Abstract

Jeune syndrome was described in 1956 as thoracic dysplasia Asphyxiating (dTAs). Its incidence is 1 per 100,000 live births. In Venezuela the first case was described by Urdaneta Carruyo in 1986. It is a member of the family of the short-rib polydactyly syndromes. Respiratory failure, secondary to pulmonary hypoplasia, is the most common cause of death. Because dTA is a rare syndrome with a high mortality presentation, we were motivated to report the case of a 11-year-old girl (diagnosed at 10 months of age), with a narrow thorax, and variable limb shortness. Thoracoplasty was performed at 9 years of age in order to achieve thoracic expansion. The patient died at 11 years of age. Conclusions: Jeune syndrome is a rare entity, which compromises the life of the patient; it has autosomal recessive inheritance pattern and requires genetic counseling to parents and a multidisciplinary management.

Keywords

Asphyxiating thoracic dysplasia, skeletal dysplasia, pulmonary hypoplasia