Perinatal Care of Infants with Congenital Birth Defects

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KEYWORDS

• Congenital anomalies • Congenital heart disease • CDH • Perinatal management
• Resuscitation

KEY POINTS

- Prenatal imaging has allowed for in utero diagnosis of congenital anomalies.
- Most neonates with congenital anomalies need only standard resuscitation, but the availability of advanced care is crucial.
- In utero intervention and specialized delivery are indicated for a small proportion of patients.
- Standardization of care using clinical algorithms might be beneficial to neonatal outcomes.

INTRODUCTION

Perinatal care for infants with congenital birth defects has changed dramatically over the past century. Previously, infants with congenital anomalies presented at birth if defect(s) were obvious on physical examination, and in the neonatal period or childhood if the defect or defects were more subtle. Delayed diagnosis is associated with worse outcomes for certain anomalies, but this is dependent on the type and severity of the anomaly and which outcome is being evaluated.[1,2] Prenatal diagnosis allows for changes in perinatal management to benefit the newborn, including increased monitoring, counseling, changes in the mode and timing of delivery, fetal intervention, and planning for postnatal care.[1] However, definitive data are rare and documented improvement is limited to a few diagnoses.[2–4] In the last decade, interventions aimed at preventing mortality have transitioned to those aimed at improving quality of life, complicating the ethical concerns regarding maternal well-being and autonomy, as well as research and innovation.[5,6]

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PRENATAL DIAGNOSIS AND MANAGEMENT

Prenatal Imaging and Diagnosis

Prenatal obstetric management changed dramatically with the application of ultrasound technology in the 1960s, and subsequently, Doppler sonography and 3-dimensional ultrasound.[1,7] Incorporation of ultrasound into routine pregnancy management allows for dating, detection of multiples, evaluation of growth, monitoring of well-being, and diagnosis of congenital anomalies and aneuploidy. Although widely available and cost-effective, ultrasound is limited by gestational age, maternal body habitus, amniotic fluid volume, image resolution, and fetal positioning.[1] The development of fetal MRI has helped overcome some of these limitations and allows for more detailed visualization. However, fetal MRI is not routinely used except for in limited circumstances when a fetal anomaly is known or strongly suspected, but further visualization beyond that which can be obtained with ultrasound is needed. More widespread use of fetal MRI is limited because of concerns regarding cost, access to equipment, and lack of trained personnel to acquire and interpret images.[1,8]

Referrals

Infants with an anomaly diagnosed as likely to impact the neonatal period should be referred to perinatology (maternal-fetal medicine).[6] Perinatologists provide counseling, follow-up, and further referral to consultation with neonatology, pediatric subspecialists, or a fetal care center as appropriate. Fetal care centers evolved through a collaborative approach between subspecialties; referral is typically indicated if the fetus is a candidate for fetal intervention or if immediate postdelivery care will be required. Centers vary in the services offered: some are multidisciplinary clinics offering diagnostic services and prenatal consultation, whereas others also offer pregnancy management, fetoscopic interventions, or open surgeries.[9]

Anticipatory Guidance and Counseling

Prenatal diagnosis of an anomaly allows for education of the family, including anticipatory guidance and likely decisions on care. Providers should attempt to present options with minimal bias, which may include prenatal intervention, postnatal treatment, palliative care, or termination of pregnancy.[6] Perinatologists and obstetricians, pediatric subspecialists, pediatric surgeons, and neonatologists can all provide unique and area-specific counseling and recommendations.[10] Counseling varies among and within different specialties; thus, it is important to ensure a coordinated approach.[10,11] Social workers, genetic counselors, and palliative care services also provide support to families. Evidence is limited regarding whether parental stress is lower if the diagnosis is made in the prenatal compared with postnatal time period, but studies have demonstrated that comprehensive, multidisciplinary counseling on fetal anomalies after diagnosis can decrease maternal anxiety.[1,2,10]

Maternal and Fetal Monitoring

Close fetal surveillance with ultrasound, nonstress testing, and potentially advanced imaging can provide updates on evolution of the disease process and fetal well-being.[2] Development or progression of intrauterine growth restriction or hydrops is a common complication of some congenital anomalies that may prompt early delivery. It is also important to monitor for maternal “mirror syndrome” with hydrops.[8] If fetal intervention occurs, the mother should be monitored closely for complications, such as uterine dehiscence or premature rupture of membranes.