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Syndromic congenital diaphragmatic hernia: current incidence and outcome

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Objective

Congenital Diaphragmatic Hernia (CDH) associated with genetic syndromes is rare. The aim of this study was to describe the incidence of CDH associated with known syndromes, with or without underlying genetic etiology, and the postnatal outcomes from a large database for CDH.

Methods

Data from the multicenter, multinational database on infants with CDH (CDHSG Registry) born from 1996 to 2020 were analyzed. Patients with known syndromes or associations, were grouped and data was analyzed and compared to those without the abnormalities.

Results

A total of 12 553 patients were entered in the registry during the study period, and 421 had reported known syndromes, representing 3.4% of all CDH cases in the registry. The overall survival to discharge for syndromic CDH was 34%. A total of 50 different associated syndromes were reported. The most common were: Fryns Syndrome (19.7% of all syndromes, 17% survival), Trisomy 18 or Edward syndrome (17.5%, 9% survival), Trisomy 21 or Down Syndrome (9%, 47% survival), Trisomy 13 or Patau Syndrome (6.7%,14% survival), Cornelia de Lange Syndrome (6,4 % of all syndromes, 22% survival) and Pallister-Killian Syndrome (5.5 % of all syndromes, 39.1% survival). The syndromic CDH group had lower birth weight and gestational age at birth, and increased incidence of bilateral CDH (2,9%) and rates of non-repair (53%). The length of hospital stay was longer, as was the need for O2 at 30 days. Extracorporeal life support (ECLS) was used only in 15% of the cases. Other malformations were more common in syndromic CDH. Those who underwent surgical repair had survival to discharge rates of 73%.

Conclusion

Syndromic CDH is rare, with low survival rates. Given higher rates of non-repair and decreased ECMO use, along with a high early mortality, decision-making regarding goals of care clearly influence outcome. Survival varies depending on the genetic cause. Early genetic diagnosis is important and may influence the decision making.