 populations) in the region.

In this study, we investigated the frequency and prevalence of neuro-genetic disorders in the northwest of Iran. The most prevalent disorders were identified as inherited deafness, spinal-muscular atrophy, Duchenne muscular dystrophy, and Down syndrome. Similar studies from different parts of the world have reported various findings: the reports of Al-Gazali et al[1] from an Arab population suggest that hemoglobinopathies including G6PD and autosomal recessive syndromes are the most prevalent disorders in the region. Some other studies in the same region have also reported similar results.[2] Gonzalez-Ferrer et al[3] studied 12,000 families in Venezuela. They identified Huntington's disease, sickle cell anemia, neural tube defects, and Down syndrome as the most frequent genetic diseases in the country. The reports of Verma et al[4] from India also show that thalassemia, Duchenne muscular dystrophy, Fragile X syndrome, spinal-muscular atrophy, thrombophilia, and spinocerebellar ataxias are the most prevalent genetic disorders in different regions of India. In the United Kingdom, fatty acid metabolic

Table 1 - Five year prevalence of neuro-genetic disorders in Northwestern Iran, 2005-2009 (per 100,000 populations).

Disease	Prevalence	95% CI
Charcot-Marie-Tooth syndrome	0.028	(-0.0268-0.0829)
Turner syndrome	0.028	(-0.0268-0.0829)
Spinocerebellar ataxia	0.028	(-0.0268-0.0829)
Microcephaly	0.056	(-0.021-0.133)
Ataxia telangiectasia	0.056	(-0.021-0.133)
Anderson-Fabry syndrome	0.084	(-0.011-0.179)
Gyrate atrophy	0.084	(-0.011-0.179)
Maple syrup urine syndrome	0.11	(0.0022-0.22)
Sanfilippo syndrome	0.14	(0.017-0.26)
Inherited myopathy	0.14	(0.017-0.26)
Leukodystrophy	0.168	(0.033-0.3)
Huntington's disease	0.196	(0.05-0.34)
Tay Sachs disease	0.22	(0.07-0.37)
Inherited blindness	0.22	(0.07-0.37)
Galactosemia	0.25	(0.087-0.41)
Fragile X syndrome	0.28	(0.10-0.45)
Inherited autosomal neuropathy	0.28	(0.10-0.45)
Becker's syndrome	0.33	(0.14-0.52)
Cerebral cavernous malformations	0.42	(0.20-0.63)
Friedreich's ataxia	0.5	(0.27-0.73)
Wilson disease	2.88	(2.32-3.44)
Phenylketonuria	2.99	(2.42-3.56)
Down syndrome	5.09	(4.35-5.83)
Duchenne muscular dystrophy	7.84	(6.92-8.76)
Spinal-muscular atrophy	11.12	(10.01-12.21)
Inherited deafness	11.37	(10.26-12.47)

CI - confidence interval

Disclosure. Funding for this project was received from the Tabriz University of Medical Sciences. The authors declare no conflicting interests with any drug, marketing, or business company.

disorders, inherited deafness, muscular dystrophies, cystic fibrosis, phenylketonuria, Fragile X syndrome, and hemophilia were reported as the prevalent genetic disorders.[5] Although consanguineous marriage may have a key role for the occurrence of neuro-genetic disorders, the impact of racial differences, ethnicity and cultural differences (namely, giving birth at an older age) cannot be ruled out in explaining the differences between reports from various places in the world.

The lack of funding did not allow us to investigate the magnitude of neuro-genetic disorders in various parts of the county. Further studies are recommended to investigate the epidemiological picture of this problem in the Eastern Mediterranean region including Iran. Estimating the true prevalence of neuro-genetic disorders may help in conducting screening programs, and to set up a population-based monitoring system for neuro-genetic disorders. This will enable health policy makers to plan realistic health care programs for the control and prevention of genetic disorders in the population.

Received 14th October 2011. Accepted 13th February 2012.

From the National Public Health Management Centre (Dastgiri), School of Medicine, Tabriz University of Medical Sciences, the Center of Excellence for Biodiversity (Bonyadi), Children Health Research Centre, Faculty of Natural Sciences, University of Tabriz, Tabriz, Iran, and the German Cancer Research Center (Mizani), Heidelberg, Germany. Address correspondence and reprint requests to: Professor Saeed Dastgiri, Department of Community and Family Medicine, School of Medicine, Tabriz University of Medical Sciences, Tabriz 515665931, Iran. Tel. +98 (914) 4157039. Fax. +98 (411) 3364668. E-mail: saeed.dastgiri@gmail.com

References

1. Al-Gazali L, Hamamy H, Al-Arrayad S. Genetic disorders in the Arab world. ***BMJ*** 2006; 333: 831-834.
2. Tadmouri G. Genetic Disorders in Arab Populations. UAE: Centre for Arab Genomic Studies Publications; 2008. p. 1-43.
3. González-Ferrer S, Pineda-Bernal L, Delgado-Luengo W, Villalobos-Cabrera H. Medical genetics in Zulia, a State of Venezuela. ***Community Genet*** 2004; 7: 153-156.
4. Verma IC, Saxena R, Lall M, Bijarnia S, Sharma R. Genetic counseling and prenatal diagnosis in India--experience at Sir Ganga Ram Hospital. ***Indian J Pediatr*** 2003; 70: 293-297.
5. Genetic Alliance UK [homepage on the Internet]. London (UK). [updated 2012 February 28; Accessed 2010 February 26]. Available from: http://www.gig.org.uk.

Related articles

Dastgiri S, Salehpoor F, Bigdeli A. An unusual time pattern of central nervous system anomalies in Northwestern Iran. ***Neurosciences (Riyadh)*** 2010; 15: 294.

Khasawneh NH, Al-Akayleh AT. Neural tube defects in newborns. ***Neurosciences (Riyadh)*** 2002; 7: 112-114.

Koul R, Chacko A, Javed J, Al-Hinai K, Zachariah M, Bulusu S, Rao TV. A profile of childhood neuropathies at Sultan Qaboos University Hospital in Oman. ***Neurosciences (Riyadh)*** 2002; 7: 92-98.

Idris MN, Sokrab TO. Autosomal dominant cerebellar ataxia type 1 in a Sudanese family. ***Neurosciences (Riyadh)*** 2002; 7: 83-85.