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Macroscopic Congenital Malformations at the Institute of Nutrition and Child Health (INSE)

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Abstract

Introduction: A congenital malformation is defined as a morphological abnormality of an organ or body region resulting from an abnormal developmental process during the formation of the embryo or fetus. Depending on their type, location and size, malformations can cause functional, psychological and aesthetic defects. The aim of this study is to document the frequency of congenital malformations, describe the characteristics of malformed newborns and their biological mothers, and identify the different types of malformations presented by newborns at the INSE. **Methods:** Descriptive cross-sectional study of clinically visible malformed newborns. It was carried out from January 1, 2021 to January 1, 2022 at the INSE neonatology unit. Epi info version 3.1 software was used for data entry and analysis. **Results:** Of a total of 2332 neonates hospitalized during the study period, 81 (3.5%) cases had at least one clinically visible congenital malformation. Nearly 84% had an age ≤ 7 days at the time of admission. The male sex was most concerned (60.5%). Newborns referred by a health facility accounted for 84%. Malformations of the digestive system accounted for 30.9% of cases, followed by those of the limbs (19.8%) and poly malformative syndrome (19.8%). **Conclusion:** This study shows that congenital malformations exist and are frequent in Guinea. Our results could therefore be the starting point for the future establishment of a national register of congenital malformations.

Keywords

Congenital Malformation, Institute, Nutrition, Child Heath

1. Introduction

Congenital malformations are morphological and functional anomalies that may or may not be visible at birth. They are one of the main causes of neonatal morbidity, mortality and disability, after prematurity, asphyxia and infection [1]. They pose a public health problem worldwide. According to the World Health Organization, around three million children are born each year with major malformations, and these are responsible for 495.000 deaths [2]. In developed countries, congenital malformations are responsible for 20% to 25% of total perinatal mortality, making them the leading cause of perinatal mortality in France [3]. In Africa, frequencies vary from one country to another (4% in Morocco in 2013, 9 cases per 1000 births in Cameroon in 2017 and 4.9% in Côte d'Ivoire) [2] [4]. Aetiologies are multifactorial, dominated by genetic and environmental factors, knowledge of which could help reduce their incidence and consequently neonatal and infant mortality rates [2]. Certain maternal infectious diseases, such as syphilis and rubella, are a major cause of congenital malformations in low- and middle-income countries. Maternal illnesses such as diabetes mellitus, certain medical conditions such as iodine or folic acid deficiency, and exposure to medicines and recreational drugs, including alcohol and tobacco, to certain environmental chemicals and to high doses of radiation are other factors at the origin of congenital malformations [5]. Congenital malformations are present at birth, even if they are not diagnosed until afterwards. Depending on their type, location and size, malformations can cause functional, psychological and aesthetic defects [6]. Although they are not a priority in the health policies of developing countries, they often pose diagnostic and therapeutic problems, and are associated with high mortality [7]. In African society, they are considered a real tragedy, given the mystico-religious considerations surrounding them on the one hand, and the real burden they place on families on the other [1]. The rate of detection of congenital disorders during the first trimester of pregnancy by biochemical analyses is better if these analyses are performed in association with ultrasound to measure nuchal translucency and other ultrasound assessments. Second-trimester ultrasound is useful for detecting major structural anomalies [3].

In Guinea, despite the progress made in pregnancy monitoring, there are still some concerns regarding the antenatal diagnosis of malformations and the urgent management of some of them. It is in this context that we initiated this work to document the frequency of congenital malformations, to describe the characteristics of malformed newborns as well as those of their biological mothers, and to identify the different types of malformations presented by newborns at the INSE.

2. Method

2.1. Study Setting

Our study was carried out in the neonatology department of the Institut de Nutrition et de Santé de l'Enfant (INSE). It is the only public reference institute at national level since 1989, receiving newborns from all regions of the country.

2.2. Type and Period of Study

This is a descriptive cross-sectional study lasting fourteen months (14), from 01 January 2022 to 03 March 2023.

2.3. Target Population

Our target population consisted of all newborns admitted to the department during the study period.

2.4. Study Population

All newborns with one or more congenital malformations received in the department during the study period.

2.5. Selection Criteria

2.5.1. Inclusion Criteria

The following are included in our study

All newborns with one or more congenital malformations, extrinsic and/or intrinsic, received in the department during the study period and whose parents have agreed to participate in the study.

2.5.2. Non-Inclusion Criteria

All newborns with poorly completed charts and those whose parents did not participate in the study were not included.

2.6. Data Collection

For data collection, we used the following media: hospitalization records, hospitalization registers and a pre-established survey form on the Kobocollect application.

2.7. Sampling and Sample Size

We carried out a census of all patients suffering from a morphological or functional anomaly, visible or not, present at birth during the study period.

At the end of this census we obtained a sample size of 81 people.

2.8. Study Variables

In our study, we defined congenital malformations as all morphological and functional anomalies that may or may not be visible at birth.

Our variables were quantitative and qualitative

Age, Gestational age, Weight, Birth weight, Height, Head circumference, CPN, Sex, Maternal data, Maternal age, Gestité, Parity, Provenance, Maternal education.

2.9. Data Analysis

Our data are collected using a survey form, recorded in a database using Kobocollect software, then downloaded in Excel files and analyzed using SPSS soft-

ware version 21.

Proportions are calculated for qualitative variables. Quantitative variables are expressed as Median.

2.10. Ethical Considerations

We sought and obtained permission from the INSE General Manager before proceeding with the study. Free and informed oral consent was obtained from the parents of the neonates prior to their inclusion in the study. Confidentiality and anonymity were respected throughout the procedure.

3. Results

A total of 2332 newborns were hospitalized during the study period. Of these, 81 had at least one or more clinically visible congenital malformations, representing a prevalence of 3.5%. Mono malformations accounted for 82.7%, versus 17.3% for polymalformed newborns.

Nearly 84% (83.9%) were aged ≤ 7 days at the time of admission. Males accounted for 60.5% of cases versus 39.5% for females. Newborns referred by a health facility accounted for 84%. Nearly 35% (34.6%) of malformed newborns had a gestational age < 37 SA versus 65.4% for those whose gestational age was > 37 SA (Table 1).

The average age of the mothers of malformed newborns was 21. Of these, 43.2% were aged between 26 and 34. Multiparous and multi gestational women accounted for 54.3% and 60.5% of cases respectively. Women who had undergone 1 or 2 ANC procedures accounted for 80.2%. Nearly 93% (92.6%) had given birth in a health facility. Eutocic delivery accounted for 69.1% of cases (Table 2).

Of the 81 types of malformation, spina bifida was the most common (9 cases, or 11.1% of all malformations), followed by omphalocele (8.6%) and trisomy 21 (8.6%). In terms of distribution by site, malformations of the digestive system and limbs accounted for the majority of cases, at 23.4% each. Eye anomalies accounted for 3.6% (Table 3).

The proportion of all deaths attributable to malformations was 37%, or 30/81. Of these, 40% were attributable to malformations of the digestive system (Figure 1).

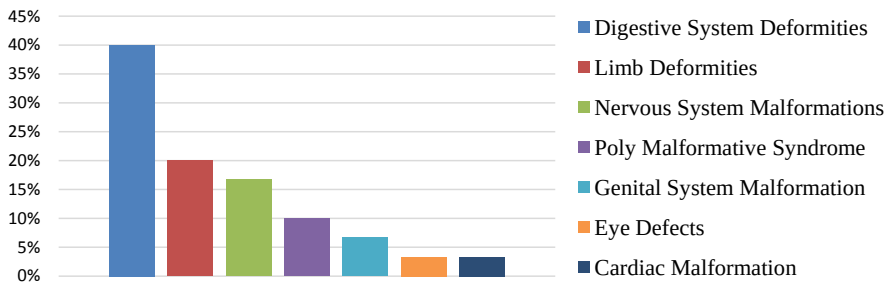

Figure 1. Distribution of deaths by type of malformation, by system.

Table 1. General characteristics of malformed newborns.

Age in days	Number	%
0 - 7	68	83.9
8 - 15	6	7.4
16 - 23	3	3.7
24 and more	4	4.9
Provenance		
CMC/HP/CS	34	42.0
Private clinic	26	32.1
Home	13	16.0
Ignace Deen	6	7.4
Donka	2	2.5
Mode of admission		
Referral	68	84.0
Spontaneous	13	16.0
Gender		
Female	32	39.5
	49	60.5
Birth weight in grams		
<2500 g	18	22.2
≥2500 g	63	77.8
Gestational age		
<37 SA	28	34.6
>37 SA	53	65.4

Table 2. Sociodemographic and obstetrical characteristics of mothers.

Characteristics	Number	%
Age in years		
<18	4	4.9
19 - 25	28	34.6
26 - 34	35	43.2
35 And more	14	17.3
Place of delivery		
CMC/HP/CS	35	43.2
Private clinic	29	35.8
Home	11	13.6
Ignace Deen	6	7.4
Parity		
Multipare	44	54.3

Continued

Pauci pare	14	17.3
Primipare	23	28.4
Gestite		
Multigeste	49	60.5
Pauci geste	12	14.8
Primigeste	20	24.7
Mode of delivery		
Eutocique	56	69.1
Dystocic	25	30.9
CPN		
1 - 2	65	80.2
3 - 4	13	16.1
≥5	3	3.7

Table3. Distribution of newborns by anatomical type and site of malformation.

Headquarters	Type of malformation	(N = 81)	%
Nervous system	Spina bifida	9	11.11
	Hydrocephalus	4	4.94
	Encephalocele	3	3.70
	Anencephaly	1	1.23
	Craniostenosis	1	1.23
		N = 18	**22.22**
Eye anomaly	Anophthalmos	1	1.23
	Congenital ectropion	1	1.23
	Microphthalmia	1	1.23
		N = 03	**3.70**
Digestive system	Omphalocele	7	8.64
	Cleft lip with or without cleft palate	6	7.41
	Anorectal malformation	2	2.47
	Oesophageal atresia	1	1.23
	Intestinal atresia	1	1.23
	Laparoscisis	1	1.23
	Cystic lymphangioma	1	1.23
		N = 19	**23.44**
Genital system	Sexual ambiguity	2	2.47
	Hypospadias	2	2.47
	Micro penis	2	2.47
	Phocomelia	1	1.23

Continued

	Vaginal prolapse	1	1.23
		N = 08	**9.90**
Members	Varus clubfoot	6	7.41
	Knee recurvatum	3	3.70
	Polydactyly	3	3.70
	Ectrodactyly	2	2.47
	Pieds bot talus	2	2.47
	Hexadactyly	1	1.23
	Pied plat light	1	1.23
	Shortening of the lower limbs	1	1.23
		N = 19	**23.44**
Polymaformative syndrome	Trisomy 21 + cleft palate	7	8.64
	Spina bifida + varus clubfoot	2	2.47
	Hydrocephalus + omphalocele	1	1.23
	Sexual ambiguity + varus clubfoot	1	1.23
	Omphalocele + superior velar cleft	1	1,23
	Omphalocele + umbilical hernia	2	2.47
		N = 14	**17.30**

4. Discussion

The frequency of congenital malformations among all pregnancies is unknown, as many conceptions result in early spontaneous abortions. Congenital malformations in stillborn and living infants therefore constitute only a fraction of all congenital malformations [3]. This is why we speak of the prevalence of birth defects, which today represents 3% to 4% of live births [3]. In this study, we recorded 81 cases of congenital malformations out of 2332 admissions, representing a hospital prevalence of 3.5%. This rate is lower than the 4% found by Sabari N *et al.* [2] in Morocco, but lower than those recorded by Nguefack CT [8] *et al.*, in Douala (1.6%). In our series, 83.9% of newborns with at least one congenital anomaly were admitted to the neonatal unit during the first week of life. This could be explained by the fact that 3/4 of cases were the result of assisted delivery, which enables clinically visible malformations to be detected early and referred to the INSE (referral structure). Physical examination of all newborns by qualified primary care practitioners is feasible in most health systems, and provides an opportunity to identify and refer many cases of congenital malformations [5]. Screening should identify opportunities to help the newborn and his or her family as widely as possible. In other words, screening should identify actionable conditions, including those that may benefit from treatment [9]. The family of a newborn who has been diagnosed should receive psychological, social and economic support from the public health organization [9]. Unfortunately, this type of support does not exist in Guinea's health establishments, as once a

child has been diagnosed with a malformation, the burden falls on the parents. A number of steps need to be taken at country level to encourage the development of prevention and care services for congenital malformations. Prevention requires the integration of basic public health approaches into health systems, particularly at the level of maternal and child health services. Many of the proposed services and interventions are already within the reach of low- and middle-income countries, while others may be added as needs and resources allow [5]. In the course of this study, we observed that boys were more affected by congenital anomalies than girls (**Table 1**). It has long been known that the overall prevalence of congenital anomalies, and that of most of these malformations considered separately, is higher in male subjects [10]. Several factors may have played a role in this increase, including higher rates of certain male-specific anomalies (hypospadias) or anomalies more common in male subjects, such as Down's syndrome and renal agenesis [11]. With regard to gestational age, nearly 35% (34.6%) of malformed neonates had a gestational age < 37 SA, compared with 65.4% of those whose gestational age was >37 SA. Our observations are similar to those of Kaboré A *et al.*, who found that malformed newborns were born at term in the majority of cases [12]. On the other hand, other authors report a higher frequency of prematurity among malformed babies [13] [14]. In our series, the average age of the mothers of malformed babies was 21 years. Of these, 43.2% were between 26 and 34 years of age. Our results corroborate those of other authors who have observed that congenital malformations are more frequent in women in the 20 - 30 age bracket [13] [15]. But this assertion is not shared by other studies, which found no association between congenital malformations and maternal age [14] [16]. In terms of obstetrical characteristics, the largest proportion of malformed newborns were born to multiparous or multigestational mothers (**Table 2**). The same observation was made by Coulibaly F G *et al.* [17], who reported that congenital malformations were more frequent in multiparous mothers (12%). In our series, 80.2% of women who had undergone 1 to 2 ANC had given birth to a malformed newborn. Pregnant women sometimes start ANC rather late, and therefore do not receive early enough the care that would prevent certain malformations, such as neural tube defects due to failure to take folic acid during pregnancy [15]. What's more, few women have the financial means to undergo prenatal examinations, in particular fetal morphological ultrasound, which would enable early detection of malformations and, in the event of severity, suggest medical termination of the pregnancy [12]. Concerning the distribution of congenital malformations by type, spina bifida is the malformation with the highest frequency (9 cases or 11.11% of all malformations). This rate is higher than that recorded by Kaboré *et al.* [12] in Burkina Fasso (4.2%), but lower than that of Gnassingbé *et al.*, who reported that spina bifida accounted for 77.59% of the 58 cases of AFTN followed up at Lomé University Hospital [18]. The high prevalence of spina bifida in our study could be explained by the fact that (80.2%) of women had only performed 1 - 2 PNCs,

and at a late stage. Yet it is during these ANC that providers counsel and prescribe folic acid to pregnant women. Folate deficiency is the most firmly established risk factor for isolated neural tube defects. Inadequate folic acid intake from all sources (naturally folate-rich foods, folic acid-enriched foods and folic acid-containing supplements) remains an important modifiable risk factor worldwide [11]. For several authors, the mothers of children with Spina bifida had not received folic acid supplementation prior to pregnancy, nor during the first trimester of pregnancy [19]. Bannick *et al.* in Uganda concluded that the limited intake of folic acid was due to a lack of information and training for women, and especially health workers, on the importance of early folic acid intake [20]. In this study, the proportion of all deaths attributable to congenital anomalies was 37%, or 30/81. In comparison with other studies, our result is significantly higher than that of Rafi *et al.* [21], who reported 87 deaths in 645 malformed newborns, or 13.8%, and that of Shamim *et al.* [22], who observed five deaths in 57 malformed newborns, or 8.77%. On the other hand, close to the 38.2% reported by Abdi-Rad *et al.* [23]. Among the causes of death, 40% were attributable to congenital digestive malformation. Our rate is lower than that recorded (45.5%) by Engbang *et al.* in Douala [24]. The multicentre nature of this study could explain this high mortality rate. The decline in infant deaths from all congenital anomalies is attributable to a variety of factors, including: increased access to and use of prenatal screening; improved antenatal care; termination of pregnancies with severe anomalies; mandatory folic acid fortification of certain foods; and lifestyle changes, such as smoking cessation during pregnancy and increased use of prenatal vitamins [11] [25].

5. Study Strengths and Limitations

This study is the first to examine and document the frequency of congenital malformations at the Institut de Nutrition et Santé de l'Enfant, to describe the characteristics of malformed newborns and their biological mothers, and to identify the different types of malformations presented by newborns. As a limitation, this study was only carried out at the Institut de Nutrition et Santé de l'Enfant, whereas there are other units in the regions and in the private sector that were not included in this study.

6. Conclusions

This study shows that congenital malformations exist and are frequent in Guinea.

Our results could therefore be the starting point for the future establishment of a national register of congenital malformations, which would play a sentinel role in our context and thus enable us to envisage a national strategy for the prevention of these malformations. With the right training, primary care practitioners can offer basic care to children with congenital malformations: recognizing them, diagnosing common problems and determining related disabilities.

Implications for Research and Practice

The results of this study will contribute to a better understanding of congenital malformations. The results of this study will help guide actions to improve the health of these children. They will also enable us to plan other, larger studies that will provide more important data. Practical training for healthcare personnel working in maternity and neonatology departments on the orientation and management of malformed newborns would be necessary.

Authors' Contributions

Study design: BMA, BA, CSH.
Data collection: BMA, DS, DKP.
Drawing up the questionnaire: BMA, DKP.
Data analysis: BMA, DMS, KO.
Initial drafting of manuscript: BMA, BA, CSH, DFB, DMA, DMM.
Manuscript revision: TAO, DIS.
The authors have read and approved the final manuscript.

Availability of Data and Materials

Data supporting the results of this study are available from [Bangoura M'mah Aminata], but restrictions apply to the availability of these data and are therefore not publicly accessible, as our research group is working on further analyses using the same data which will then be submitted for publication. However, these data are available on reasonable request from the corresponding author [Bangoura M'mah Aminata].

Consent for Publication

Not applicable.

Conflicts of Interest

The authors declare that there is no competing interest.

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Inquiry Form

A) IDENTIFICATION OF THE NEW-BORN

File No. ..... Dossier No…… Type…….

Sex: O Male O Female

Number of days lived: .....

Date of birth: ...../...../.....

Term of pregnancy.....

Date of admission: ...../...../.....

Maternal age: .....

Mother's level of education:

O Primary; O Secondary; O Higher education/vocational; O Koranic; O No schooling;

OMunicipality: O Dixinn; O Kaloum; O Ratoma; O Matam; O Matoto; Coyah; O Dubréka; O Other:.....

Provenance: O Ignace Deen; O CMC Matoto; O CMC Matam; O Donka; O CMC Kaloum; O CMC Ratoma; O CMC Dixinn; O Private clinic; O Regional/Prefectural Hospital

Other: .....

.....

Maternal history: O Hypertension: O Yes O No; Diabetes: O Yes O No

Urinary tract infection: O Yes O No; Epilepsy: O Yes O No; Malaria: O Yes O No; Fetid leucorrhoea: O Yes O No; Eclampsia: O Yes O No

Serology: O SRV; O Rubella; O Toxoplasmosis; Other serologies: .....

.....

Ecography: O Yes O No

If Yes Result malformation: .....……

Notion of taking medication: O Yes O No

Drugs taken: O FAF

Other drugs taken: .....…….

Vices: O Oui O Non

O Alcool; O Tabac; O Drogue

Other vices: .....

Anamnèse

Gender: .....; Parity: .....; Abortion: .....; Stillbirths: .....; Deaths: .....

Antenatal consultation (ANC): O Yes O No

Number of ANC 1st trimester: .....

Number of ANC 2nd trimester: .....

Number of ANC 3rd trimester: .....

Mother's blood group

0 A+; O A−; O B+; O B−; O AB+; O AB−; O 0+; O 0−

Examens paracliniques du bébé

Examens réalisés: O oui O Non

Types d'examens réalisés

☐ Blood glucose; ☐ NFS; ☐ Echography; ☐ 9. ETF; ☐ GS/RH; ☐ CRP; ☐ GE(dp); ☐ Bilirubin; ☐ Creat

If other, please specify: .....

Blood glucose result: .....

Baby's GS/RH result: 0 A+; O A−; O B+; O B−; O AB+; O AB−; O 0+; O 0−

If CRP, Result: .....

If GE(dp) Results: .....

If bilirubinemia results: .....

If Créat Result: .....

If CBC, white blood cell result: .....

If ETF Result, ETF: .....

If ultrasound of the baby, results: .....

B) TYPES OF MALFORMATION

NERVOUS SYSTEM:

Nervous System Defects

O Anencephaly; O Encephalocele; Other; O Myelomeningocele; O Hydrocephaly; O Microcephaly; O Holoprosencephaly

If Other, specify: .....

EYES AND EARS:

Eye and/or ear deformities

O Anophthalmia; O Microphthalmia; O Micro Anotitis; O Other

If Other, please specify:

CLEFT LIP AND PALATE

O Orofacial cleft malformations; O Cleft lip with or without; O Cleft palate; O Macroglossia; O Choanal atresia

If other, please specify: .....

Digestive system: O omphalocele; O prune belly syndrome; O Digestive system malformations; O Anorectal malformation (MAR); O laparoschisis; O bladder extrophy; O oesophageal atresia;

If other, please specify: .....

GENITAL SYSTEM

O Sexual ambiguity; O Cryptorchidism; O Hypospadis; O Epispadias; O Ambiguity

If other, please specify: .....

.....

Membres

O Varus clubfoot; O Knee recurvatum; O Syndactyly

If other, please specify: .....

CARDIAC MALFORMATION

Types of cardiac malformation: .....

Associated diagnoses: O Perinatal asphyxia; O IMF; O INNP; O INNT; O Prematurity;

If other, specify: .....

Treatment of associated diseases

O Ampi; O Genta O; O Novatax;
If other, please specify: .....
ISSUE: O Transferred; o Died; o Improved
Date of discharge: ...../...../.....
Length of hospital stay: .....