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Research Article

Congenital Malformations In 101 Neonatal Cases

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Abstract

Introduction: A congenital malformation refers to a birth defect or abnormality that is present at birth. These malformations can affect various parts of the body, such as organs, limbs or systems. They can result from genetic factors, environmental factors during pregnancy or a combination of both.

Method: This work summarizes a retrospective descriptive and analytical study of all newborns with congenital malformations admitted to the neonatal intensive care unit of the Mohammed VI University Hospital in Tangier over a period from April 2023 to July 2024.

Results: From a sample of 833 newborns, we collected 101 cases of congenital malformations, representing a hospital frequency of 12.12%. The most frequent malformations were polymalformative syndromes (n = 58), followed by cardiovascular malformations (52), digestive and abdominal wall malformations (49), face and neck (24), central nervous system (19), osteoarticular (15), urogenital and renal (12).

Conclusion: Congenital malformations constitute a real public health problem in our context and pose a problem of etiological diagnosis and adequate management.

Keywords: Antenatal Diagnosis; Pregnancy Monitoring; Consanguinity; Congenital Malformation; Newborn; Prognosis; Karyotype; Genetic Counseling; Multidisciplinary Care

Introduction

Congenital malformations are a group of diverse disorders of prenatal origin that can be caused by single gene abnormalities, chromosomal disorders, multiple hereditary factors, environmental teratogens and micronutrient deficiencies. Certain maternal infectious diseases, such as syphilis or rubella, are a significant cause of congenital malformations in low and middle-income countries. Maternal diseases, such as diabetes mellitus, certain medical conditions, such as iodine or folic acid deficiency and exposure to medications and drugs including alcohol and tobacco, certain environmental chemicals and high-dose radiation are other factors that cause congenital malformations.

In Morocco, few studies have focused on congenital malformations, so this work sets the following objectives:

1. To identify the epidemiological, clinical, therapeutic and evolutionary profile of congenital malformations in the neonatal intensive care unit of the Mohammed VI University Hospital in Tangier
2. Determine the main risk factors involved in the occurrence of congenital malformations in our contex
3. Establish preventive measures to address the occurrence of these malformations
4. Evaluate the management and prognosis of newborns with congenital malformations

Methodology

This work summarizes a retrospective descriptive and analytical study of all newborns with congenital malformations admitted to the neonatal intensive care unit of the Mohammed VI University Hospital in Tangier over a period from April 2023 to July 2024.

Results

In a sample of 833 newborns, we collected 101 cases of congenital malformations, representing a hospital frequency of 12.12%. Regarding maternal characteristics, the average age of the mothers was 31.3 years, the average parity was 2.66 with extremes between 1 and 8. In our series, 23.7% of mothers had a history of miscarriage. For prenatal follow-up, 56.4% of mothers had at least 3 prenatal consultations. Iron and folic acid supplementation was identified in 62.4% of mothers. Regarding the mother's pathological history, 22.8% of mothers had a chronic pathology, including diabetes mellitus in 10.9% of cases and high blood pressure in 5.9% of cases. The notion of taking medication was found in 5% of mothers. Fenugreek intake during pregnancy was reported in 2% of cases, toxic habits including alcohol and tobacco consumption were not reported in any mother, however, 3 mothers (3%) were exposed to passive smoking at home. Only one malformed newborn was born through medically assisted procreation.

Regarding geographical origin, we observed a predominance of urban origin among mothers of malformed newborns in 89.1% of cases. Family history of congenital malformations was found in 7% of cases while parental consanguinity was found in 22 of the cases, it was 1st degree in 18% of the cases. Regarding neonatal characteristics, the mean age of diagnosis was 3.98 days, the sex ratio was 1.10 with a slight male predominance and sexual ambiguity in 2% of cases. The majority were full-term newborns with a total of 67 cases, including 51 born vaginally, eutrophic (92.1%) and from Tangier (n = 60).

The most frequent malformations were polymalformative syndromes in 58 cases, followed by cardiovascular malformations in 52 cases, digestive and abdominal wall malformations in 49 newborns, face and neck malformations in 24 cases, central nervous system malformations in 19 cases, osteoarticular malformations in 15 cases, urogenital and renal malformations in 12 cases. For cardiovascular malformations, patent ductus arteriosus was the most common malformation in 15 cases, while esophageal atresia was the most common digestive malformation in 19 cases. Mongoloid facies was the most common malformation of the face and neck in our study 5.9% (N = 6), followed by micro-retrognatism in 3% of cases. Hydrocephalus was the most common central nervous system anomaly with 10.89% of cases (N = 11), followed by spina bifida 7.92% (N = 8) and Arnold Chiari malformation 2.97% (N = 3).

Among the musculoskeletal system anomalies, club feet were the most common malformation with 4%. Sexual ambiguity and hypospadias were the most common among urogenital and renal malformations, respectively 3% (N = 3) and 2% (N = 2). Malformations of other organs and systems were uncommon in our study, particularly those of the respiratory system and cutaneous-phaneric system and the oral cavity, which accounted for less than 4%. Karyotyping was performed in (n = 8) newborns. Morphological examinations: Chest X-ray, abdomen without preparation, barium enema, ultrasounds (heart echo, trans-fontanellar ultrasound, abdomino-pelvic and renal ultrasound), CT scan or CT angiography, magnetic resonance imaging were widely performed in our study, systematically as part of the malformation assessment and according to the warning signs. Newborn care was surgical in 59 cases and medical in 30 cases. Eleven parents received psychological support and 12 parents received genetic counseling. In our sample, 35 of the newborns did not survive during hospitalization.

Congenital Malformations	Number of Cases	Percentage
Poly malformations	58	23.77%
Cardiovascular malformations	52	21.31%
Digestive malformations/abdominal wall	49	20.08%
Facial and neck malformations	24	9.83%
Central nervous system malformations	19	7.78%
Osteoarticular malformations	15	6.14%
Urogenital and renal malformations	12	4.91%
Malformations of the oral cavity	9	3.68%
Skin malformations	3	1.22%
Respiratory malformations	3	1.22%
Total	244	100%

Table 1: Summary of congenital malformations presented by newborns.

Discussion

The World Health Organization defines congenital malformations as morphological and functional anomalies present at birth that originate in the genetic makeup of the embryo or in an extrinsic defect in its development in utero induced by infectious, metabolic, medicinal causes, ionizing radiation and toxic substances (1 and 2). It is estimated that 5 to 10% of malformations are due to exogenous or environmental causes and 20 to 30% to genetic or endogenous causes (gene mutations or chromosomal abnormalities) [1]. They constitute a real public health problem at the global level due to the high morbidity and mortality associated with them [3]. Each year, approximately 8 million children or 6% of the total number of births worldwide, are born with a serious congenital disorder according to the WHO [3]. However, the prevalence of major congenital malformations varies among types, severity and geographic regions due to lack of data and underreporting of cases [4].

In the United States, they affect about 3% of all pregnancies [5]. This frequency is even higher in resource-limited (6.4%) or middle-income (5.6%) countries [3,6]. In sub-Saharan Africa, different figures are reported in the literature. This prevalence is 55 in Ethiopia, 58 in the Democratic Republic of Congo, 66 in Uganda and 88 in Burkina Faso per 10,000 births respectively [3,7,8]. Globally, congenital malformations are the fifth leading cause of potential loss of life and significant causes of mortality, morbidity and disability in children [3]. Despite their clinical importance and despite the adverse consequences for the affected individual, a burden for the family and for caregivers, few studies have been conducted on the predisposing risk factors. On the etiological level, 50% of malformations are still of unknown cause [11]. Certain maternal infectious diseases, such as syphilis or rubella, are an important cause of congenital malformation in low- and middle-income countries [3].

Other factors that cause birth defects include certain maternal conditions, such as iodine or folic acid deficiencies and exposure to medications and drugs, including alcohol; and rarely to certain environmental chemicals and high-dose radiation [3]. Most congenital malformations are detected by diagnostic means (genetic counseling, prenatal diagnosis, obstetric ultrasound and medication (folic acid intake). Their prognosis depends on the early diagnosis and treatment [2]. Advances in imaging have greatly increased the possibilities of detection antenatal and improved the quality of postnatal analysis of certain malformations.

In our study, 101 cases of congenital malformations were recorded over a period of 15 months, representing a frequency of 12.12%. This result tends to be closer to that reported by Kaboré, et al., who found a frequency of 9.2% [13]. A study conducted by Sabiri, et al., at the Souissi maternity hospital in Rabat reports a prevalence of 4% out of a total of 1000 births, this rate is significantly lower than that found in our work [14]. Concerning maternal age in our study the age group over 35 years was the most represented with 35.80%, our result tends to be closer to that reported by Youssra (27.7) and Sabiri, et al., (27.2 +/- 5) and Ayayen (28 years) [2,14,15].

On the other hand, Sabbou M and Youl H, found the highest rate for the age group of 20 to 25 years with a rate of 23.88% and 27.27% respectively, Hama A, et al., found that the age group of 20-29 years is the most represented (50.63%) with an average age of 24.91+/6.02 years and extremes of 12 to 43 years [1,16,17]. This result is close to those of Kaboré A and Coll and Alou S, who found respectively 48.8% and 70.6% for the same age group with an average age of 25 years and 26 years [13,18].

Regarding iron and folic acid supplementation, 62.4% of mothers (N = 63) in our study had benefited. Our results are consistent with those reported by a study carried out in Senegal (N = 66) [2]. Unlike the study by Kamla, where none of the mothers had been supplemented, Werler, et al., showed that taking 400 ug/day of folic acid one month before and one month after the last period significantly reduced the risk of occurrence of neural tube closure defects by 70% [14,22].

In Canada, there was a 40% reduction in the prevalence of neural tube defects and a 46% reduction in spina bifida cases after the implementation of folate fortification measures, compared to births occurring in 1996, which corresponds to the end of the pre-fortification period [15]. Furthermore, a recent study by Feng, et al., demonstrated that folate supplementation significantly reduces the risk of congenital heart disease [25].

Concerning maternal pathologies, the frequency of congenital malformations is high in women with chronic pathologies, particularly diabetes [14]. In our study, 22.8% of mothers had a chronic pathology, this was diabetes mellitus in 10.9% of cases and arterial hypertension in 5.9% of cases. Our result is significantly higher than that reported by Youssra M, with a rate of 4.17% [25].

The study conducted by Sabiri, et al., concluded that long-standing maternal diabetes was significantly linked to the occurrence of congenital malformations and that alterations in periconceptional glycemic control increase the risk of malformations [14].

This risk is particularly well known for diabetes pre-existing during pregnancy. However, several studies based on patient registries or cohort studies, prospective or retrospective, have reported an increased risk of malformations in diabetes [14,28]. Bateman, et al., demonstrated that both treated and untreated hypertension were associated with a significant increased risk of cardiac malformations [30].

For medication intake in our study, it was found in 5% of mothers. This result is superimposable to that reported by Ayayen [2]. For Youssra M, the notion of taking medication was found in 20.83% of cases. This result is significantly higher than that found in our study [15].

The use of levothyroxine (2 cases), methyldopa (1 case), beta2-mimetic (1 case) and a cough suppressant (1 case) was observed. On the other hand, Ayayen (2) noted the taking of an antihypertensive (Alpha-methyl dopa), Antibiotic (amoxicillin-clavulanic acid), Antifungal (Fluconazole) and an Antiemetic (metopimazine).

According to the study by Sabiri, et al., in the group of malformed newborns, 55% of women took medication during pregnancy, mainly antiepileptics such as phenobarbital, carbamazepine or sodium valproate [14]. The use of these substances during pregnancy increases the prevalence of congenital malformations by two to six times. The teratogenic effect of drugs is observed especially when they are taken during the first trimester of pregnancy [2].

For the intake of fenugreek during pregnancy it was reported in 2% of cases in our series. On the other hand, according to Youssra M, 16.67% of mothers of malformed newborns reported the notion of consumption of fenugreek during pregnancy [15]. In Morocco, a study reported the occurrence of congenital malformations such as hydrocephalus and spina bifida after ingestion of fenugreek during pregnancy [31]. The notion of taking medicinal plants was not found in our study.

For the study of toxic habits, in our series, no mother of malformed newborn had not smoked or consumed alcohol during pregnancy. In contrast, 3 mothers (3%) were exposed to passive smoking at home. This result is significantly lower than that reported by Youssra M who noted that 25% of mothers were exposed to passive smoking [15].

Rabah, et al., reported in their study that 55.44% of mothers were exposed to tobacco, both actively and passively and they concluded that there was a significant association between maternal smoking and the occurrence of congenital malformation [35]. In our study, there was a predominance of urban origin among mothers of malformed newborns (89.1%). Therefore, geographical origin represents an important risk factor in our study.

This result is superimposable with those of Sabbou M, Kaboré A, et al., and Youssra M, who found that congenital malformations are more frequent in urban areas than in rural areas [13,15,16]. Family history of congenital malformations was found in 7% of cases. This result is significantly lower than that found in the study by Hama A, et al., where family history of malformation was found in 16.46% of cases [1]. On the other hand, Youssra M found a percentage of 4.17% concerning family history of malformations [15].

In the study conducted at the Souissi maternity hospital in Rabat, a family history of congenital malformations was present in 27.5% of the group of malformed newborns and in 2.2% of the group of healthy newborns. Therefore, heredity represents a statistically significant risk factor for the occurrence of congenital malformations. For consanguinity, twenty-two malformed newborns were the result of consanguineous marriages, a rate of 22%. This consanguinity was first-degree in 18% of cases.

Our result is lower than that reported by Youssra M, who found a consanguinity rate of 33% [15]. The study carried out at the Souissi maternity hospital in Rabat (14) found a higher rate of consanguinity among malformed newborns (48.7%). A study carried out in Senegal noted a consanguinity rate of 40%, with a predominance of second-degree consanguinity [2]

According to a study conducted by the National Institute of Hygiene, the prevalence of consanguinity in Morocco is estimated at 15.25% nationwide [39]. On the other hand, Youl H, found a consanguinity rate of 13.6%. Significantly lower than that found in our study [17]. For the type of congenital malformations, it was found that polymalformative syndromes were present in 23.77% and occupied the first rank. Our result is lower than that of Alou S, who found that polymalformative syndromes are the most frequent with a rate of 34.1% [18].

On the other hand, Hama A, et al., found that polymalformative syndromes represented a rate of 22.78% and occupied the 4th rank [1]. Cardiovascular malformations were present in 21.31% of cases, ranking second. Our result is consistent with that reported by Ayayen [2].

On the other hand, Kabore reported that these malformations were in the first rank. In our series, digestive and abdominal wall malformations occupied the 3rd rank with a rate of 20.08%. This result is consistent with that reported by Hama A, et al., who showed that abnormalities of the digestive tract and anterior abdominal wall represented 21 cases or 26.58% and came third
The same observation was made by several authors such as: Kamla J, et al., found 23.3% and ranked them third [22].

On the other hand, Alou S, noted that malformations of the digestive tract and the abdominal wall comes in 4th place [18]. It has been found that malformations of the face and neck come in 4th place rank with a rate of 9.83%. On the other hand, Ayayen noted that malformations of the face and neck were in 1st place. While Hama A, et al., objectified that these malformations were ranked last.

Nervous system anomalies were present with a rate of 7.78% or 5th place. This result is close to that reported by Ayayen who found that central nervous system malformations occupied 6th place with 5.8% [2]. Concerning osteoarticular malformations, a rate of 6.14% was observed or 6th place. This result does not agree with any study.

On the other hand, Hama A, et al., reported that 31.64% of malformations concern the limbs, which is the first in the series [1]. Youl H and Youssra M, found respectively 42.42% and 37.5% of limb malformations in the First rank [15]. In our series, urogenital and renal malformations came in 7th place with a rate of 4.91%. This result was shared by several authors who found that these malformations were not frequent [1].

Kaboré A et al [13] observed 8% of urogenital malformations in 4 years. Alou S, reported 2.4% in his series [13]. Youl H, collected only 3 cases in 6 months, i.e. 4.54% [17]. In our series, we found that antenatal diagnosis was performed in 22 cases, at a rate of 21.8%. Our result was significantly higher than that reported by Ayayen who reported that antenatal diagnosis was performed in only 9% of cases [2]. This deficit in antenatal diagnosis is explained on the one hand by poor ultrasound monitoring of pregnancies and on the other hand by the lack of qualified personnel for the correct interpretation of these ultrasounds.

Karyotyping was performed on 8 malformed newborns, representing a rate of 7.9%. This result was slightly higher than that reported by Ayayen who found that karyotyping was performed in only 3 cases. In our study, no newborns benefited from cytogenetic tests such as molecular studies and FISH. The same observation was made in a study carried out in Senegal [2]. Genetic counseling was performed on only 12 parents, at a rate of 11.9%.

Conclusion

In our context, congenital malformations are dominated by poly-malformative syndromes, followed by malformations of the cardiovascular system, digestive system and abdominal wall, malformations of the face, neck and central nervous system. The prognosis of these malformations depended on the type and quality of care, explaining the high mortality of these disorders, as well as the functional disabilities. Specialized care by a multidisciplinary team is crucial to optimize the long-term results of these malformed newborns. Screening of couples with a pattern of consanguinity, monitoring of pregnancies and prenatal diagnosis should be priorities of public health programs in our context.

Conflict of Interests

The authors declare that they have no conflicts of interest.

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Author Contributions
The authors contributed equally to the work.

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