

# Congenital Malformations in Neonate in Dakar: A Prospective Study of Clinical and Prognostic Profiles in a Referral Hospital

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## Abstract

**Introduction:** Congenital malformations (CM) are a major cause of neonatal mortality in developing countries. This study aims to describe the epidemiological, clinical, and evolutionary profile of CM in a tertiary referral center in Senegal. **Patients and Methods:** This is a prospective descriptive study conducted over eight months (January-August 2020) in the neonatal unit of the Albert Royer National Children's Hospital (CHNEAR) in Dakar. Newborns (0 - 2 months) with visible or clinically diagnosed malformations were included. **Results:** Out of 437 admissions, 80 newborns had a CM, representing a hospital prevalence of 18.3%. The sex ratio was 1.2. Regarding anthropometry, 20% had low birth weight (<2 kg) and 27% presented with intrauterine growth restriction (IUGR). Parental consanguinity was high (40%), mainly of the second degree (62.5%). The most common malformations involved the face and neck (27.2%), followed by osteoarticular (15.6%) and cardiovascular (15.6%) systems. Antenatal diagnosis was made in only 9% of cases, while 40% of mothers had no obstetric ultrasound. The in-hospital mortality rate was 55%. **Conclusion:** The high prevalence and mortality rate of CM at CHNEAR, coupled with poor prenatal screening, highlight the urgent need to improve morphological ultrasound training and surgical and genetic facilities in Senegal.

## Keywords

Congenital Malformations, Neonatology, Low Birth Weight, Consanguinity, Senegal

## 1. Introduction

Congenital malformations (CM) are defined by the World Health Organization (WHO) as morphological or functional abnormalities present at birth [1] [2]. They constitute a major public health problem, affecting approximately 6% of births worldwide and representing a significant cause of neonatal mortality [1]. Developed countries have effective registries and surveillance systems in place to enable prevention and early management [3]. Furthermore, variations in prevalence according to gender are observed worldwide, with certain anomalies often being more prevalent in males [4]. The situation remains a cause for concern in developing countries [5]. In sub-Saharan Africa, the actual prevalence is often underestimated due to the lack of national registries [6], inadequate prenatal diagnosis, and unreported early deaths. In Senegal, few prospective studies have addressed this issue comprehensively [7]. Most of the available data are retrospective or focused on specific pathologies. The Albert Royer National Children's Hospital (CHNEAR) in Dakar, a level 3 facility, receives a high concentration of complex pathologies. The objective of this study was to establish the epidemiological, clinical, and prognostic profile of congenital malformations in the CHNEAR neonatal unit in order to identify risk factors and challenges in care.

## 2. Patients and Methods

**Study setting and type:** This is a prospective descriptive study conducted over eight months period from January 1 to August 31, 2020, in the neonatal and pediatric intensive care unit at CHNEAR in Dakar. Patients were followed from admission until discharge (death or home return) to evaluate hospital outcomes.

**Study population:** The target population included all newborns aged 0 to 2 months admitted to the department during the study period. The inclusion criteria were the presence of a clinically detectable or imaging-confirmed congenital malformation, with informed consent from the parents. Incomplete records were excluded.

**Data collection:** Data were collected through a comprehensive clinical examination, a review of medical records and maternal health records, and the results of paraclinical tests (imaging, biology). The variables studied included the socio-demographic characteristics of the parents (age, consanguinity, socioeconomic status), obstetric history, types of malformations (classified by system), therapeutic management, and hospital progress.

**Ethical considerations and analysis:** Informed consent was obtained from parents in all cases. Data were analyzed using SPSS version 12.0 software.

## 3. Results

**General epidemiological data:** During the study period, 437 newborns were admitted, 80 of whom had one or more congenital malformations, representing a hospital prevalence of 18.3%. The sex ratio (M/F) was 1.2. The average age at admission was 16 days (range: 0 - 58 days). Most patients (42.5%) came directly-

ferred to their homes, while the others were referred by healthcare facilities.

**Anthropometric parameters and trophicity:** Regarding anthropometric parameters at birth, the majority of neonates (74%) had a birth weight between 2 and 4 kg. However, low birth weight was a significant finding, with 20% of the malformed children ( $n = 16$ ) weighing less than 2 kg at birth. In terms of linear growth, 73% of the patients had a height between 30 and 40 cm, while 7% ( $n = 6$ ) measured less than 30 cm. The head circumference was below 30 cm in 28% of cases ( $n = 22$ ). Furthermore, intrauterine growth restriction (IUGR) was diagnosed in 27% of the neonates ( $n = 22$ ). Among these cases of hypotrophy, 55% ( $n = 12$ ) were below the 10th percentile and 59% ( $n = 13$ ) were classified as disharmonious.

**Maternal and family factors:** The average maternal age was 28 years. The socioeconomic status was considered low in 54% of cases. A major risk factor was parental consanguinity, found in 40% of couples ( $n = 32$ ); among these, second-degree consanguinity was most frequent (62.5%,  $n = 20$ ), followed by first-degree consanguinity (18.7%,  $n = 6$ ). Maternal obstetric history showed that 19% of mothers ( $n = 15$ ) had a history of miscarriages and 1% ( $n = 1$ ) had a previous in utero fetal death (IUFD). Furthermore, prenatal care was suboptimal: 40% of mothers had no obstetric ultrasound, and the antenatal diagnosis rate was only 9%.

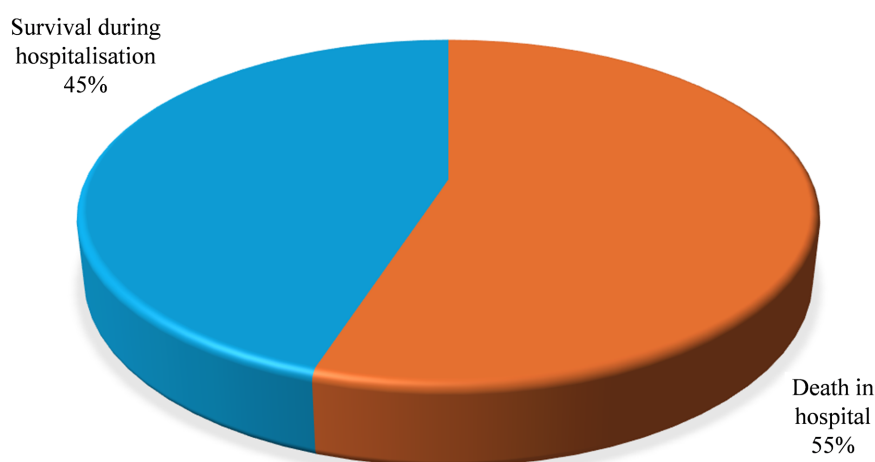
**Types of malformations:** A total of 257 anomalies were recorded in the 80 patients, an average of 3.2 malformations per child (**Table 1**). The most affected systems were: face and neck (27.2%) with low-set ears, hypertelorism and cleft lip and palate; osteoarticular (15.6%) with clubfoot and polydactyly; cardiovascular (15.5%) with VSD, ASD and complex heart disease; digestive (10.9%) with atresia (esophagus, duodenum) and Hirschsprung's disease; polymorphic malformations (9.2%) including trisomy 21 ( $n = 5$ ) and trisomy 18 ( $n = 3$ ).

**Table 1.** Distribution of patients according to the type of malformation.

Congenital malformations	Staff numbers	Frequency (%)
Facial and neck malformations	70	27.2
Osteoarticular malformations	40	15.5
Heart defects	40	15.5
Digestive malformations	28	10.8
Polymalformations	24	9.2
Urogenital and renal malformations	16	6.2
Nervous system malformations	15	5.8
Skull malformations	14	5.4
Abdominal wall malformations	5	2
Respiratory malformations	5	2
Skin and appendage malformations	1	0,4
<b>Total</b>	<b>257</b>	<b>100</b>

**Management and prognosis:** In terms of diagnosis, karyotyping could only be performed in 3 patients (3.8%) due to financial constraints. In the absence of routine cytogenetic confirmation via karyotyping, polymalformative syndromes such as trisomies 21 and 18 were identified on the basis of recognised clinical phenotypic criteria (Hall score for trisomy 21). Heart defects were confirmed by cardiac Doppler ultrasound performed by specialist paediatric cardiologists.

Management was mainly palliative (87.5%), consisting of comfort care and monitoring. Only 12.5% of children underwent curative surgery (digestive atresia, omphaloceles). The prognosis was poor, with a case fatality rate of 55% with 44 deaths (**Figure 1**). Mortality was particularly high among patients with multiple malformations (58.3%) and those with heart disease (30%) or abdominal wall malformations (60%) (**Table 2**).



**Figure 1.** Patient outcomes.

**Table 2.** Case fatality rate according to the type of malformation.

Malformation	Total number	Number of deaths	Case fatality rate
Polymalformations	24	14	58.3%
Cardiovascular malformations	40	12	30%
Digestive malformations	28	6	21.4%
Urogenital malformations	16	3	18.8%
Nervous system malformations	15	3	20%
Abdominal wall malformations	5	3	60%
Osteoarticular malformations	40	2	5%
Respiratory malformations	5	1	20%

## 4. Discussion

### 4.1. Prevalence and Hospital Profile: Selective Recruitment

Our study reports a hospital prevalence of 18.3%, a rate significantly higher than

those observed in the subregion, particularly in Cameroon (1.63%) [8] and Gabon (2.29%) [9]. This major difference can be explained by the status of the CHNEAR [7]. As a level 3 facility and national reference center, the department handles the most complex neonatal pathologies and transfers from peripheral facilities. In addition, our methodology included minor dysmorphic features (face and neck), which are often underreported in retrospective studies. Our sex ratio of 1.2 is consistent with international data suggesting an overall male predominance for most types of malformations [4]. However, this figure remains below Western data, probably reflecting a persistent underestimation linked to prehospital deaths and undocumented spontaneous abortions in our context [5]. This prevalence remains lower than in Morocco, where different environmental risk factors have been discussed [10].

#### **4.2. Consanguinity and Genetic Determinants**

The high prevalence of parental consanguinity (40%) in our cohort is a crucial risk factor for recessively inherited malformations. In our series, the high frequency of second-degree unions (62.5%) suggests a significant risk for the expression of autosomal recessive disorders, as described by Hamamy [11]. The lack of accessible genetic testing (only 3.8% of karyotypes performed) limits our ability to provide an accurate prognosis.

#### **4.3. Maternal Profiles and Screening Gaps**

The average maternal age of 28 years observed in our study reflects a young reproductive population. However, the precariousness of socioeconomic status, found in 54% of cases, remains a major concern. According to the ANSD [12], such low income is often associated with poor nutritional status and limited access to specialized prenatal care, which can increase the vulnerability to congenital anomalies [1] [8] [13]. Parental consanguinity, identified in 40% of couples, represents a significant cultural risk factor in our series. The high prevalence of second-degree (62.5%) and first-degree (18.7%) unions suggests a high risk for the expression of autosomal recessive disorders, as highlighted by Hamamy [11]. This genetic burden is further evidenced by the 19% of mothers with a history of miscarriages and the 1% who experienced a previous in utero fetal death (IUFD). These findings may indicate recurrent lethal malformations or specific structural patterns within these families, as also observed in other regional series [11] [13]. Furthermore, the prenatal screening paradox in our context is alarming. Despite a high attendance at prenatal consultations, 40% of mothers had no obstetric ultrasound, and the antenatal diagnosis rate was only 9%. This suboptimal care highlights the critical lack of morphological ultrasound equipment and trained personnel in peripheral health centers, a challenge frequently encountered in low-resource settings [8] [12]. As noted in other African series, such late or absent diagnoses prevent organized neonatal management and contribute to the high mortality rates observed in reference centers [1].

#### 4.4. The Paradox of Prenatal Care: Quantity vs. Quality

Although 54% of mothers had more than three prenatal consultations (CPN), antenatal diagnosis of the malformation was made in only 9% of cases. This discrepancy highlights the uneven quality of care: 40% of mothers did not have any obstetric ultrasounds, often due to a lack of financial resources or a shortage of healthcare services in rural areas. In addition, the almost total absence of screening for TORCH infections (33.8% not performed and no positives detected) contrasts with studies in Cameroon [8]. This suggests that current antenatal care is ineffective in screening for malformations, turning the delivery room into a place where anomalies are suddenly discovered, thereby delaying early surgical treatment.

#### 4.5. Anthropometric Parameters and Prognosis

In our study, low birth weight (LBW) was observed in 20% of the neonates. This prevalence is significant and often associated with a higher risk of neonatal mortality in cases of congenital anomalies [1] [3]. Our findings align with several African series, notably in Gabon [9] and Morocco [10]. Furthermore, intrauterine growth restriction (IUGR) was present in 27% of our cases. This high rate of hypotrophy, particularly the disharmonious type (59%), suggests chronic fetal suffering that could be linked to the malformative process itself or to shared etiological factors, such as maternal nutritional status [1] [11]. The head circumference, which was below 30 cm in 28% of our patients, is also a concerning clinical sign. In the literature, a reduced head circumference is frequently associated with specific polymalformative syndromes and neurodevelopmental delays [2] [4]. Finally, the combination of low birth weight and severe malformations significantly increased the case fatality rate in our series. As noted in other studies conducted in the sub-region, notably in Cameroon [8] and Senegal [7].

#### 4.6. Lesion Profile

The distinctive nature of facial and osteoarticular abnormalities unlike traditional series, our study places malformations of the face and neck in first place (27.2%). This unusual result stems from our exhaustive listing of dysmorphic features (low-set ears, hypertelorism) that are often included indiscriminately in polymalformations syndromes by other authors. Osteoarticular (clubfoot, polydactyly) and cardiac malformations rank second (15.6%). It is interesting to note that neural tube defects are less frequent (6th position) than in other African series [14]. This could indicate the beginning of the effectiveness of the folic acid supplementation policy, from which 82.5% of our pregnant women benefited, corroborating Honein's observations on the impact of fortification [15].

#### 4.7. Diagnostic Impasse and Neglected Psychological Burden

Our study illustrates the "genetic divide" between the North and the South. A definitive diagnosis (karyotype) was only made in 3 patients (3.8%) due to a lack of

financial resources and accessible laboratories. This lack of an etiological diagnosis prevents accurate genetic counseling for future pregnancies, leaving families in uncertainty. In terms of treatment, the rate of curative surgery is alarming (12.5%). Most patients (87.5%) received palliative or expectant care. Unlike developed countries where neonatal surgery is standardized, we face a critical shortage of technical facilities that condemns children with conditions that are curable elsewhere (omphaloceles, digestive atresia).

#### **4.8. Neglected Psychological Burden**

No parents received formal psychological support, a finding shared by Oulai in Côte d'Ivoire [16]. The announcement of a disability at birth is a violent trauma, often aggravated by cultural beliefs that induce guilt. The absence of psychologists in the maternity and neonatal wards at CHNEAR is a major shortcoming that isolates families in their distress.

#### **4.9. Prognosis and Mortality**

With an in-hospital case fatality rate of 55%, our series reveals a grim prognosis, much more severe than the 33% in Cameroon [8]. This excess case fatality rate can be explained by the severity of the cases recruited and the lack of immediate surgical solutions. The mortality was primarily driven by multiple malformations (31.8% of deaths) and complex heart conditions (27.3% of deaths). However, the prognosis is worsened by classic neonatal vulnerability factors: 32% of newborns were preterm and 20% weighed less than 2 kg. In the absence of immediate curative surgical solutions for 87.5% of patients, these comorbidities contribute synergistically to the fatal outcome. Case fatality rate is particularly critical among abdominal wall defects (60%), confirming that without improvements in surgical facilities, the survival of these newborns remains compromised.

#### **4.10. In-Hospital Mortality and Challenges in Patient Care**

The overall mortality rate of 55% observed at CHNEAR is particularly alarming. This disparity can be explained by our status as a level 3 referral centre. However, the international study PaedSurg Africa published in *The Lancet* (2021) demonstrates that mortality in low-income countries is seven times higher than in high-income countries, mainly due to delays in transfer and a lack of surgical intensive care units [17]. In our series, the interaction between the anatomical abnormality, prematurity (32%) and low birth weight (20%) creates extreme vulnerability.

#### **4.11. Limitations of the Study**

The single-center and hospital-based nature of the study constitutes a selection bias, as it does not reflect actual community mortality (deaths at home). In addition, the lack of genetic confirmation for most dysmorphic syndromes limits the nosological accuracy of our results.

## 5. Conclusion

Congenital malformations at CHNEAR are characterized by their severity and late diagnosis. Consanguinity appears to be a major contributing factor. To reduce this high mortality rate, it is imperative to strengthen prenatal diagnosis, make genetic testing accessible, and improve neonatal surgery facilities. The creation of a national registry of malformations is necessary to better understand and prevent these pathologies in Senegal.

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## Conflicts of Interest

No conflicts of interest in relation to this article.

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