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# Management of Congenital Disorders in a Resource-Limited Country: Organizational Model of a Specialized Working Group within a Perinatal Network

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## Abstract

Objective: To evaluate the organizational model of a perinatal network and its relevance in a resource-limited country. Methodology: This was a mixed prospective qualitative and quantitative study conducted over a 2-year period, from January 1, 2022, to December 31, 2023. This study took place in Senegal, a country with limited resources and a weakness of hyperspecialized medical technical resources. There was no policy for the management of fetal malformations. The qualitative part was carried out through overt participant observation. The human resources and the organization of the perinatal network were described. For the quantitative part, all fetuses managed during the study period were included. The studied parameters related to neonatal care and outcomes. Qualitative variables were described using dispersion parameters, and quantitative variables were described using proportions. Results: The perinatal network includes several specialists across six hospitals. Of these hospitals, only one provided emergency pediatric surgery. The network included highly specialized human resources in prenatal diagnosis, congenital heart defects, pediatric surgery, anesthesia, and other medical specialties in perinatology. Advanced ultrasound was centralized by an obstetrician. The team decided on the follow-up methods, timing, and mode of delivery. The newborn was immediately transferred to the appropriate specialty. Over the 2-year period, 201 fetuses were managed. The rate of cesarean delivery was 76.3%. Neonatal mortality was 51.4%. Discussion: Centralizing care improves the quality

of prenatal diagnosis and management of congenital defects. Mortality remains high when emergency surgery is not well available. This mortality is also due to the lack of a single center offering all perinatal care and so, the transfer of newborns. The cesarean rate increases due to underlying conditions and organizational factors. Conclusion: Public policies should prioritize the centralization of care for congenital disorders to reduce the costs of disability and mortality.

## Keywords

Congenital Disorders, Perinatal Network, Resource-Limited Country, Antenatal Diagnosis, Care Centralization

## 1. Introduction

Congenital disorders are relatively common health events, accounting for approximately 3% of births [1]. Congenital anomalies are the leading cause of infant mortality, morbidity, and worldwide disabilities. Around 303,000 newborns die each year before reaching one month of age due to congenital anomalies [2]. Furthermore, congenital malformations represent a significant portion of disability-adjusted life years (DALYs), approximately 57.7 million [2] [3].

In high-income countries, centralizing the management of congenital malformations enables the provision of highly specialized care [4]. These advancements reduced mortality rates to below 10% for previously lethal conditions [3]. Moreover, many anomalies are rare diseases, and centralizing care improves practitioners' experience [4].

In resource-limited countries, fetal medicine is either nonexistent or underdeveloped. Thus, such hyperspecialized centers do not exist. Children with life-threatening anomalies are at risk of death during transfers to care centers [3]. Diagnosis and management are inadequate due to a lack of experience and insufficient technical resources.

In Senegal, faced with the high preventable mortality linked to the lack of centralized care for congenital anomalies, a group of specialists formed a perinatal network to improve the outcomes of these conditions. The objective of this study was to evaluate the organizational model of the perinatal network and its relevance in a resource-limited country.

## 2. Patients and Methods

This was a mixed prospective cross-sectional study, both qualitative and quantitative, conducted over a 2-year period from January 1, 2022, to December 31, 2023. The study was carried out in several hospitals in Dakar (Senegal).

For the qualitative component, we conducted overt participant observation. The sample included all specialists and hospitals involved in the organization and

care of fetuses with congenital defects. The limited number of specialists prevented us from reaching data saturation. The data collection tool consisted of on-site notes. The data collected were related to the number and type of specialists in the working group, the medical facilities available in each hospital, and the organizational model of the perinatal network. Data triangulation was achieved through complementary data collection methods. We used inductive thematic analysis, describing the most significant themes.

In the quantitative component, we included in the study all fetuses with a detected anomaly who were managed within the perinatal network both antenatally and postnatally. We did not include malformations that were not prenatally diagnosed in the network. Data were recorded prospectively using the SOSEPERD® database, which operates on Filemaker® software. This database is updated regularly. The parameters studied included the diagnosis, management, outcomes and lethality of the congenital disorders. Data collected from the Filemaker® database were exported to Excel® and then, analyzed using the Statistical Package for Social Sciences 20.0 (SPSS).

We calculated the specific lethality rates of certain anomalies. Qualitative variables were described using position measures, and quantitative variables were described using proportions. This study received approval from the local ethics committee under number 22UD/24. All patients registered in the database consented to the anonymous use of their data.

## 3. Results

### 3.1. Qualitative Results

The perinatal network includes several specialists working across six different hospitals and the University of Dakar. Among these hospitals, the Dalal Jamm National Hospital Center (DJNHC) had a maternity ward and a Neonatal Intensive Care Unit (NICU) with a capacity of 12 incubators and radiant warmers. The antenatal diagnosis specialist worked at this hospital. The Pikine National Hospital Center (PNHC) had a pediatric surgery department and is the only hospital offering emergency pediatric surgery. However, its neonatology unit was limited, with only two incubators. The Albert Royer Children's Hospital (ARCH) had the largest NICU with a capacity of 38 incubators but lacked a maternity ward. This hospital also had a pediatric surgery department, but emergency surgeries were not performed. Moreover, specialized consultations in genetics, neurosurgery, pediatric dermatology, and pediatric nephrology were available. The hospital also had a cardiovascular surgery department. The Diamniadio Children's Hospital had both a neonatology unit and a pediatric surgery department but without maternity ward. Additionally, a cardiologist specializing in congenital malformations worked at Thiès Hospital, located in another region of Senegal. Similarly, the fetal pathology specialist worked in the southern area of the country (**Table 1**).

Table 1. Distribution of care and specialists in the different hospitals.

| Hospital | Available care or specialist |
|---|---|
| Dalal Jamm National Hospital Center | Prenatal diagnosis |
| | Neonatal intensive care |
| | Maternity unit |
| Pikine National Hospital Center | Emergency neonatal surgery |
| | Maternity unit |
| Albert Royer Children's Hospital | Neonatal intensive care |
| | Planned neonatal surgery |
| | Genetics consultation |
| | Pediatric nephrology |
| | Neurosurgery |
| | Cardiac surgery |
| | Pediatric dermatology |
| Diamniadio Children's Hospital | Planned neonatal surgery |
| | Neonatal intensive care |
| Abass Ndao Hospital | Pediatric endocrinology |
| Thies Hospital Center | heart defect specialist |

Due to the limitations of each hospital, specialists decided to unite and create a perinatal network to coordinate patient care. The network included:

- 4 obstetricians, including 1 specializing in antenatal diagnosis,
- 4 neonatologists,
- 1 neurosurgeon,
- 2 cardiologists specializing in congenital heart defects,
- 8 pediatric surgeons, including 2 specialized in urology and 1 in orthopedic surgery,
- 1 endocrinologist,
- 2 geneticists,
- 2 pediatric nephrologists,
- 2 radiologists specializing in pediatric imaging,
- 1 anesthesiologist specialized in neonatology,
- 1 fetal pathology specialist, and
- 1 psychiatrist.

These specialists carried out their daily activities in the different hospitals (Table 1).

The perinatal network had provided a 24/7 phone line for all obstetricians across the country.

After screening, obstetricians in different regions informed the prenatal diagnosis specialist through the phone line directly. The prenatal diagnosis specialist

scheduled an appointment at Dalal Jamm Hospital. Thus, these patients were evaluated at DJNHC for detailed morphological ultrasound and possible invasive sampling. After a significant number of fetuses with congenital disorders are collected, a multidisciplinary team meeting is held at ARCH. Various physicians provided indications, according to their specialty, for antenatal and postnatal care, and delivery conditions. Thus, the mode and timing of delivery were determined during the meeting. In some cases, prenatal consultations were organized with the specialist who will care for the newborn. These consultations help to provide information to parents about the infant's subsequent care and prognosis. After the meeting, parents were informed, and additional procedures were carried out if necessary. Psychological support was recommended for all parents.

Patients with late screening, in the third trimester or in emergency conditions were discussed directly with the relevant postnatal specialty after diagnostic ultrasound. The patient's specific care pathway, which is antenatal care, mode and timing of delivery, was established.

Specifically in cases of congenital heart defects, a diagnostic ultrasound was performed in the presence of the pediatric cardiologist. A fetal echocardiography appointment was arranged between the obstetrician, the cardiologist, and the patient at DJNHC. This collaboration during the ultrasound allowed for a more precise diagnosis. The pediatric cardiologist also provided guidance on monitoring depending on the evolving nature of the pathology and the need for in utero treatment. The presence of cardiologist was also essential for parents' counseling. The neonatologist always performed the initial care and the newborn was, then transferred to the appropriate care facility. The Emergency Medical Assistance Service (EMAS) was involved in newborns' transfer.

The perinatal network set up a WhatsApp® group including all specialists to maintain continuous communication on clinical cases and their progress outside of formal meetings.

A database using FileMaker software contained the electronic records of all infants. The records were updated during evaluations and care. Specialists had specific login passwords to access the online patient records and update them.

### 3.2. Quantitative Results

During the study period, 201 abnormal fetuses were treated. The average age of the patients was 28 years (standard deviation: 5.367). The screening was performed late, with an average gestational age of 27.1 weeks at the time of diagnosis.

The conditions treated during the study period are summarized in **Table 2**. We performed invasive tests (amniocentesis or fetal blood sampling) in 9.7% of the patients for diagnostic purposes. Antenatal care included amniodrainage, amnioinfusion, bladder drainage, thoracic drainage and antiarrhythmic treatments. The rate of cesarean section was 76.3%. Surgery was performed on 29.1% of the newborns. We recorded a high mortality rate: 51.4%. Among these deaths, 68.4% were avoidable, as only 31.6% of the conditions were recognized as lethal. These lethal

conditions were primarily anencephaly, exencephaly, thanatophoric dysplasia, bilateral renal agenesis, alobar holoprosencephaly, bilateral multicystic dysplastic kidney, Meckel-Gruber syndrome …

**Table 2.** Distribution of fetal defects monitored by the perinatology network.

| Fetal defect | Prevalence (%) | Fetal defect | Prevalence (%) |
|---|---|---|---|
| **Digestive tract and abdominal wall** | | **Heart and thoracic malformation** | |
| Omphalocele | 3.5 | Hypoplastic left ventricle syndrome | 4.0 |
| Gastroschisis | 3.5 | Heart block | 3.0 |
| Anorectal malformation | 1.5 | Conotruncal heart defects | 2.0 |
| Esophageal atresia | 1.5 | Ventricular septal defect | 0.5 |
| Pentalogy of Cantrell | 1.5 | Tricuspid valve dysplasia | 0.5 |
| **Central nervous system** | | Tachyarythmia | 1.5 |
| Ventriculomegaly | 7.0 | Ebstein anomaly | 0.5 |
| Holoprosencephaly | 4.0 | Diaphragmatic hernia | 1.5 |
| Dandy-Walker malformation | 4.0 | Congenital lobar emphysema | 0.5 |
| Anencephaly | 3.0 | Hydrothorax | 0.5 |
| Spinal dysrraphism | 2.0 | **Kidney and urinary tract anomalies** | |
| Exencephaly | 2.0 | Posterior urethral valves | 7.0 |
| Galen vein aneurysme | 0.5 | Ureteropelvis junction obstruction | 3.5 |
| Agenesis of corpus callosum | 0.5 | Multicystic kidney dysplasia | 3.0 |
| **Short long bone** | | Kidney agenesis | 1.5 |
| Tanatophoric dysplasia | 3.0 | Urethral atresia | 1.5 |
| Hypophosphatasia | 1.5 | Autosomal recessive | 1.5 |
| Achondroplasia | 0.5 | polycystic kidney disease | 0.5 |
| **Twin anomalies** | | Bartter syndrome | 0.5 |
| Twin to twin transfusion syndrome | 3.5 | Autosomal dominant polycystic kidney disease | 0.5 |
| Conjoined twin | 3.5 | **Other defects** | |
| **Fetal tumors** | | Unexplained fetal hydrops | 3.0 |
| Sacrococcygeal teratomas | 1.5 | Turner syndrome | 2.0 |
| Ovarian teratoma | 0.5 | Epidermolysis bullosa | 1.5 |
| Congenital mesoblastic nephroma | 0.5 | Others* | 10 |

Others* = Turner syndrome, congenital adrenal hyperplasia, Limb-body-wall-complex, OEIS complex, Meckel-Gruber syndrome, Vacterl association, Beckwith-Wiedemann syndrome, Hutchinson-Gilford progeria syndrome, amniotic bands, Zellweger syndrome, trisomy 21, dysferlinopathy, Cytomegalovirus infection.

The specific lethality of urethral atresia, hypoplastic left ventricule syndrome,

diaphragmatic hernia, OEIS complex syndrome, and hydrothorax was 100%. One of the conjoined twins was successfully separated in one year. The specific lethality of gastroschisis was 60% (Table 3).

Table 3. Specific lethality of different congenital defects.

| Congenital defect | Specific lethality |
|---|---|
| Urethral atresia | 100% |
| Twin to twin transfusion syndrome | 40% |
| Epidermolysis bullosa | 100% |
| Conotruncal heart defects | 100% |
| Bilateral multicystic kidney dysplasia | 100% |
| Hypoplastic left heart syndrome | 100% |
| Diaphragmatic hernia | 100% |
| Hydrothorax | 100% |
| OEIS Complex | 100% |
| Gastroschisis | 60% |
| Anorectal malformation | 100% |
| progeria syndrome | 100% |
| Conjoined twin | 75% |

## 4. Discussion

### 4.1. Key Findings

The perinatal network has highly specialized human resources, and care is optimized through multidisciplinary meetings and coordination. However, the availability of emergency surgery is significantly lacking, and the absence of a centralized care unit necessitates the use of the emergency medical services (EMAS) for patient transfers. The use of invasive diagnostic tests remains insufficient, as does antenatal treatment. The cesarean section rate is very high: nearly 3 out of 4 women deliver by cesarean. Additionally, several conditions still have a high specific lethality rate.

### 4.2. Interpretation of the Results

Antenatal diagnosis was primarily based on ultrasound for most fetuses. A study by Goley *et al.* evaluated the role of ultrasound in antenatal diagnosis in countries with limited and moderate resources. In this systematic review, antenatal diagnosis rates varied from 1.8% in Nigeria to 90.5% in China, and even 100% in Thailand. There is also significant disparity within individual countries [2].

These inconsistent results are linked to both, the quality of the equipment and

the expertise of the practitioner. In this study, the authors associate diagnostic performance with training and experience. Indeed, the highest detection rates were achieved by practitioners who either centralized all fetal anomaly ultrasounds or had 5 to 6 years of experience. For example, a radiologist trained in fetal anomalies in the United Kingdom detected 93.8% of anomalies. In Turkey, a detection rate of 90.5% was achieved by a radiologist who centralized all fetal anomaly ultrasounds. In Romania, a detection rate of 97.4% was obtained by obstetricians specialized in prenatal diagnosis [2].

This organization enables a better antenatal diagnostic rate via ultrasound. However, the use of invasive procedures for biological diagnosis remains low, largely due to the cost of this technique. Technological advancements in genetics (karyotyping, FISH, microarray, exome sequencing, targeted testing) and other biological tests have had a significant impact on diagnosing congenital anomalies. Fetal imaging alone, often does not allow for a differential diagnosis. Establishing a diagnosis is crucial for decision-making regarding subsequent management, such as in utero intervention or mode of delivery [5]. These tests also allow for genetic counseling. Given that 50% of congenital malformations have a genetic component [6], implementing genetic testing in Low-resources countries is essential to improve care.

In this perinatal network model, care is centralized around specialized human resources. Although there is no single center bringing together all of these human and technical resources.

The strengths of the network were linked to the quality of highly specialized physicians in various areas of perinatology. Additionally, the network allowed for coordinated care through meetings and planned the patient's care pathway from the antenatal to postnatal period. Finally, by managing a large number of congenital disorders, the network enabled specialists to gain more experience in fetal medicine and the management of fetal defects.

The main weakness was the lack of a single center where all specialists worked in coordination. This situation required transfers after birth, which increased morbidity and mortality. Likewise, specialists were not always present during unexpected births.

Recently, in Europe, diagnostic and care centers for fetal malformations have been centralized. These centers provide highly specialized care that improves the prognosis for malformations [7].

In Finland, a government decision centralized all pediatric cardiac surgeries, organ transplants, and biliary atresia surgeries in a referral hospital in Helsinki. This centralization significantly improved neonatal survival rates. For example, the survival rate after biliary atresia surgery increased from around 60% to nearly 100% post-centralization [4].

In Sweden, health authorities decided to centralize cardiac surgeries. Thirty-day postoperative mortality after open-heart surgery decreased from 9.5% in 1988 to 1.9% in 1997 [4].

The mortality rate is 51.4% in our study. According to Sitkin *et al*., this rate is

around 10% in some countries [3]. Part of our mortality rate is also due to the lack of pregnancy termination options because of the prevailing legislation.

The perinatal network recorded a high cesarean section rate (76.3%). This rate may be due to the need to schedule delivery and so, ensuring the presence of the specialist responsible for the affected organ and optimizing newborn care. For example, the pediatric cardiologist must travel from one region to another for delivery of fetus with heart defect requiring immediate care. The high cesarean rate may also be due to underlying fetal condition.

In the literature, cesarean rates vary depending on the pathology. The rate ranges from 20.2% to 44.9% in cases of congenital heart defects or neural tube defects. In cases of fetal surgery for neural tube defects, the cesarean rate is 100%. The cesarean delivery rate varies between 22.9% and 82.4% in cases of gastroschisis. In cases of fetal tumors beyond 37 weeks, the cesarean section incidence is 63.8% [8].

The specific lethality of certain life-threatening, but treatable conditions if emergency surgery is performed, remains very high in our network. This elevated rate is related to the lack of availability of emergency surgery, the need for transfers, and the inadequate technical infrastructure.

For example, the lethality of diaphragmatic hernias and conotruncal heart defects is 100%. Mortality in cases of gastroschisis is 60%. In the literature, the mortality rate for gastroschisis ranges from less than 5% to 33.2% [9]-[11]. Regarding diaphragmatic hernias, it ranges from 18% to 25.8% [12]-[14]. The mortality associated with conotruncal heart defects is 10.7% in Italy [15] (**Table 4**).

**Table 4.** Specific lethality of certain congenital defect.

| Studies | Diaphragmatic hernia | Gastroschisis | Conotruncal heart defect |
|---|---|---|---|
| Politis *et al.* | 21.3% | - | - |
| Gutpa *et al.* | 25.8% | - | - |
| Zalla *et al.* | 18% | - | - |
| Egger *et al.* | - | 33.2% | - |
| Georgeades *et al.* | - | <5% | - |
| Jwa *et al.* | - | 23.5% | - |
| Anaaclerio *et al.* | | | 18% (syndromic disease)<br>10% (non-syndromic disease) |

These figures highlight a significant gap that needs to be addressed by upgrading the technical equipment.

## 5. Conclusion

Fetal malformations are a frequent health event, accounting for 3% of births, but each condition is often rare. Multidisciplinary care and life-threatening conditions

that require immediate intervention, necessitate the centralization of care in these cases. Indeed, the improvement of outcomes has been demonstrated in the literature in this context. Centralization must encompass both human and structural resources. A significant gap remains in care delivery in resources-limited countries. Centralizing care in these settings should be a priority.

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## Conflicts of Interest

We have no conflict of interest to declare.

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