

Isolated versus non-isolated congenital diaphragmatic hernia: does it matter?

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OBJECTIVES

To compare prenatal characteristics, genetic evaluation, and neonatal outcomes between isolated and non-isolated congenital diaphragmatic hernia (CDH).

METHODS

This retrospective cohort study included fetuses diagnosed with CDH at a tertiary center between 2012 and 2025. Cases were classified as isolated or non-isolated CDH based on the prenatal or postnatal presence of additional structural anomalies or molecular finding. Fetuses undergoing fetal endoscopic tracheal occlusion (FETO) were excluded. Prenatal imaging, genetic evaluation, and neonatal outcomes were compared between groups.

RESULTS

A total of 293 fetuses were included; 176 (60%) had non-isolated CDH and 117 (40%) isolated CDH. Baseline demographics were similar between groups. Non-isolated cases were more likely to undergo genetic counseling and testing ($p < 0.05$). Initial observed-to-expected lung-to-head ratio (o/e LHR) was lower in non-isolated cases (35.9 ± 15.7 vs 44.2 ± 18.6 , $p = 0.014$). Distribution of CDH defect type differed significantly ($p = 0.003$), with type C defects more common in non-isolated CDH. Despite these differences, neonatal outcomes were similar. ECMO use was higher among isolated cases but not statistically significant (43.3% vs 36.4% , $p = 0.34$). Pulmonary hypertension was common in both groups, with a non-significant trend toward higher prevalence in non-isolated CDH (86.1% vs 77.8% , $p = 0.16$), and survival rates were comparable (68.7% vs 69.3% , $p = 0.92$).

FIGURE 1. PRENATAL LUNG MEASUREMENT

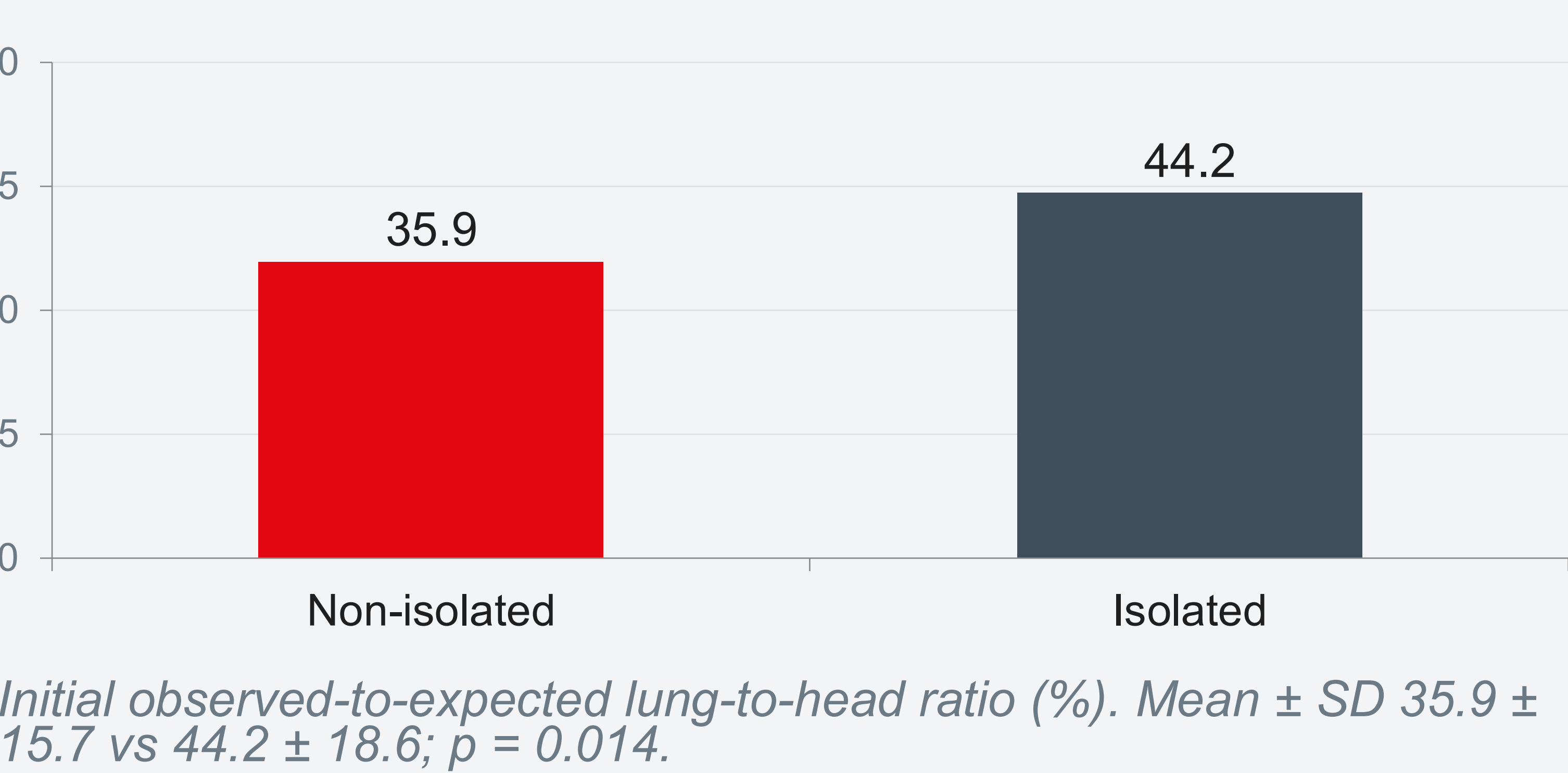
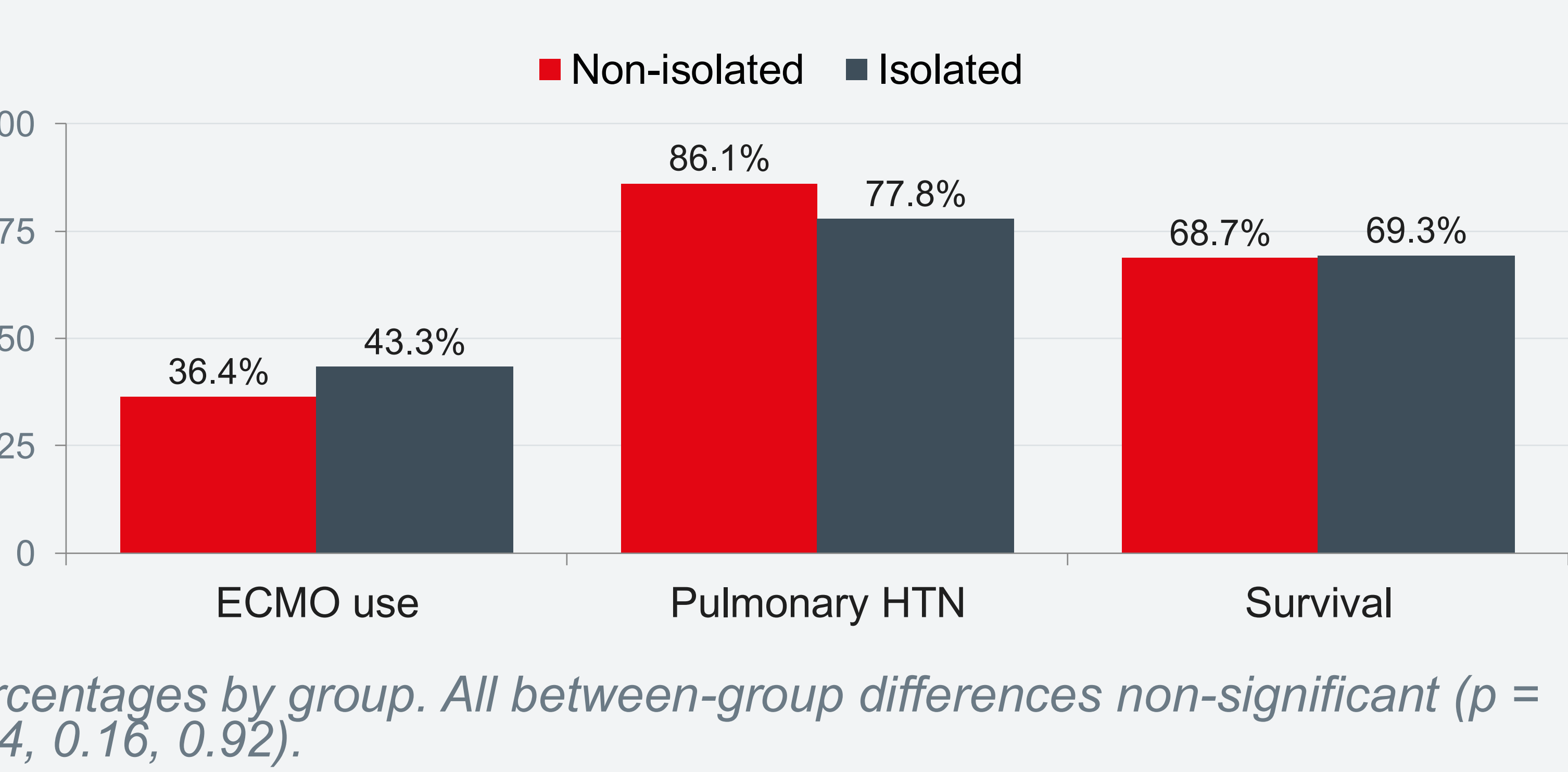


FIGURE 2. NEONATAL OUTCOMES



AT A GLANCE

293
fetuses with CDH included

60% / 40%
non-isolated vs isolated

35.9 vs 44.2
initial o/e LHR (%), $p = 0.014$

68.7 vs 69.3
survival (%), $p = 0.92$

CONCLUSIONS

Non-isolated CDH is associated with lower prenatal lung measurements and increased genetic evaluation
But neonatal outcomes including survival and ECMO use were similar.