Visible congenital malformations in Ouagadougou (Burkina Faso).

Abstract

Objective: To gain a better understanding of congenital malformations and to help reduce neonatal morbidity and mortality, particularly in a context of resource-limited countries. **Methods.** This descriptive cross-sectional study with retrospective and prospective data collection over the period from November 2019 to January 2022 was conducted in the Neonatology Unit of the Centre Hospitalier Universitaire Pédiatrique Charles de Gaulle of Ouagadougou, Burkina Faso (West Africa). Records of neonates with a visible malformation, neonates whose parents were accessible by telephone were included. **Results**. The frequency of congenital malformations was 8.4%. They affected females (51.1%), first-born siblings (25%) and full-term babies (69.6%). Osteoarticular malformations (38%), those of the eye, ear, face and neck (22.81%), followed by those of the digestive system (21.64%) were the most frequent. The case-fatality rate was 54.3%. **Conclusion**. Visible congenital malformations are a major cause of morbidity and mortality among neonates in our unit. To help reduce the morbidity and mortality associated with malformations, awareness-raising, early detection, prevention and improved medical care for affected neonates are needed. This is all the more true in countries where medical resources may be limited and surveillance systems need to be put in place.

Keywords: **Newborn, Congenital anomalies, Prevalence, Risk factors, Diagnosis, Mortality**

Introduction

Congenital malformations are defined as abnormalities of body structure or function present at birth. Also known as congenital anomalies, these conditions develop during the prenatal period and can be identified before, at birth or later in life [WHO 2010]. In current practice, the focus is generally on major structural malformations. These are defined as structural changes that have significant medical, social or aesthetic consequences for the individual concerned, and usually require medical intervention (e.g. cleft lip and spina bifida). These major anomalies (e.g. neural tube defects, heart disease, Down's syndrome) account for most of the death, morbidity and disability associated with congenital anomalies. In contrast, minor congenital anomalies (e.g. single palmar crease, clinodactyly), although more frequent, are structural changes that do not pose a significant health problem during the neonatal period and generally have limited social or aesthetic consequences for the individual concerned. Major anomalies may be associated with minor ones [WHO Birth defects].
In this article, we will focus on structural malformations.
It is estimated that 6% of neonates worldwide are born with a congenital anomaly, resulting in hundreds of thousands of deaths. However, the actual number of cases could be much higher, as statistics often fail to take into account pregnancy terminations and stillbirths [WHO 26/08/2023]. In 2021, 192,862 malformed births were reported in Europe [Loane 2023]. Congenital disorders are one of the main causes of the global burden of disease, and low- and middle-income countries are disproportionately affected (94%). These areas are also less likely to have facilities to treat reversible conditions such as clubfoot, leading to more pronounced and long-lasting effects [WHO 28/08/2023]. In Africa, in the absence of national surveillance programs, the data reported are patchy, mostly hospital-based. A frequency of 6.8% in Côte d'Ivoire [Bénié 2021], 7.9% in Niger [Nouhou 2022], for example, has been reported. In Burkina Faso, recent data show a frequency of congenital malformations of 3% in Ouahigouya [Ouermi 2021] and 9.2% in Ouagadougou [Kaboré 2020]. In our context of a

country with limited resources, malformations pose a problem of identification, etiological diagnosis, therapeutic management and psychosocial support.

The aim of this work was to study visible congenital malformations, focusing on their epidemiology, clinical features and evolution. The information thus gathered should be of use to health professionals in improving management, and to health decision-makers in devising strategies to combat these emerging pathologies.

Patients and methods

Study context

CHUP-CDG is a 3rd level referral hospital in Burkina Faso dedicated solely to the care of children's health problems. It has a pediatric surgery department and a medical department. The neonatology unit (NU) where this study took place is part of the latter department. With a capacity of 40 places, the NU receives neonates from maternity units in Ouagadougou and other regions of the country.

Type and period of study

This was a descriptive cross-sectional observational study with both retrospective and prospective data collection covering the period from November 24, 2019 to January 31, 2022.

Inclusion criteria

All neonates with a major or minor structural anomaly recognizable on physical examination at birth or within 28 days of birth hospitalized in the UN during the period whose parents gave their consent were included in this study.

Non-inclusion criteria

Internal malformations without clinical expression (e.g. certain non-cyanogenic cardiopathies), functional malformations (e.g. congenital metabolic disorders), deformities and neonates whose parents could not be reached by telephone were not included in this study.

Data collection and analysis

Information sources included hospitalization registers, unit activity reports, hospital statistical yearbook and patient files. Diagnosis was made by the pediatrician on the basis of a clinical examination of the neonate. A data collection form designed for the purpose of this study was used to collect the data. Data collection techniques included reviewing patient files and telephoning parents to fill in information missing from the medical record. The data collected were entered on a computer using Access software (Microsoft, USA), then transferred and analyzed using Epi Info software (CDC, Atlanta). For all qualitative variables, frequencies were generated and expressed as percentages (%). For quantitative variables, means were calculated with standard deviation.

Studied variables

The variables studied were the number of malformed neonates hospitalized; the child's age and sex; the parents' age, residence and occupation; age of pregnancy at birth, birth weight, ante-natal history (gravidity, stillbirth, abortion), number of antenatal care visits (ANCs), antenatal check-up, site, type, and number of malformations, existence of consanguinity between the two parents, pathologies during pregnancy, use of teratogenic drugs during pregnancy, contact with pesticides, length of hospital stay, mode of discharge (normal, death, transfer, discharge against medical advice).

Results

Frequency of malformations

A total of 1,158 neonates were admitted to the NU during the period. There were 97 cases of visible congenital malformations, giving a frequency of 8.4%.

Descriptive characteristics of parents

The average age of the mothers of malformed neonates was 29.5 ± 7 years [17, 46]. Their age ranged from 20 to 35 in 68.5% of cases. They were housewives in 56.2% of cases.

The average age of the fathers was 39.6 ± 9.1 years [21, 68], with 59.5% aged over 35. They were farmers in 24.7% of cases.

Parents lived in urban areas in 79.8% of cases, in Ouagadougou in 50.6% of cases.

Table 1 shows the social-demographic characteristics of the parents of malformed neonates.

Table 1: Social-demographic characteristics of parents of malformed neonates admitted to the neonatology unit of the Centre hospitalier universitaire Charles de Gaulle, Ouagadougou, Burkina Faso, 2019-2022 (n=89).

Parents' social-demographic characteristic	**Frequency**	**%**
Mother's age group (years)		
< 20	10	11.2
20-35	61	68.5
> 35	18	20.2
Father's age range (years)		
21-35	36	40.4
> 35	53	59.5
Residence		
Urban	71	79.8
Rural	18	20.2
Mother's occupation		
Housewife	50	56.2
Employee	12	13.5
Informal sector	11	12.4
Student	11	12.4
Retailer	5	5.6

Parents' social-demographic characteristic	Frequency	%
Father's occupation		
Farmer	22	24.7
Retailer	21	23.6
Employee	21	23.6
Informal sector	21	23.6
Student	2	2.2
Gold digger	2	2.2

Maternal obstetrical history

The number of pregnancies per woman varied from one to ten, with the majority (34.8%) between two and three . Mothers had had at least one abortion in 6.7% of cases, and at least one stillbirth in 11.2%.

In 51.7% of cases, mothers had attended at least four ANCs.

Infectious disease tests included syphilitic (48.3%), toxoplasmic and rubella (3.4%) serologies.

Obstetrical ultrasound was performed in 78.6% of women. An antenatal diagnosis of malformation was made in 14.3% of cases.

Table 2 shows the distribution of mothers according to their obstetrical history.

Table 2: Obstetric history of mothers of malformed neonates admitted to the neonatology unit of the Charles de Gaulle University Pediatric Hospital, 2019-2022, Ouagadougou, Burkina Faso (n = 89).

Maternal obstetrical history	**Frequency**	**%**
Gesture class		
Primigravida	22	24.7
Paucigravida	31	34.8
Multigravida	24	27.0
Large multigravida	12	13.5
Stillbirth		
0	79	88.8
1	8	9.0
2	1	1.1
3	1	1.1
Abortion		
0	83	93.3
1	4	4.5
2	1	1.1
4	1	1.1

Characteristics of neonates

On admission, the average age of the neonates was 1.8 ± 2.9 days [0, 15].

Table 3 shows the social-demographic characteristics of malformed neonates,

Table 3: Social-demographic characteristics of malformed neonates admitted to the neonatology unit of the Centre hospitalier universitaire Charles de Gaulle, Ouagadougou, Burkina Faso, 2019-2022 (n = 92).

Social-demographic characteristics of neonate	**Frequency**	**%**
Age range (days)		
0 - 3	77	83.7
4 - 28	15	16.3
Gender		
Male	42	45.6
Female	47	51.1
Ambiguity	3	3.3
Sibling rank		
1st	23	25.0
2nd	19	20.6
3rd	14	15.2
4th	12	13.0
5th	13	14.1
≥ 6th	11	12.0

The neonates were born at term in 69.6% of cases (64/92). They were premature in 28.3% (26/92) and born post-term in 2.2% (2/92).

Mean birth weight was 2225 ± 707 g [945, 4100]. Low birth weight (LBW) neonates accounted for 65.2% (60/92) of the sample.

Characteristics of malformations

Type and site

Osteoarticular malformations accounted for 38.0% of cases, malformations of the eye, ear, face and neck for 22.8%, and digestive malformations for 21.6%.

The frequency and location of malformations are shown in Table 4.

Table 4: Type and site of malformations in neonates admitted to the neonatology unit of the Centre Hospitalier Universitaire Pédiatrique Charles de Gaulle, Ouagadougou, Burkina Faso, 2019-2022 (n = 171).

Anatomical type and site of malformations	**Frequency**	**%**
Osteoarticular system	**65**	**38.0**
Clubfoot	23	13.4
Polydactyly	19	11.1
Genu recurvatum	5	2.9
Flat foot	4	2.3
Hand club	3	1.7
Amelia	2	1.2
Hand hypoplasia	1	0.6
Agenesis of the fingers	1	0.6
Thigh angulation	1	0.6
Genu flexum	1	0.6
Genu valgum	1	0.6
Femur hypoplasia	1	0.6
Micromelia	1	0.6
Hypoplastic leg bones	1	0.6
Thoracic excavation	1	0.6
Eye, ear, face and neck	**39**	**22.8**
Micro retrognathia	9	5.3
Low-set ears	8	4.7
Short neck	7	4.1

Anatomical type and site of malformations	Frequency	%
Exophthalmos	2	1.2
Microphthalmia	2	1.2
Arhinia	2	1.2
Hypotelorism	1	0.6
Hypertelorism	1	0.6
Slanted eyes	1	0.6
Congenital cataracts	1	0.6
Limited palpebral opening	1	0.6
Hypoplasia of the pinna	1	0.6
Absence of ear orifice	1	0.6
Facial dysmorphia	1	0.6
Single nostril	1	0.6
Digestive system	**37**	**21.6**
Omphalocele	13	7.6
Cleft lip and palate	10	5.8
Siamese twins	4	2.4
Macroglossia	3	1.7
Ogival palate	3	1.7
Microglossia	2	1.2
Hypoplasia of the abdominal wall muscles	2	1.2
Anal imperforation	1	0.6
Narrowed oral cavity	1	0.6
Central nervous system	**17**	**9.9**
Spina bifida	7	4.1
Encephalocele	2	1.2
Microcephaly	2	1.2

Anatomical type and site of malformations	Frequency	%
Macrocephaly	2	1.2
Craniostenosis	1	0.6
Sacra-coccygeal dimple	1	0.6
Urogenital system	**10**	**5.8**
Hypoplasia of the external genitalia	4	2.4
Sexual ambiguity	3	1.2
Hypospadias	3	1.7
Skin and appendages	***3***	**1.8**
Ichthyosis congenita	1	0.6
Single transverse palmar fold	2	1.2

Number of malformations

There were 171 types of malformation, an average of 1.9 malformations/neonate [1, 6]. Isolated malformations accounted for 53.3% of cases, and poly malformations 46.7%. Among poly malformations, four syndromic entities were identified. These were Prune Belly syndrome (two cases), Pierre Robin sequence (two cases), Down's syndrome (two cases) and osteogenesis imperfecta (two cases).

Etiological factors

Consanguinity

Consanguinity between the two parents was found in 2.2% of cases.

Maternal pathologies

There were three cases of malaria, one case of pneumonia and two cases of pre-eclampsia. There were no cases of diabetes.

Drugs

During the third trimester of pregnancy, two mothers self-medicated with an antimalarial drug (artemether-lumefantrine), while one mother took an antibiotic (amoxicillin). No cases of drug use were reported in the first or second trimester.

Pesticides

In 7.9% of cases, mothers were in contact with unspecified agricultural pesticides during pregnancy.

In-hospital outcome

The mean length of hospital stay was 17.5 ± 19.7 days [1, 104]. Discharge was normal in 44.6% of cases, and death in 54.3%. One case was transferred to neurosurgery.

Discussion

Frequency of malformations

In this study, the frequency of malformations was 8.4%, that result was higher than that reported by some authors in Côte d'Ivoire [Bénié 2021] (6.8%), Niger [Ramatou 2022, Nouhou 2022] (2.6%, 7.9% respectively), Democratic Republic of Congo (DRC) [Mashako 2017] (3.4%), [Monzango 2018] (5.1%), Egypt [El Awady 2021] (7.4%), India [Madhura 2020] (6.1%). On the other hand, it is lower than that reported in an earlier study in the CHUP-CDG in 2020 [Kaboré 2020].

Apart from some methodological differences between the studies, the difference in the frequency of congenital malformations between regions, countries and within the same country can be explained by several factors. 1) Genetic factors: Genetic variations within populations can influence the prevalence of congenital malformations. Certain ethnic groups or populations may be more predisposed to certain malformations because of their specific genetic heritage [Anthony 2005, Egbe 2015]. 2) Environmental factors: Environmental factors, such as exposure to toxins, teratogens, air or water pollution, can vary from country to

country, and even from region to region within a country. These environmental differences can contribute to variations in the frequency of congenital malformations. 3) Healthcare and access to medical services: Differences in healthcare systems and access to quality prenatal and postnatal care can play a significant role. Countries or regions with limited access to adequate healthcare may have a higher prevalence of congenital malformations due to late diagnosis or insufficient medical intervention. 4) Level of economic development: Developed countries may have more advanced medical resources and infrastructure, which can improve the detection and management of congenital malformations. In developing countries, on the other hand, resources may be more limited, which could lead to an underestimation of malformations. 5) Social-cultural factors : Cultural and social differences may also play a role in how congenital malformations are perceived, reported and treated. Certain malformations may be stigmatized or ignored in certain cultures, which can influence reported prevalence rates.

It is important to note that the frequency of congenital malformations is complex and multifactorial, and that these explanations are not exhaustive. Further research and epidemiological studies are needed to better understand the differences observed between and within countries.

Risk factors

The majority of mothers were between 20 and 35 years of age. This result is similar to that reported in Burkina Faso [Kaboré 2020] , Cameroon [Kamla 2017], and India [Madhura 2020] and can be explained by the higher fertility rate in this age group. We noted the presence of malformations in children born to young mothers under 20 years of age (11.24%). This result is superposable with that of Monzango *et al* [Monzango 2018], who found 18.9% of mothers aged between 18 and 21. The influence of maternal age on the occurrence of malformations depends on the type of malformation. Structural malformations are more

frequent in younger mothers (under 20), while chromosomal malformations are more prevalent in older mothers (over 35) [Nazer 2007, Tennant 2010].

The fathers of malformed neonates were around 40 years old on average. This result is close to that of Bénié *et al* [Bénié 2017]. In our society, husbands are generally older than their wives. This could explain this result. One study [Zhu 2005] found that the risk of extremity malformation for offspring was significantly increased by 37% in fathers aged 40-44, by 64% in fathers aged 45-49, and the risk of Down's syndrome multiplied by 4.5 in fathers aged 50 and over compared with those aged 20-29.

In this study, the proportion of malformed children was higher among housewives, in agreement with West Africans authors [Kaboré 2020, Segbedji 2022]. Among fathers, the majority of malformations were found among farmers, again in agreement with Kaboré *et al* [Kaboré 2020]. In reality, these results reflect the occupational distribution of men and women in our country [INSD]. Some studies have shown a significant link between parental occupation and the occurrence of congenital malformations. For example, mothers working in leather manufacturing and the textile industry had a high risk of giving birth to a malformed child [Bianchi 1997], while this risk was multiplied by 4 in the case of a mining or blue-collar father [Lubala 2012]. Other studies have not produced formal evidence of associations between paternal occupational exposure to solvents and neural tube defects and between maternal exposure to pesticides and orofacial clefts [Baldacci 2018].

In this study, the trend shows a progressive increase in malformations from primigravida to a peak in paucigravida, followed by a decrease in multigravida, and finally a minimal frequency in large multigravida. The possible interpretation of this trend is as follows: 1) As primigravida (24.7%) are in their first pregnancy, they generally have a lower risk of malformations, as risk factors related to previous pregnancies are absent. 2) The paucigravida

(34.8%) have already had a few pregnancies, but not as many as the multigravida. The increasing frequency of malformations can be attributed to the potential accumulation of risks linked to previous pregnancies, but it may also be due to other individual factors. 3) The curve curves downwards for multigravida (27%), meaning that the risk of malformations is lower than for paucigravida. This could be explained by the fact that multigravida women have already experienced pregnancies without serious malformations, which may indicate a certain genetic resistance or favorable lifestyle habits. Finally 4) The frequency of malformations is lowest among large multigravida women (13.5%). This group, which has experienced a large number of pregnancies, has undoubtedly adapted biologically, has a better understanding of prenatal care and has implemented strategies to reduce risks.

These explanations are hypotheses, and there could be other complex factors at play, such as the woman's age, genetics, environmental factors, access to healthcare and lifestyle habits, which influence the frequency of malformations in each group. To obtain a more precise explanation, in-depth studies and analysis of clinical data would be necessary.

There was a preponderance of malformed female neonates in this study. This result corroborates that reported by Mekonnen et al [Mekonnen D 2021] and Mekonen et al [Mekonen HK 2021]. In contrast, other authors found a higher frequency of male neonates [Ahuka 2015, Madhura 2020, Segbedji 2022, Nouhou 2022]. The study of the relationship between gender and malformations has shown a male representation in many registries, although some malformations (central nervous system, cleft palate) are more frequent in females, while others (urogenital, gastrointestinal) are the preserve of males [Lisi 2005].

The majority of neonates were full-term in this study, that is consistent with the findings of some authors [Shalaby 2020, El Ghanmi 2020, Segbedji 2022] but contradicts those of other authors [Daliri 2018, Madhura 2020, Mekonen HK 2021] who report more premature

neonates. It is thought that the type of malformation may influence the term of the pregnancy. In the case of a major or chromosomal malformation, the pregnancy may end in abortion or premature birth, whereas in the case of a minor malformation the pregnancy may progress to term.

In this study, most neonates had LBW. This finding is in agreement with the conclusions of authors such Daliri et al [Daliri 2018] but it contradicts Mekonen HK et al [Mekonene HK 2021]. This observation can be explained by the fact that many of the malformations, whether minor or structural, restricted fetal growth, resulting in the birth of LBW neonates.

Concluding with factors related to the mother and the newborn, study results concur that the sex of the newborn, premature birth, low birth weight, the mother's age, consanguineous marriages, multiple births, family history of congenital malformations and the risk of chronic diseases in the mother during pregnancy increase the risk of birth of children with congenital malformations [Daliri 2018].

Etiological factors

In this study, a proportion of 2.2% of malformed neonates from consanguineous marriages was noted, this figure being significantly lower than the rate of consanguinity of 8% reported in India [Padmanabhan 2020], 37.5% in Qatar [Al-Dewik 2023]. This difference is attributable to the impact of social-cultural practices specific to different populations. Consanguinity is thought to play a significant role in increasing the prevalence of genetic disorders, mainly autosomal recessive disorders [Ben-Omran 2019]. According to World Health Organization (WHO) [WHO 26/08/23] data, consanguinity contributes to an increase in the prevalence of genetically rare congenital malformations by doubling the risk of such malformations in children born to first cousins.

We have found highly febrile pathologies (malaria, pneumonia) in the last trimester of pregnancy in women who have given birth to malformed children. We have no evidence that this fever is involved in the occurrence of the malformations observed in the neonates of the women concerned. Fever is known to play a role in the occurrence of congenital malformations, especially in the first trimester of pregnancy [Nazer 2005].

In this study, we noted that 3.4% of pregnant women took medication during the third trimester without consulting a qualified health professional. These drugs were artemether-lumefantrine and amoxicillin. Artemether and lumefantrine are two antimalarial drugs that do not appear to be contraindicated for pregnant women. Similarly, amoxicillin, a member of the penicillin family of antibiotics, does not appear to present any teratogenic risk, whatever the term of pregnancy. However, it is important to note that the use of illicit drugs before or during pregnancy increases the risk of perinatal morbidity and congenital malformations, as demonstrated by a recent study [Lee 2023].

In this study, 7.9% of mothers claimed to have come into contact with pesticides during pregnancy, as part of their farming activities alongside their husbands, who were farmers in 24.7% of cases. Studies have shown that parental exposure to pesticides increases the risk of congenital malformations [Tennant 2010]. A rigorous national policy of controlling the marketing, monitoring the use, and even banning these proven carcinogens would be useful in preventing possible malformations.

Anatomical type and site of malformations

Osteoarticular malformations (38%), malformations of the eye, ear, face and neck (22.8%) and digestive malformations (21.6%) were the most common in this study. According to the WHO [WHO 26/08/23], the most common serious congenital disorders are congenital heart and neural tube defects and Down's syndrome. Data from recent studies in Africa, the Middle-

East and Asia show a difference in malformation frequencies depending on the organs and systems affected (Table 5).

Table 5: Frequency of some anatomical types and site of malformations in Africa, Middle-East and Asia

Author, country	**Anatomical type and location of malformations n (%)**				
	Circulatory system	Nervous system	Musculoskeletal system	Digestive system	Urogenital system
Our study	-	17 (9,9)	65 (38)	37 (21,6)	10 (5,8)
Kaboré e*t al* [Kaboré 2020] (n = 134) (Burkina Faso, 2020)	81 (37,8)	24 (11,2)	45 (21)	13 (6,1)	
Kamgaing *et al* [Kamgaing 2018] (n = 78) (Gabon, 2018)	-	43 (55,1)	20 (25,6)	31 (39,7)	
Bénié *et al* [Bénié 2021] (n = 103) (Ivory Coast, 2021)	-		44 (43)	38 (36,9)	17 (16,5)
Ajao *et al* [Ajao 2019] (n = 67) (Nigeria, 2019)	11 (16,5)	4 (6)	8 (12)	5 (7,5)	8 (12)
Cavaliere *et al* [Cavaliere 2021] (n = 143) (Mozambique, 2021)	14 (10)		45 (31)	-	-
Longombe *et al* [Longombe 2015] (n = 89) (DRC, 2015)		33 (30,8)	38 (35,5)	13 (12,1)	5 (4,7)
Mashako *et al* [Mashako 2017] (n = 52) (DRC, 2015)		7 (13,5)		25 (48)	3 (5,8)
Mekonen HK *et al* [Mekonen 2021] (n = 383) (Ethiopia, 2021)		263 (68,7)		11 (2,9)	12 (3,1)
Forci *et al* [Forci 2020] (n = 245) (Morocco, 2020)	(8)	(18)	(33)		(7,5)
El Awady *et al* [El Awady 2021] (n = 74) (Egypt, 2021)	24 (32,4)		14 (18,9)	7 (9,7)	4 (2,7)
Al-Dewik *et al* [Al-Dewik 2023] (n = 332) (Qatar, 2023)	117 (35)	20 (6)			39 (12)
Padmanabhan *et al* [Padmanabhan 2019]	31 (35,6)	9 (10,3)	23 (26,4)	8 (9,2)	12 (13,8)

(n = 87) (india, 2019)					
Sinha *et al* [Sinha 2022] (n = 124) (India, 2022)	15 (12,1)	26 (21)	48 (38,7)	24 (19,3)	7 (5,6)

In fact, every anatomical structure in the body is susceptible to malformation. The frequency with which different structures are found malformed varies considerably, depending on the physiological impact of the malformation during the prenatal period [Stevenson].

Outcome

The mortality rate associated with malformations is very high in this study, with more than half of malformed neonates dying. Other authors have reported more moderate mortality rates, such as Madhura *et al* [Madhura 2020] (8%), Bénié *et al* [Bénié 2021] (11%), Kaboré *et al* [Kaboré 2020] at 13.9%, and Sinha *et al* [Sinha 2022] at 30.5%. This result lies between the significantly higher rates observed in other African countries, such as Mozambique according to *Cavalière et al* [Cavaliere 2021] (50%) or the DRC according to Mashako *et al* [Mashako 2017] (59.6%). This variation in case-fatality rates could be explained by differences in malformation severity, levels of medical care, risk factors and etiology,

There is therefore a need for continued research, as these results underline the importance of monitoring congenital malformations and associated lethality, in order to identify long-term trends, assess the effectiveness of interventions and guide health policies aimed at improving maternal and child health.

Limits of study

Due to several limitations, the implications of the results of this study could be restricted. The descriptive, retrospective aspects and the fact that the study was conducted in a single center could lead to a reduction in both the quantity and precision of the data, thus limiting the

generalization of the results. It should also be noted that the underlying causes of malformations were not explored in depth in this study.

Conclusion

This study has shown that malformations are common in our hospital. The diversity of malformation types highlights the complexity and variability of structural or functional problems encountered in neonates. The impact on different groups (sex, birth order) of neonates can give indications of some potential risk factors. The higher frequency of osteoarticular, eye, ear, face/neck and digestive malformations suggests specific areas of pediatric health concern in this population. The very high case-fatality rate of congenital malformations underscores the importance of medical care and interventions to improve outcomes.

List of abbreviations

ANCs: Antenatal care visits

CHUP-CDG: Centre hospitalier universitaire Chrales de Gaulle

DRC: Democratic Republic of Congo

LBW: Low birth weight

NU: Neonatal unit

WHO: World health organization

Ethical Approval and consent

We obtained an institutional approval letter from Centre hospitalier universitaire Charles de Gaulle prior to the commencement of the study with approval number 2022-1540/MSHP/SG/CHUP-CDG/DRH/SRF, July 22, 2022. The necessity to obtain written consent was waived because of the retrospective nature of this study. In addition, the data recorded was anonymized. Above all, this study was entirely conducted as per the Declaration of Helsinki ethical principles for medical research on human subjects.

References

1. WHO. Birth defects. Malformations congénitales. [Internet] https://apps.who.int/gb/ebwha/pdf_files/WHA63/A63_10-en.pdf. (September 16, 2023). French
2. WHO. Birth defects surveillance: a manual for programme managers, second edition. Geneva: World Health Organization; 2020. Licence: CC BY-NC-SA 3.0 IGO.
3. WHO. Congenital disorders. Key facts. https://www.who.int/news-room/fact-sheets/detail/birth-defects. (August 26, 2023).
4. Loane M, Given JE, Tan J, Reid A, Akhmedzhanova D, et al. Linking a European cohort of children born with congenital anomalies to vital statistics and mortality records: A EUROlinkCAT study. PLoS ONE. 2021;16(8):e0256535. https://doi.org/10.1371/journal.pone.0256535
5. Bénié AC, Lohourou GF, Akobé AJR, Traoré I, Kpangni AJB, et al. Les malformations congénitales cliniquement visibles. Rev Int Sci Med Abj. 2021;23(1):17-23. French
6. Hama AN, Hamani I, Boureima D, Hamadou M, Ousmane A, et al. Malformations congénitales au Centre de Santé Mère et Enfant de Tahoua. Health Sci Dis. 2022;23(3):95-8. French
7. Ouermi AS, Dao L, Lengane NI, Kpadonou DM, Guira I, et al. Pronostic immédiat des malformations congénitales reconnaissables à la naissance au Centre hospitalier universitaire de Ouahigouya (Burkina Faso). J Rech Univ Lomé. 2021;23(1):323-31. French
8. Kaboré A, Nagalo K, Compaoré K, Bélemviré B, Bama A, et al. Malformations congénitales : étude descriptive hospitalière à Ouagadougou (Burkina Faso). Health Sci Dis. 2020;21(3):90-9. French

9. Ramatou IB, Mahamoud Omid AA, Mamane Kabirou DMK, Oumarou H, Habibou A. Epidémiologie des malformations congénitales à l'Hôpital National Amirou Boubacar Diallo de Niamey. Med Afr Noire. 2022;69(6):375-80. French
10. Mashako RM, Nsibu NC, Nkinamubanzi M, Bitwe MR, Kajibwami E, et al. Les malformations congénitales à l'Est de la République Démocratique du Congo : défis et perspectives. Int J Innovation Sci Res. 2017;33(2):256-61. French
11. Monzango SGL, Kanguidia MJ, Mbanzulu PN. Malformations congénitales visibles à la maternité des cliniques universitaires de Kinshasa. Kis Med.2018;8(1):319-323. French
12. El Awady H, Al Gameel A, Ragab TM, Hassan N. Congenital malformations in neonates in Fayoum Governorate, Egypt. East Mediterr Health J. 2021;27(8):790-7. https://doi.org/10.26719/emhj.21.033.
13. Madhura F, Pratibha S. Incidence of congenital anomalies in neonates – a tertiary care experience. Int J Pediatr Res. 2020;7(2):97-103. https://doi.org/10.17511/ijpr.2020.i02.09.
14. Anthony S, Kateman H, Brand R, Den Ouden AL, Dorrepaal CA, et al. Ethnic differences in congenital malformations in the Netherlands: analyses of a 5-year birth cohort. Paediatr Perinat Epidemiol. 2005;19(2):135–44.
15. Egbe AC. Birth defects in the newborn population: race and ethnicity. Pediatr Neonatol. 2015;56(3):183–8. https://doi.org/10.1016/j.pedneo.2014.03.010.
16. Kamla JI, Kamgaing N, Nguifo FEJ, Fondop J, Billong S, et al. VP. Épidémiologie des malformations congénitales visibles à la naissance à Yaoundé. Health Sci Dis. 2017;18(4):53-62.

17. Nazer HJ, Cifuentes OL, Aguila RA, Ureta LP, et al. Edad materna y malformaciones congénitas: un registro de 35 años. 1970-2005. Rev Med Chil. 2005;135(11):1463-9. http://dx.doi.org/10.4067/S0034-98872007001100014. Spanish

18. Tennant P, Raza F, Bythell M, Rankin J. Maternal age and the risk of structural congenital malformations. Arch Dis Child - Fetal Neonatal Ed. 2010;95(Suppl 1):Fa4-Fa4.http://dx.doi.org/10.1136/adc.2010.192310.1.2.

19. Zhu JL, Madsen KM, Vestergaard M, Olesen AV, Basso O, et al. Paternal age and congenital malformations. Hum Reprod. 2005;20(11):3173-7. https://doi.org/10.1093/humrep/dei186.

20. Segbedji KAR, Tchagbele OB, Takassi OE, Kombieni K, Talboussouma SM, et al. Epidémiologie des anomalies morphologiques néonatales en milieu hospitalier à Kara (Nord Togo) de 2014 à 2019. Eur Sci J. 2022;18(3):1. French https://doi.org/10.19044/esj.2022.v18n3p1.

21. Institut National de la Statistique et de la Démographie (INSD). 5th General Population and Housing Census of Burkina Faso. 2nd edition (August 2022). French https://www.insd.bf/sites/default/files/2023-08/INSD_Rapport_SYNTHESE%20DES%20RESULTATS%20DEFINITIFS_1.pdf (September 16, 2023).

22. Bianchi F, Cianciulli D, Pierini A, Costantini AS. Congenital malformations and maternal occupation: a registry based case-control study. Occup Environ Med. 1997;54(4):223-8.

23. Lubala KT, Kalenga MK. Facteurs de risque associés aux malformations congénitales apparentes à Lubumbashi, une ville minière de la RDC, étude cas- témoins. Pan Afr Med J. 2012;Revision 1082-1. French

24. Baldacci S, Gorini F, Santoro M, Pierini A, Minichilli F, et al. Environmental and individual exposure and the risk of congenital anomalies: A review of recent epidemiological evidence. Epidemiol Prev. 2018;42(3-4 suppl1):1-34. https://doi.org/10.19191/EP18.3-4.S1.P001.057.

25. Mekonnen D, Taye M and Worku W. Congenital anomalies among newborn babies in Felege- Hiwot Comprehensive Specialized Referral Hospital, Bahir Dar, Ethiopia. Sci Rep. 2021;11:11027. https://doi.org/10.1038/s41598-021-90387-0.

26. Mekonen HK, Berhe Y, Berihu BA, Teka H, Hadgu A, et al. A silent epidemic of major congenital malformations in Tigray, northern Ethiopia: hospital- based study. Sci Rep. 2021;11:21035. https://doi.org/10.1038/s41598-021-00240-7.

27. Longombe AO, Bahati K, Fefe I. Panorama of visible birth defects in Eastern Democratic Republic of Congo (DRC) during the war period. Ind J Appl Res. 2015;5(8):118-20.

28. Lisi A, Botto LD, Rittler M, Castilla E, Bianchi F, et al. Sex and congenital malformations, an international perspective. Am J Med Genet A. 2005;134(1):49-57. https://doi.org/10.1002/ajmg.a.30514.

29. Elghanmi A, Razine R, Jou M, Berrada R. Congenital malformations among newborns in Morocco: A retrospective study. Pediatr Rep. 2020;12:7405. https://doi.org/10.4081/pr.2020.7405.

30. Daliri S, Safarpour H, Bazyar J, Sayehmiri K, Karimi A, et al. The relationship between some neonatal and maternal factors during pregnancy with the prevalence of congenital malformations in Iran: a systematic review and meta-analysis, J Matern Fetal Neonatal Med. 2018;32(21):3666-3674. https://doi.org/10.1080/14767058.2018.1465917.

31. Padmanabhan R, Venkatasubramanian R, Heber A. Prevalence and pattern of congenital malformations among neonates in a Medical college hospital - A retrospective study. Int J Sci Stud. 2019;6(12):28-31.

32. Al- Dewik N, Samara M, Younes S, Al- jurf R, Nasrallah G, et al. Prevalence, predictors, and outcomes of major congenital anomalies: A population- based register study. Sci Rep. 2023;13:2198. https://doi.org/10.1038/s41598-023-27935-3.

33. Ben-Omran T, Al Ghanim K, Yavarna T, El Akoum M, Samara M, et al. Effects of consanguinity in a cohort of subjects with certain genetic disorders in Qatar. Mol Genet Genomic Med. 2020;8:e1051. https://doi.org/10.1002/mgg3.1051.

34. Lee CTC, Chen VCH, Lee JKW, Wu SI, Cheng G, et al. Substance use before or during pregnancy and the risk of child mortality, perinatal morbidities and congenital anomalies. Epidemiol Psychiatr Sci. 2023;32(e43):1-10. https://doi.org/10.1017/S2045796023000549.

35. Kamgaing EK, Rogombe SM, Maniaga RK, Tsague G, Minko JI, et al. Congenital malformations seen in Libreville, management and evolution. EC Pediatr. 2018;7(5):422-34.

36. Ajao AE, Adeoye IA. Prevalence, risk factors of congenital anomalies among neonatal admissions in OGBOMOSO, Nigeria. BMCPediatr. 2019;19:88. https://doi.org/10.1186/s12887-019-1471-1.

37. Cavaliere E, Trevisanuto D, Da Dalt L, Putoto G, Pizzol D, et al. Congenital malformations in neonates admitted to a neonatal intensive care unit in a low-resource setting. J Matern Fetal Neonatal Med. 2022;35(25):6363-7. https://doi.org/10.1080/14767058.2021.1912003.

38. Forci K, Alami MH, Bouaiti E, Slaoui M, Mdaghri Alaoui1A, et al. Prevalence of congenital malformations at the “les Orangers” maternity and reproductive health

Hospital of Rabat: descriptive study of 470 anomalies. BMCPediatr. 2020;20:272. https://doi.org/10.1186/s12887-020-02179-6.

39. Sinha A, Tripathi S, Nigam N, Kumar M, Singh SN. Profile of neonates born with congenital birth defects in a tertiary care hospital of North India: An observational study. Clin Epidemiol Glob Health. 2022;14:100999. https://doi.org/10.1016/j.cegh.2022.100999.

40. Stevenson RE, editor. Human malformations and related anomalies. 2nd ed. Oxford; New York: Oxford University Press (Oxford monographs on medical genetics; 2006.