Primary Congenital Pulmonary Hypoplasia: A Case Report and Review of Literature

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Pulmonary hypoplasia caused by congenital acinar dysplasia is an extremely rare maldevelopment of the lungs and carries a high mortality rate. We report a case of severe primary pulmonary hypoplasia with absence of alveoli. The patient was a 1-hour-old liveborn male neonate and was twin “B” of a diamniotic-dichorionic twin pregnancy. The mother is a 34-year-old, G3P2 (2002), with a history of gestational diabetes mellitus. On routine antenatal ultrasound, twin B was diagnosed with an unknown cardiac defect. At 34.4 weeks of gestation, both twins were delivered via cesarean section due to premature rupture of membranes and breech position of twin B. Apgar scores were 7/8 and 1/1, at 1 and 5 minutes for twin A and B, respectively. Resuscitative measures were initiated but were unsuccessful. Of note, the second child was diagnosed with DiGeorge syndrome and an unknown cardiac defect. Postmortem examination revealed acrocyanosis, bilateral pulmonary hypoplasia with a combined lung weight of 15.2 g (N = 34.1 ± 9.4 g) and a lung-to-birth weight ratio of 0.97% (N > 1.2%). Microscopic examination revealed severe bilateral pulmonary hypoplasia with almost complete absence of alveoli. The bronchial branching pattern was normal. There were no other congenital anomalies identified. Primary congenital pulmonary hypoplasia is rare. Due to the rarity of this condition, it is poorly documented on what is the cause. There are reports of familial cases which suggest an autosomal recessive mode of inheritance. Further research on the etiology, such as the status of gene mutation or loss of heterozygosity, is warranted in the future.