

## **Congenital Malformations: The Importance of Early Detection**

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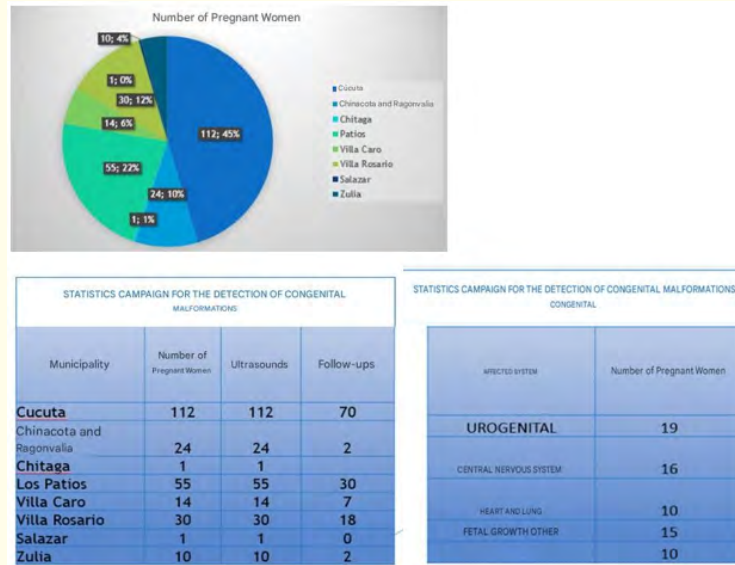
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A congenital anomaly is a defect present at birth in an organ or region of the body; these are anatomical alterations that occur during the intrauterine stage. They affect between 5 - 10% of all live births and account for 7 - 8% of neonatal deaths. Congenital malformations are the leading cause of mortality worldwide in children under 1 year of age, also causing high morbidity in childhood and nearly 3.2 million disabilities annually. Genetic, environmental, and multifactorial causes have traditionally been identified as the causes of these anomalies, with genetic factors being the most common cause.

Congenital defects represent a significant medical problem for society, as they are largely responsible for perinatal and infant morbidity and mortality. In many cases, they cause complications and sequelae associated with varying degrees of disability that compromise the individual's development and social integration. The prevalence of congenital anomalies in Latin America is estimated at 3 - 8%, a figure that justifies the establishment of screening programs aimed at improving prenatal detection rates. In 2012 - 2013 in Norte de Santander, Colombia, perinatal mortality was 11.6/100,000, and the number of cases of fetuses with multiple malformations (mostly incompatible with life), according to statistics from the Maternal-Fetal Medicine Unit of the H.U.E.M. (Cucuta) and the NORFETUS Fetal Diagnosis and Therapy Unit, was 61 cases that year.

The NORFETUS Maternal-Fetal Medicine Unit in Cucuta, Colombia, founded in 2013, has provided access to advanced ultrasound scans to many pregnant women in the Norte de Santander region of Colombia over the years, with the following statistics: 2,375 pregnant women in 2013 - 2014, 3,696 pregnant women in 2015, 5,628 pregnant women in 2016 - 2017, 7,074 pregnant women in 2018, and 7,657 pregnant women in 2019 - 2020. The most frequently detected congenital malformations worldwide are: Genitourinary, heart disease, and fetal central nervous system disorders.

The Early Detection and Management of Congenital Malformations program in Norte de Santander, led by the Norte de Santander Governor's Office and with scientific and technological support from the NORFETUS Maternal-Fetal Medicine Unit, began in 2016- 2017, offering nearly 300 pregnant women annually in the subsidized health system to access advanced ultrasounds for early identification of abnormalities in their babies. The idea is to provide coverage to most pregnant women in the department. In 2016, the pilot program began in the municipalities of Norte de Santander with the highest perinatal mortality rates between 2014 and 2015, which were: Cucuta, Villa Rosario, Ocaña, Los Patios, Pamplona, and Tibu. The first 252 ultrasound evaluations were performed, with the following statistics:



Figure

In 2016 - 2017, 70 fetuses were found to have malformations, most of which were urogenital and central nervous system abnormalities. In 2018, a total of 68 fetuses had malformations, distributed as follows: Urinary tract abnormalities 19 cases, fetal growth abnormalities 16 cases, Heart defects 10 cases, central nervous system abnormalities 4 cases, placental and amniotic fluid disorders 4 cases, and other abnormalities 15 cases. However, once these congenital fetal abnormalities have been detected and identified early on, they are monitored until birth, with multidisciplinary support in some cases. From the outset, emphasis is placed on providing support to parents and close family members by a specialist in perinatal psychology, in order to reduce the psycho-affective and emotional consequences of having a child with a malformation. Here we examine the four stages of the grieving process triggered by the news of a high-risk pregnancy or the birth of a child with difficulties, detailing the main psychological mechanism used by parents in each of the stages and formulating recommendations for managing their attitudes in this situation. The course of the pregnancy is entrusted to monthly medical monitoring; every warning sign has a medical response; childbirth is a process, reliable and controlled. The loss of a pregnancy, the early diagnosis of an unfavorable and irreversible situation, or the death of a newborn are experienced as an unparalleled tragedy and undoubtedly provoke much more intense psychological reactions.

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