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How Accurate Is Diagnosis of Congenital Anomalies Made by Family Physicians?

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ABSTRACT

Background: Although family physicians have a key role in clinical management of many diseases and in community health, the accuracy of the diagnosis for congenital anomalies by family physicians still needs more investigations. The aim of this study was to assess the accuracy of family physicians in case detection and diagnosis of congenital anomalies in rural areas, northwest of Iran.
Methods: In a community-based study of 22500 children born between 2004 and 2012, all 172 cases of congenital anomalies diagnosed by family physicians were assessed by a qualified pediatrician in 47 health houses in rural areas of Tabriz District, northwest Iran. A group of 531 children was compared as control subjects.
Results: The overall sensitivity and specificity of family physicians' diagnosis for congenital anomalies were estimated 98% (95% Confidence Interval (CI): 95.9 to 100) and 100% (95% CI: 99.3 to 100), respectively. Sensitivity for diagnosis of congenital heart diseases was 97% (95% CI: 93 to 100), and for genitourinary tract, it was 86% (95% CI: 59 to 100). Specificity was estimated 100% for both groups of heart and genitourinary tract anomalies.
Conclusion: The performance of family physicians was found accurate enough in the diagnosis of congenital anomalies. Health care system may consider family physician program as an effective approach to detect and clinical management of congenital anomalies.

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Introduction

"Congenital anomalies affect 1 in 33 infants with 3.2 million birth defect-related disabilities every year in the globe. They may result in long-term disability with significant impacts on individuals, families, health care systems and communities".[1] They are the leading cause of prenatal mortality and childhood morbidity and disability in many countries. Congenital anomalies are the most common causes of death in children (1–59 months) in Iran.[2] Previous studies reported a prevalence ranging from 49 to 283 per 10 000 births in the country.[3-9] Total prevalence of congenital anomalies was 1.9 per 100 births between 2000 and 2011 in east Azerbaijan, northwest Iran.[10]

According to the World Health Organization (WHO) report 2008, the five main

features that generally identify a functioning Primary Health Care (PHC) system includes effectiveness and safety; person-centeredness; comprehensiveness and integration; continuity; and availability of a regular entry point (gate-keeping) for access to secondary and tertiary care.[11] Evidences show that a functioning PHC sector offers crucial advantages within health systems in terms of the health status of the public, improving health-care-related outcomes, lower health disparities, more equitable access to care, better quality of care and lower costs.[12] Health care spending in countries with such a system has been shown to be less than in settings with non-functioning or limited primary care system.[13]

Figure 1 illustrates the health system network of Iran. The PHC network in rural Iran has been described as an 'incredible masterpiece'.[14] Mostly financed by the state budget, nationwide implementation of the PHC networks started in 1981 in Iran with the establishment of health houses in rural areas. Following the introduction of PHC, health indices in Iran are now similar to the best in the region.[11] However, in recent years the system has lacked the level of flexibility to respond to emerging needs: increased life span of the population, rising burden of non-communicable diseases, migration to urban areas and growing expectations of the public for further access to qualified doctors.[15] In particular, the current PHC system seems unfit for addressing the observed shifts in disease patterns towards chronic conditions and non-communicable diseases.[16] In response, since 2004, the government has instituted health sector reform in rural areas through the introduction of two new policies including family physician program to improve the quality of services, and rural health insurance for all to enhance affordability.

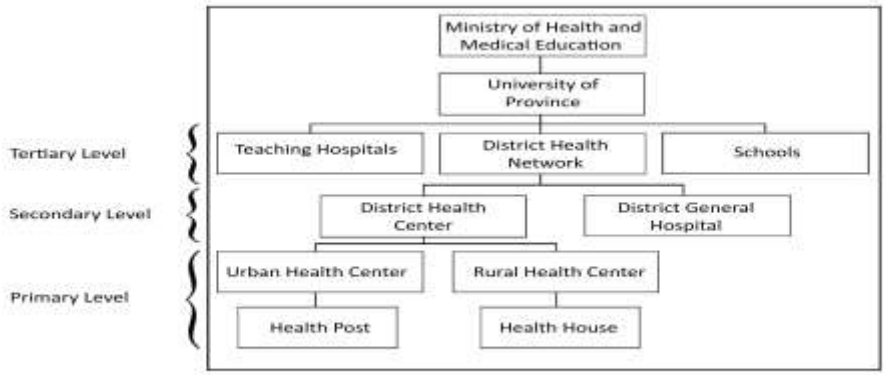

Fig. 1: Health system network in Iran

Important changes occur following establishment of this plan can be categorized into two categories. First, service delivery is for 'a defined population'. Second, is referral system in which it is predicted that one should utilize specialized services if needed, and in case s/he is introduced by her/his family physician, the insurance should pay most of his cost. The Ministry of Health is obliged to expand the reform to urban areas if the evidences prove its positive effects on health indices. Diagnosis and clinical management of congenital anomalies are now included in the services provided for family physician program. Thus each family physician examines all newborns in his /her catchment area with regular time intervals, proposed protocol, and applies laboratory tests when needed in order to screen them with respect to congenital anomalies. S/he also monitors and follows up all confirmed cases.

Accurate diagnosis and effective surveillance of congenital anomalies can play a major role in decreasing financial and emotional burden experienced by families caring for a child with birth defect(s). Studies examining the performance and effectiveness of the family physicians in various diseases have been published both inside and outside Iran.[17-23] A study in Sanandaj, western Iran showed the positive impact of the family physician program on function of health units in terms of availability of physicians and midwives and also insurance coverage at health centers in rural Area.[24] Hatam et al. reported that although the family physician plan has led to better and more regular service delivery and has better equipped the health centers, this plan has resulted in increased referrals to pharmacies, laboratories, and radiology centers and more referrals to family physicians in Fars province, southern Iran.[25] To our knowledge, however, there is no published data about the validity of birth defects diagnosis made by family physicians in the country.

The aim of this study was to determine the accuracy of family physicians in case detection and diagnosis of congenital anomalies in in rural areas, northwest of Iran.

Materials and Methods

The research setting was all 17 rural health care centers and their 47 affiliated health houses in Tabriz district, northwest Iran, in which 49 family physicians, 27 midwives and 96 Behvarzes worked. Rural population of Tabriz is about 158731 people. At this population, 22500 children under 8 years (live births born between 2004 and 2012) recruited for this study.Congenital anomalies were defined as structural defects, chromosomal abnormalities, inborn errors of metabolism and hereditary disease diagnosed before, at, or after birth.[26] The definition of the congenital anomalies was based on the standard coding system of the International Classification of Diseases (Version10) based on the primary diagnosis of anomaly.

Nearly all births regardless of the birthplace are recorded in rural areas. Each child has a health file including his/her health related life events. The coverage for health/medical information is nearly complete for those under 8 years of age.

The physicians in Iran are general practitioners with an MD degree without any academic specialty education in family medicine that could be different from countries where family physicians graduate from a specialty degree program in family medicine. They are admitted directly from secondary school to a 7-year undergraduate medical education as medical students. After initial phase of training typically on basic biological sciences students then spend 2.5 years in hospital rotations. Then they receive an additional 18 months of clinical internship. Following the internship, students graduate with an official medical license from the ministry of health. Then they start their clinical practice in either public health centers or private clinics and hospitals. Physicians working in public health centers have normally taken extra courses through different training packages and modules organized by district health centers. These individuals are then called "Family Physicians" in the country health system.

All 172 under 8 year of age alive cases of congenital anomalies confirmed by family physicians during their surveillance period (from birth up to 8 years of age) according to their health files with two control groups were studied. The first control group comprised all brothers and sisters of the cases (control group 1). Another group was selected from under 8 children who did not show any congenital anomalies in their health/medical records (control group 2).

A qualified general pediatrician who was an academic member of Tabriz University of Medical Sciences and had profound experience in the diagnosis and clinical management of congenital anomalies examined all subjects in the health houses and ordered the standard necessary tests according to the type of their anomalies if needed between May 2013 and September 2013. Data collec-

tion instrument was a checklist including demographic characteristics and information regarding the household (i.e. family size, age, sex). It also covered variables regarding the congenital anomaly description, laboratory and diagnosis tests.

Ethical Considerations

The study received ethics approval from the committee of ethics in Tabriz University of Medical Sciences. Informed consents were obtained from the mothers of all participants.

Data were analyzed using the STATA 11 statistical software package (STATA Corporation, College Station, TX). We calculated diagnostic indices (sensitivity and specificity) and 95% confidence interval (CI) for each index.

Results

A total of 721 children less than 8 years of age were invited to participate in the study of which 172 were cases with congenital anomalies according to their family physicians. Of 549 subjects without birth defects, 207 allocated to control group 1 and 342 to control group 2. Five children of control group 1 and 13 subjects of group 2 refused to attend the study for personal reasons. In total, 703 children were examined by a qualified pediatrician. The mean age of the study cases, control group1 and control group 2 were 47.7 (Standard Deviation: 27.6), 43.6 (Standard Deviation: 28) and 45.3 (Standard Deviation: 27.4) months, respectively. Table 1 shows the characteristics of study participants.

Table 1: Characteristics of study participants, northwest of Iran

		Cases* **n (%)**	**Control group 1**** **n (%)**	**Control group 2***** **n (%)**
Gender	Male	100 (58)	108 (53)	185 (56)
	Female	72 (42)	94 (47)	144 (44)
Age(month)	1-12	26 (15)	20 (10)	48 (15)
	13-59	76 (45)	105 (52)	177 (54)
	60-96	70 (40)	77 (38)	104 (31)

* Under 8 year of age with congenital anomalies/** Under 8 year of age children of cases' brothers and sisters
*** Under 8 year of age without congenital anomalies

According to the family physicians' diagnosis total prevalence of congenital anomalies was 113 per 10 000 live births between 2004 and 2012. Of 254 children diagnosed with birth defects, 82 had died due to different reasons and 172 survived.

Anomalies of nervous system and congenital heart diseases, ear/eye defects accounted proportionally for more than 55% of anomalies in the region. By contrast, categories of digestive system anomalies, genitourinary tract and defects of respiratory system accounted all for less than 10% of anomalies. The prevalence of anomalies of nervous system increased from 19 per 10 000 births in 2004-2006 to 32 in 2010-2012. In addition, the ear and eye defects showed an upward trend from 9 per 10 000 births in 2004-2006 to 15 in 2010-2012.

All 172 children assessed as having congenital anomalies of any kind by family physicians, were found presenting birth defects according to the diagnosis made by the study pediatrician. In 531 subjects classified by family physicians as not having birth defects, 2 cases identified with congenital anomalies based on the pediatrician. One diagnosed with ventricular septal defect categorized as congenital heart disease anomaly and another suffered from undescended testis, which grouped in the genitourinary tract anomalies.

Table 2 presents diagnostic indices of family physicians. The sensitivity and specificity of diagnosis made by family physicians for congenital anomalies were 98% (95% CI: 95.9 to 100) and 100% (95% CI: 99.3 to 100) respectively. Sensitivity for diagnosis of con-

genital heart diseases was 97% (95% CI: 93 to 100), and for genitourinary tract, it was 86% (95% CI: 59 to 100). Specificity was estimated 100% for both groups of heart and genitourinary tract anomalies.

Table 2: Sensitivity and specificity for the diagnosis of congenital anomalies made by family physicians, northwest Iran

Congenital Anomaly Category	Number of Anomalies	Sensitivity	95% Confidence Intervals	Specificity	95% Confidence Intervals
Nervous system anomalies (including NTDs)	47	100	–	100	–
Congenital heart diseases	46	97	93-100	100	–
Ear and eye anomalies	22	100	–	100	–
Musculoskeletal and connective tissue anomalies	11	100	–	100	–
Cleft lip with/without palate	10	100	–	100	–
Down's syndrome	10	100	–	100	–
Anomalies of limb	8	100	–	100	–
Digestive system anomalies	7	100	–	100	–
Genito-urinary tract anomalies	6	86	59-100	100	–
Respiratory system anomalies	3	100		100	–
Others	2	100	–	100	–
All congenital anomalies	172	98	95.9-100	100	–

Discussion

The family physician program is considered as one of the most remarkable reforms in the health care system to increase accessibility to physicians especially in deprived areas. In this study we investigated the capability of family physicians in diagnosing and clinical management of congenital anomalies in northwest of Iran.

Our findings indicated that family physicians assessment is highly sensitive and specific in detecting and managing congenital anomalies. To our knowledge there is no published original study concerning the diagnosis validity of family physicians for the congenital anomalies. However many studies focused on the accuracy of family physicians in diagnosis of various diseases and disorders.[27-31] A study in the north of Iran showed the positive effect of family physicians on prenatal care.[32] Rouhani et al. reported significant impact of the family physicians on the utilization of curative care in the country.[33] A report from western Iran showed the key role of the physicians on the increased utilization of the hospitalization.[34]

The high accuracy of diagnosis and clinical management of congenital anomalies in this study may be a result of personal and institutional characteristics of the current health system of the country. Rural Iran benefits from a well-established PHC network. The network is well organized and is credited with the improvements in health outcomes that have been observed since the 1980s in rural areas. Since 2004, the family physician program integrated to PHC network and strengthened its potentials through increasing accessibility to physicians, providing health insurance to completely rural population and improving referral system. As a result, case finding (i.e. birth defects) as one of the most important elements of the program has been activated. Other strength of PHC is its remarkable information system. This might have resulted in accurate detection of cases as well. The system gathers all health related data and provides health information from their catchment area, which is passed on to the higher levels. Since each Behvarz covers only about 1500 people and s/he are well anchored in her/his community, the likelihood of missing data is very low. Thus, the family physician program

team has proper access to health related information of the population under coverage and can properly screen and follow up them.

Another possible explanation to high validity of detecting congenital anomalies could be the characteristics of family physicians. These doctors provide comprehensive care to patients of all ages with all sorts of conditions. They also screen for early signs of serious conditions including congenital anomalies. Because they focus on the "whole patient" and provide treatment over long times, family physicians are uniquely suited to providing preventive care and managing chronic and complex conditions for everyone in the community. Many patients have long-standing relationships with their family doctor. Over time, the family physician comes to know a great deal about the patients' health history and medical needs.

We had a limitation in this study. Data came from live births only, stillbirth deliveries or very early fatalities were not enrolled into the study due to practical barriers. This may cause an underestimation of the accuracy and it means that if it was possible to do the study on stillbirth deliveries, as possible source of survival bias, due to the fact that stillbirth usually occurs because of more severe congenital anomalies we could expect higher accuracy. This is because diagnosis of severe cases is easier than subtle cases and we expect higher accordance between the physician and the pediatrician in diagnosing severe cases.

Conclusion

In conclusion, the performance of family physicians was found accurate enough in the diagnosis of congenital anomalies. Health care system may consider family physician program as an effective approach to detect and clinical management of congenital anomalies.

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Competing Interests

The authors declare that there is no conflict of interests.

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