

Congenital lung malformations in fetuses with congenital diaphragmatic hernia

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Objective

To describe the prevalence and prenatal characteristics of congenital lung malformations (CLM) in fetuses with congenital diaphragmatic hernia (CDH).

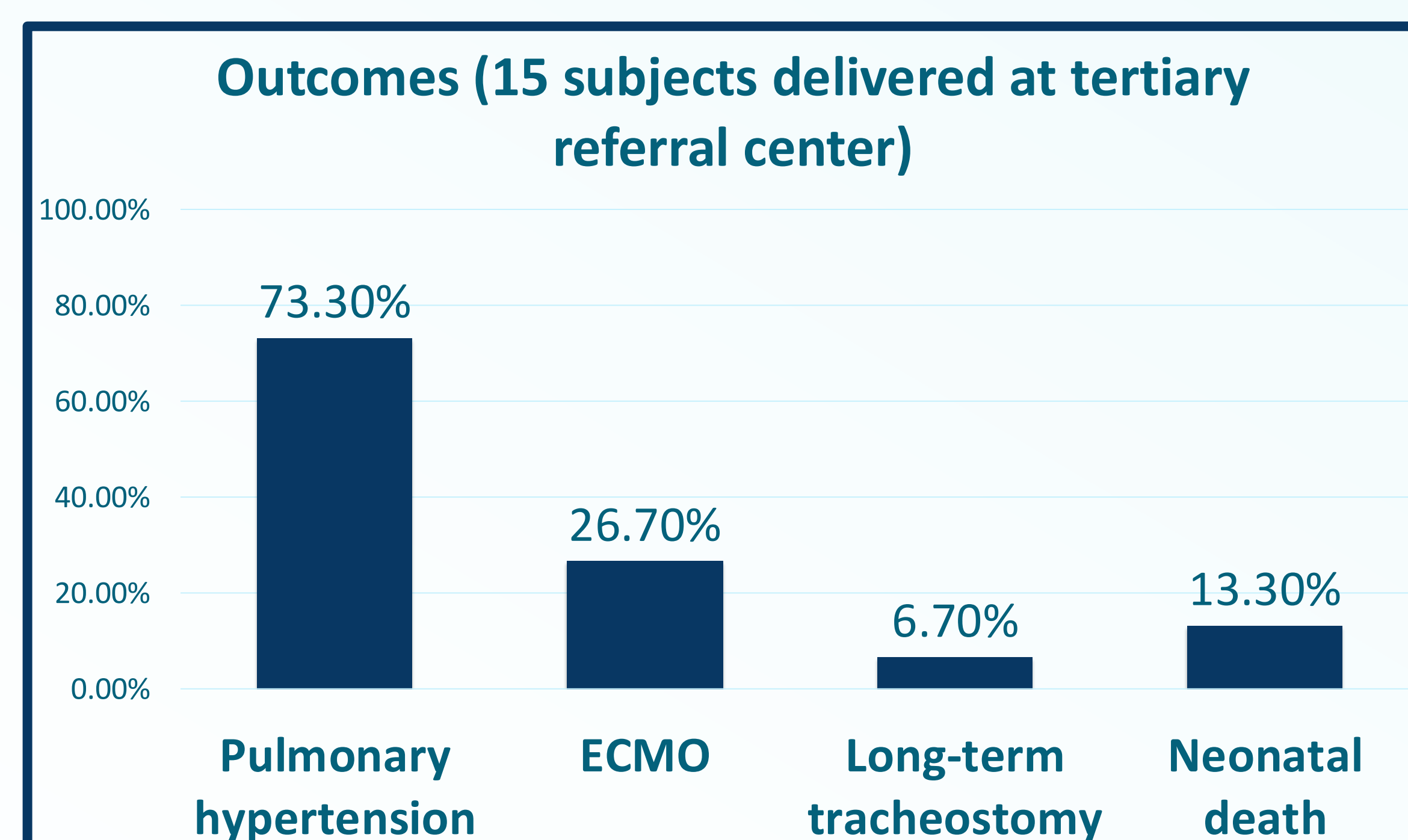
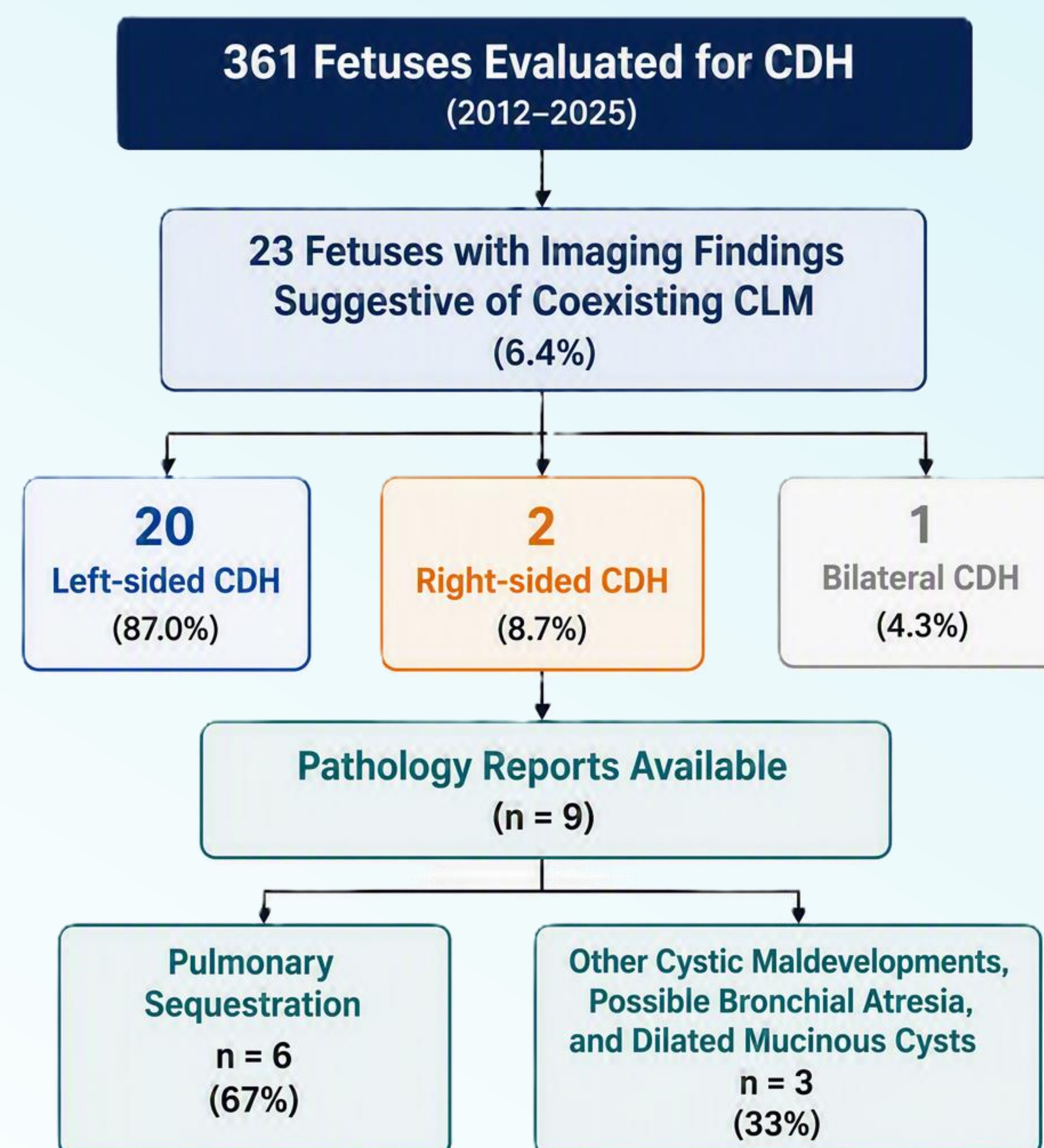
Introduction

Congenital diaphragmatic hernia (CDH) is associated with significant morbidity due to pulmonary hypoplasia. It may present as an isolated defect or with associated structural, chromosomal, or genetic anomalies. Previous cohort studies have reported a co-occurrence of CDH and bronchopulmonary sequestrations of 3.4-3.9%.^{1,2} In this study, we analyzed the co-occurrence of lung masses and CDH in patients seen at a tertiary referral center. We examined several factors related to disease detection, progression, and prognosis in patients with both CDH and lung masses.

Methods

- This retrospective cohort study included fetuses evaluated for CDH at a tertiary referral center from 2012 to 2025.
- Cases with imaging findings suggestive of co-occurring CLM were identified via prenatal ultrasound and/or fetal MRI reports.
- This included findings such as echogenic lung masses, cystic lesions, or systemic arterial supply.
- Prenatal imaging findings, genetic testing results, pathology reports, and neonatal outcomes were analyzed.

Results



91.3% (21/23) of CLMs were ipsilateral to the CDH

Median observed-to-expected total fetal lung volume was 33% (IQR 25-37%)

Three of the 23 (13%) patients underwent FETO

There was one incidence of intrauterine fetal demise

22% of subjects who underwent genetic testing had abnormal results

Maternal Demographics

- Average age: 30
- 60.87% White, 17.39% Black or African American, 4.35% Asian, 4.35% other or multiple races selected, 13.04% unable to obtain
- 73.91% Not Hispanic or Not Latino, 21.74% Hispanic or Latino, 4.35% unable to obtain

Conclusion

- Imaging findings suggestive of CLM were identified in approximately 6% of fetuses with CDH in this cohort.
- CLMs were predominantly ipsilateral to the diaphragmatic defect. This finding could suggest a possible developmental relationship between the two anomalies.
- CLMs were most commonly identified as pulmonary sequestration on both prenatal imaging and postnatal pathology reports.

Limitations

- Only 9 of 23 cases of CLM had available pathology reports, limiting the ability to confirm imaging findings.
- The study did not directly compare CDH+CLM patients with a matched group of CDH patients without CLM. Therefore, further comparative studies are needed to determine the clinical significance and prognostic impact of this association.

References

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