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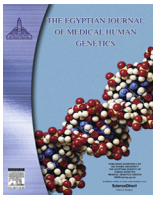

Original article

Study of congenital malformations in infants and children in Menoufia governorate, Egypt

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ABSTRACT

Congenital anomalies is one of the main causes of physical disabilities, stillbirths and neonatal deaths. The exact etiology of most congenital anomalies is unidentified but genetic and environmental causes are accused.

We aimed to study congenital anomalies regarding frequency, clinical pattern and associated risk factors.

A cross-sectional study was conducted on 100 infants and children with congenital anomalies attended to our pediatric genetic clinic at Menoufia University Hospital from October 2016 to October 2017. Detailed history taking, clinical examination and investigations including cytogenetic study were done.

Out of 100 cases, 51% have isolated anomalies and 49% have multiple anomalies, 14.2% had chromosomal abnormalities, 44.8% were diagnosed as genetic syndromes, while we did not reach the final diagnosis in 40.8% of cases. According to the ICD-10 classification of congenital anomalies musculoskeletal system anomalies were the most common in 48% of cases, followed by anomalies of the eye, ear, face and neck in 44% of cases. Anomalies of nervous system, circulatory system, genital organs, urinary system, chromosomal abnormalities, cleft lip and cleft palate occur in 26%, 22%, 18%, 12%, 7% and 6% of cases respectively.

Gastrointestinal anomalies in only 4% of cases taking into account that one case may have more than one affected system. According to the guidelines for case classification for the National Birth Defects Prevention Study, 2003, 51% had major anomalies, 18% had minor anomalies while 31% had both. Some cases had undergone immediate intervention e.g. meningomyelocele, encephalocele, omphalocele and gastroschisis. While other cases required later intervention e.g. hypospadius, cleft palate and cleft lip. Male gender, consanguineous marriage and lack of maternal folic acid supplementations were found in 54%, 43% and 59% of cases respectively, constituted the main risk factors.

Subjects and methods: proper physical examination, cytogenetic and molecular studies are important for the early intervention so prevention will be possible.

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1. Introduction

Congenital anomaly has been defined according to the World Health Organization as any morphological, functional, biochemical or molecular defects that may develop in the embryo and fetus from conception until birth, whether detected at birth or later [1]. Approximately 3 million fetuses and infants are born each year with major malformations [2]. The prevalence of congenital and genetic disorders in infants and young children in Egypt ranges from 2.8% in urban areas to 8.4% in rural areas [3].

The causes of congenital anomalies are divided into single gene defects, chromosomal aberrations, multifactorial disorders, teratogenic factors and those of unknown etiology. Even with the great advances in genetics over the last decade, the etiology of more than 50% of malformations is still unknown [4]. Approximately 2–3% of neonates have a single major malformation, and 0.7% has multiple major defects [5].

Structural anomalies can be classified into anomalies that are due to abnormal tissue development (malformation and dysplasia) and others which arise after tissue development (deformation and disruption) [6]. The anomalies which affect an infant's life expectancy, health status, physical or social functioning may be described as "major" anomalies. In contrast, minor anomalies are

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those with little or no impact on health or short-term or long-term function [7].

Early and precise diagnosis of a child with multiple congenital anomalies is important for management, genetic counseling concerning etiology, recurrence risk, prenatal diagnosis, screening and recommendation for evaluation of other family members. Diagnosis of a child who presents with multiple congenital anomalies is still a complex issue.

2. Aim of the work

To study congenital anomalies as regards their frequency, clinical pattern, nature and the linked risk factors as well as to integrate an approach to reach a diagnosis of a dysmorphic child.

3. Patient and methods

The current study is a cross-sectional study which was conducted on 100 infants and children with congenital anomalies who attended our pediatric genetics clinic or admitted in the pediatric department-Menoufia university hospital from October 2016 to October 2017. Their ages ranged from 1 day to 12 years .

The cases were classified according to the guidelines for case classification for the national birth defects prevention study, 2003 [7] into cases with major anomalies and cases with minor anomalies with some cases having both. The anomalies also were classified according to the affected system according to the International Statistical Classification of Diseases and Related Health Problems, 10th version, for 2007 [8]. Also cases were classified into cases with single anomalies and cases with multiple anomalies which were further exposed to categorization according to their pattern trying to reach a diagnosis. Down syndrome was excluded to avoid high figures of chromosomal abnormalities.

After taking written informed consent of parents of affected children. The work has been carried out in accordance to the code of Ethics of the World Medical Association (Declaration of Helsinki For Experiments in Humans). Detailed history was taken regarding gender, gestational age, residence, maternal and paternal age at the time of conception, history of maternal illness or drug intake, maternal exposure to infections or teratogens, smoking (passive or active), folic acid supplementation, mode of delivery, history of previous abortions or stillbirths and obstetric complications. Three generation family pedigree was constructed for each case.

Physical characteristics were reported including the general appearance, body shape and size, craniofacial examination, neck examination as regards length, webbing and neck swelling and examination of extremities regarding symmetry, shortening of limbs and abnormalities of the fingers and toes. Anthropometric measures including internipple distance & internipple index were taken. Craniofacial anthropometric measures were taken including horizontal measures (head circumference, head length, head breadth, intercanthal distance, interpupillary distance, outer canthal distance, palpebral fissure length, philtrum width and commissural distance) and vertical measures (ear length, nose length, philtrum length, lower lip to chin). These measures were interpreted according to charts for craniofacial anthropometry [9,10]. Photographs were taken to document the dysmorphic features and the parents gave their approval for the publication of these photographic materials.

Investigations were asked including TORCH screening, abdomino-pelvic ultrasound and echocardiography for all cases and specific imaging studies as indicated for each case such as skeletal survey, C.T skull and MRI brain. Karyotyping was done for cases with multiple anomalies. Also Intelligent Quotient test was asked as indicated.

Cases with multiple anomalies were diagnosed by comparison with known cases indicated by the diagnostic search engine databases e.g. OMIM, Face2gene library and Genetic Home Reference. Results were analyzed by descriptive statistical techniques recording the number and percentage of the studied variables.

4. Results

We had studied 100 cases whose ages ranged from 1 day to 12 years. They were 54 males and 46 females. Demographic data revealed that regarding gestational age, 88% were full terms, 12% were preterms. As regards maternal age parameters, 71% of mothers were between 20 and 35 years, 23% were above 35 years and only 6% were below 20 years. History of consanguinity was

Table 1
Classification of the cases according to the provisional diagnosis.

Group I Single anomalies (n = 51)	Group II Multiple anomalies (n = 49)		
	Group IIa: Chromosomal anomalies (n = 7)	Group IIb: Syndromes, sequence, association or developmental field defect (n = 22)	Group IIc: Unknown diagnosis (n = 20)
Encephalocele (n = 1) Cystic hygroma (n = 1) Macroglossia (n = 1) Cleft palate (n = 4) Cleft palate and lip (n = 2) Cupped ear (n = 2) Preauricular tags(n = 2) Pectus excavatum (n = 1) Gastroschisis (n = 1) Phocomelia (n = 2), Polydactyly (n = 5) Syndactyly (n = 3) Polysyndactly (n = 3) Umbilical hernia (n = 5) Inguinal hernia (n = 6) Hypospadias (n = 4) Epispadias (n = 2) Undescended testicles (n = 3) Talipes equinovarus (n = 3)	46,XY,t(2:9)(q21:q31) 46,X,t(X:13)(p22.2:q12) 46,XY, add(17)(p13) 46,XY,del(4)(p16) 46,XX, del (18)(p11.2) 46,XX,deletion (18)(q) 45,XX,der(13:14)(q10:q10)	Seckel syndrome (n = 4) Treacher Collins syndrome (n = 1) Kartagener syndrome (n = 1) Caudal regression syndrome (n = 1) Apert syndrome (n = 2) Arnold chiari malformation (n = 1) Noonan syndrome (n = 1) Rubinstien Taybi syndrome (n = 1) Achondroplasia (n = 3) Osteogenesis imperfecta (n = 1) VACTERL association (n = 2) Meningomyelocele sequence (n = 2) Acrorenal polytopic developmental field (n = 2)	Cases who have anomalies whose combination cannot be categorized into syndrome, association, sequence or developmental field defects and karyotyping was normal

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