Chromosome 22q11 deletion in a patient with pulmonary atresia, intact ventricular septum, and confluent branch pulmonary arteries

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Ver ResearcherID y ORCID

CARDIOLOGY IN THE YOUNG
Volumen: 28
Número: 3
Páginas: 467-470
DOI: 10.1017/S104795111700227X
Fecha de publicación: MAR 2018
Tipo de documento: Article
Ver impacto de la revista

Abstract

In this study, we report a patient with pulmonary atresia with intact ventricular septum (PA/IVS), confluent pulmonary arteries supplied by an arterial duct, and chromosome 22q11.2 microdeletion. The 22q11.2 deletion syndrome has been associated with anomalies of the outflow tracts, such as tetralogy of Fallot with either pulmonary stenosis or atresia, but we are aware of a solitary case described with pulmonary atresia when the ventricular septum is intact. The presence of genetic malformations can have long-term co-morbidities. By describing our patient, we aim to create awareness of this rare association.

Palabras clave

Palabras clave de autor: Chromosome 22q11 deletion; pulmonary atresia; intact ventricular septum; confluent branch pulmonary arteries; right aortic arch
KeyWords Plus:DEFECTS

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Editorial

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Información de la revista

- **Impact Factor:** Journal Citation Reports

Categorías / Clasificación

Áreas de investigación: Cardiovascular System & Cardiology; Pediatrics
Categorías de Web of Science: Cardiac & Cardiovascular Systems; Pediatrics

Información del documento

Idioma: English
Número de acceso: WOS:000429627500019
ID de PubMed: 29233233
ISSN: 1047-9511
eISSN: 1467-1107