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Original Article

Congenital anomalies in Tunisia: Frequency and risk factors

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ABSTRACT

Background. – Despite the high risk of recurrence of congenital malformations, there are no well-accepted preventive measures in developing countries like Tunisia. It is recommended that thorough epidemiological studies of congenital anomalies in this country are needed. The aim of this study is to assess the frequency and types of congenital anomalies in Tunisia and research some risk factors associated with occurrence of these anomalies.
Methods. – In this retrospective study, all the fetuses who were autopsied during 21 years period from February 1991 to December 2011 ($n = 9678$) at Service of embryofetopathology in the Center of Maternity and Neonatology of Tunis (CMNT) were studied. The classification of malformations was based upon the anatomical system affected. The differences in fetal/maternal characteristics between cases with or without congenital malformations were assessed using Chi^2 test.
Results. – Of the all 9678 autopsied fetuses, 4498 (46.47%) were diagnosed as being malformed fetuses. Anomalies of limbs (22.71%) and digestive and abdominal wall defects (14.76%) were mostly detected, followed by congenital brain defects (13.41%) and nephrourologic abnormalities (11.23%). A marked association of parental consanguinity with increased congenital anomalies rates was found ($P < 10^{-6}$, OR = 1.89, CI = 1.69–2.13).
Conclusion. – In Tunisia, surveillance and epidemiological evaluation of congenital anomalies underline the high frequency of these events. This will help to better target congenital anomalies prevention and screening policies in our population.

Introduction

Birth defects or congenital anomalies include all structural and functional alterations in embryonic or fetal development resulting from genetic, environmental or unknown causes, which result in physical and/or mental impairment [1]. Based on the World Health Organization report, about 3 million fetuses and infants are born each year with major congenital anomalies [2]. They are found in approximately 3% of newborns [3]. Birth defects can be isolated abnormalities or part of a syndrome and continue to be an important cause of neonatal and infant morbidity and mortality [4].

The pattern and prevalence of congenital anomalies may vary over time or with geographical location, reflecting a complex interaction of known and unknown genetic and environmental factors including socio-cultural, racial and ethnic variables [5].

The causes of congenital malformations may be divided into five broad groups [6–10]:

- single gene defects (mutant genes);
- chromosomal abnormalities;
- multifactorial disorders which are the result of interaction between genetic predisposition and presumed environmental factors;
- teratogenic factors
- those of unknown cause.

Abbreviations: CMNT, Center of Maternity and Neonatology of Tunis; WA, Weeks of amenorrhea.

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In most of the cases, the causes are complex and obscure and assumed to be a multifactor effect involving gene factors, environmental factors or their interaction [11].

Prevention of congenital anomalies must be based on epidemiological surveillance that reveals the extent of the problem and can even identify the risk factors [12]. In Tunisia, surveillance and epidemiological evaluation of congenital anomalies underline the high frequency of these events. This will help to better target congenital anomalies prevention and screening policies in our population [13]. The aim of this study is to assess the frequency and types of congenital anomalies in Tunisia and research some risk factors associated with occurrence of these anomalies.

Methods

This retrospective study concerning the period from February 1991 (date of the first autopsy) through December 2011 was carried out in the Service of embryofetopathology in the Center of Maternity and Neonatology of Tunis (CMNT), Tunisia. This service receives stillborns and deceased fetuses from all the other regional hospitals and private clinics in addition to those from the center.

The present study included 9678 consecutive cases of fetal autopsy over a period of 21 years. Only those cases, where both maternal and fetal records were available were included in the study. Autopsy was performed as per thorough protocol which included external examination, internal examination of thoracic and abdominopelvic cavity as well as removal of viscera, examination of head and neck, brain and spinal cord, examination of placenta and umbilical cord.

All reports on fetal examinations were recorded by the practitioners. The data of autopsied fetuses and their mothers – fetal sex, fetal weight, fetal term, mother's age, maternal blood group, consanguinity, origin, residence... – were pooled. The classification of malformations was based upon the anatomical system affected.

The differences in fetal/maternal characteristics between cases with or without congenital malformations were assessed using chi-square test by the statistical program "Epi- Info-version 6" [14]. To make statistical comparisons, 95% confidence intervals were reported. The rates were considered statistically significant at the 5% level ($P \leq 0.05$).

Results

Sample description

During the period between 1991 and 2011, 9678 fetuses were autopsied in service of embryofetopathology in CMNT. Among these fetuses, 46.47% of cases ($n = 4498$, $N = 9678$) have shown at least one congenital anomaly (minor or major) (Fig. 1).

The origin of fetuses was noted in 8710 cases. The rate of cases who came from CMNT was 72.87% ($n = 6347$). Against, 27.12% ($n = 2363$) of cases have been sent by other hospitals and private clinics.

The sex was noted in 9399 cases. Among these cases, there were 4991 (53.1%) males and 4408 (46.89%) females with a sex ratio (males/females) of 1.13.

Fetal weight was noted in 8835 cases. The average weight was 1026.04 g, with extremes ranging from 2 to 6800 g.

Fetal term was noted in 8734 cases. The median term fetal was between 24 and 25 weeks of amenorrhea (WA), with extremes ranging from 9.5 to 40.5 WA.

The state of birth was specified for 7728 fetuses. The rate of stillborn fetuses was 88.49% ($n = 6839$). However, 11.5% ($n = 889$) of cases have been deceased in neonatal period.

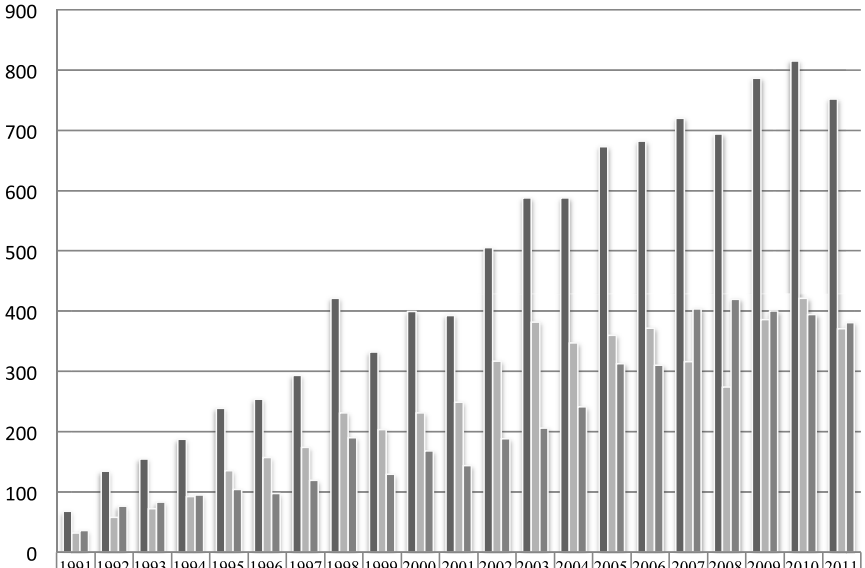
The total number of mothers was 9354, due to presence of the twins in our series.

Maternal age was noted in 7930 cases. The average maternal age was 31 years, with extremes deriving from 14 to 51 years.

The number of gestation was specified in 8094 mothers. The average number of gestation was 2.59, with extremes going from 1 to 14.

Parity was specified in 8094 mothers. It ranged between 0 and 13, with an average of 1.6.

The blood type was identified for 5375 mothers. The positive rhesus was the most common with a percentage of 87.64%



	1991	1992	1993	1994	1995	1996	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006	2007	2008	2009	2010	2011
All autopsied fetuses	68	134	155	187	239	254	293	421	332	399	393	505	588	588	673	682	720	694	786	815	752
Fetuses without congenital anomalies	32	58	72	92	135	157	174	231	203	231	249	317	382	347	360	372	316	274	386	421	371
Fetuses with congenital anomalies	36	76	83	95	104	97	119	190	129	168	144	188	206	241	313	310	404	420	400	394	381

Fig. 1. Distribution of fetuses with or without congenital anomalies.

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