

Non-isolated congenital diaphragmatic hernia: outcomes in genetic versus structural causes

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OBJECTIVES

To examine imaging and outcomes in cases of non-isolated congenital diaphragmatic hernia (CDH) in subgroups according to structural, genetic, or structural+genetic comorbidity.

METHODS

This was a retrospective cohort study of fetuses with non-isolated CDH at a tertiary center between 2012 and 2025. Cases were categorized as structural, genetic, or structural + genetic. Prenatal imaging findings and neonatal outcomes were compared across groups.

RESULTS

A total of 210 non-isolated CDH cases were included: 101 (48.1%) structural, 31 (14.8%) genetic and 78 (37.1%) structural+genetic.

The structural+genetic group was evaluated at an earlier gestational age (median [IQR]: 26.0 [23.1, 29.1] vs 28.4 [25.1, 33.4] vs 26.8 [23.6, 29.1], p= 0.031). Initial observed-to-expected lung-to-head ratio (o/e LHR) was lowest in the structural+genetic group (median [IQR]: 28.0 [21.0–30.2] vs 33.5 [27.0–45.5] vs 37.5 [26.0–46.0], p = 0.049). Postnatal defect size was largest in the structural+genetic group (median [IQR]: 85.0 [75.0–90.0] vs 70.0 [50.0–87.0] vs 55.0 [20.0–80.0], p = 0.019).

Despite these differences, ECMO utilization, pulmonary hypertension, and survival were similar across groups.

FIGURE 1. PRENATAL LUNG MEASUREMENT

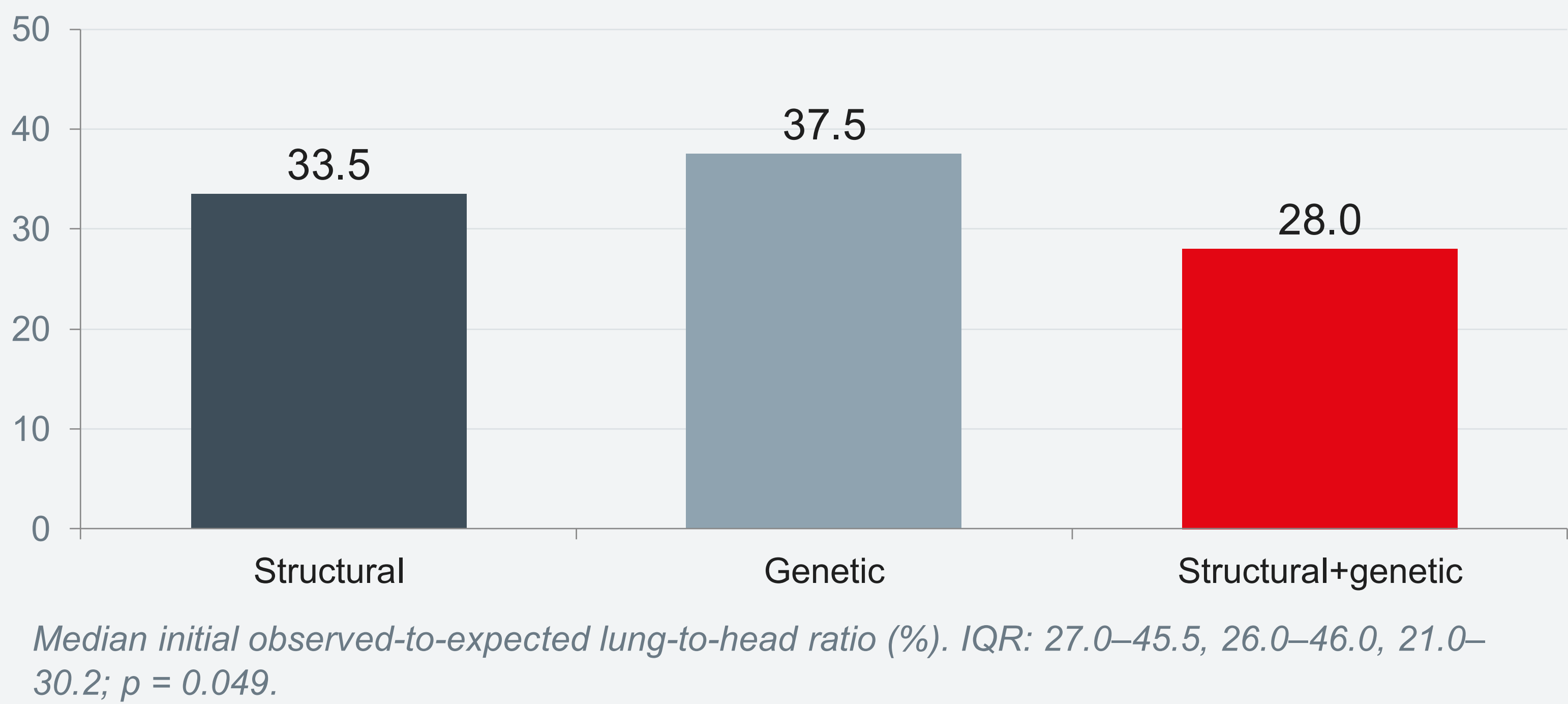
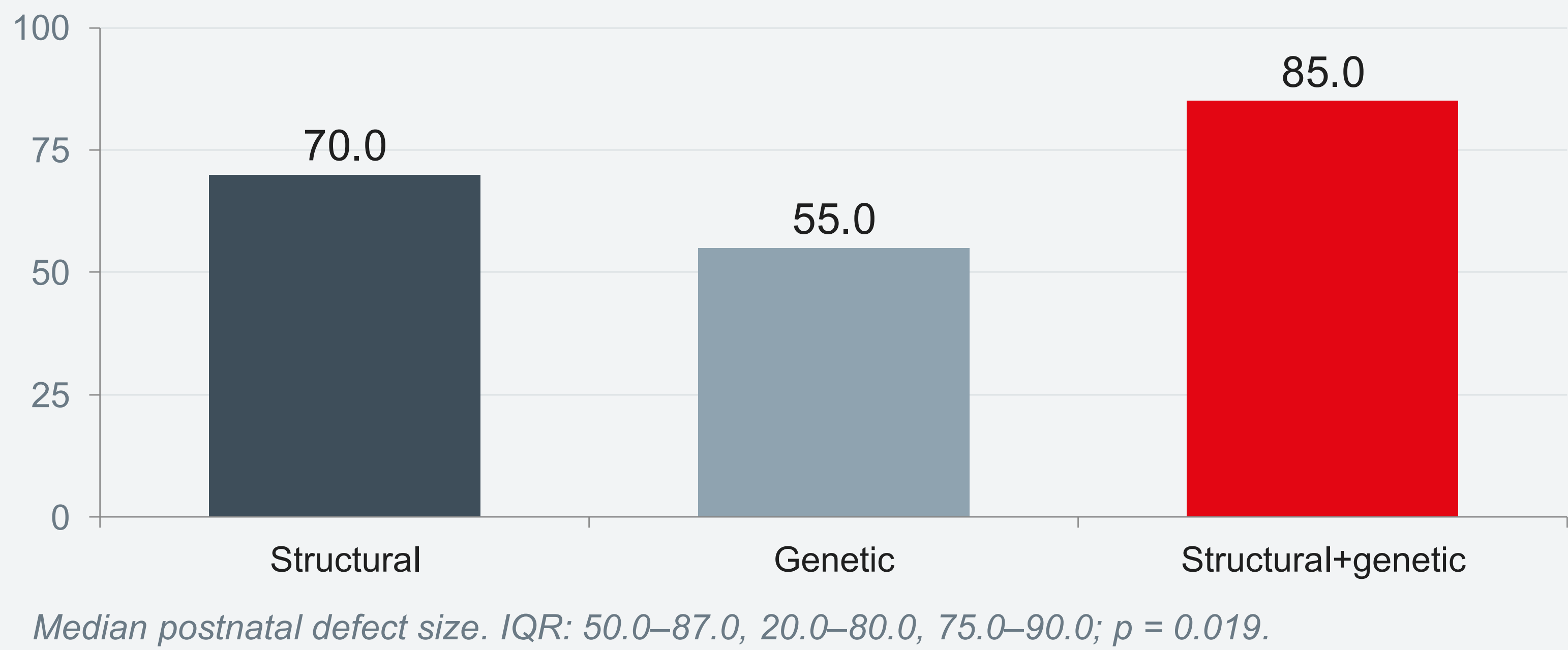


FIGURE 2. POSTNATAL DEFECT SIZE



AT A GLANCE

210
non-isolated CDH cases

101 / 31 / 78
structural / genetic / structural+genetic

28.0
lowest median o/e LHR, structural+genetic
(p = 0.049)

85.0
largest median defect size,
structural+genetic (p = 0.019)

CONCLUSIONS

- Non-isolated CDH cases demonstrate substantial heterogeneity, highlighting the importance of genetic testing and counseling along with prenatal imaging.
- Cases with structural+genetic findings show lower o/e LHR but similar short-term neonatal outcomes compared with other non-isolated subgroups.