Rare Association Of Severe Hypoplasia Of The Abdominal

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Rare Association Of Severe Hypoplasia
Rare association of severe hypoplasia of the abdominal aorta with imperforate anus, colonic atresia, and choledochal cyst. Komuro H(1), Takahashi MI, Matoba K, Hori T, Hirai M, Gotoh C, Kaneko M. Author information: (1)Department of Pediatric Surgery, Graduate School of Comprehensive Human Sciences, University of Tsukuba, 1-1-1 Tennodai, 305-8575 Tsukuba, Ibaraki, Japan. hiro-kom@md.tsukuba.ac.jp

Rare association of severe hypoplasia of the abdominal ...
Hypoplasia of the abdominal aorta (HAA) is a rare condition that causes marked hypertension. Although multiple etiologies have been postulated for HAA, congenital structural anomalies are rarely observed except in cases associated with some hereditary syndromes.

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We report in a 17 years old patient a rare case of severe hypoplasia with gelatinous bone marrow transformation whose specific etiology remains mysterious in spite of a detailed diagnostic and etiologic assessment. Marrow gelatinous transformation (also called serous fat atrophy or starvation marrow) is characterized by the association of marrow hypoplasia and interstitial infiltration of a...

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Pontocerebellar hypoplasias (PCH) are a group of rare heterogeneous conditions characterized by prenatal development of an abnormally small cerebellum and brain stem, which is usually associated with profound psychomotor retardation.

Pontocerebellar Hypoplasia - NORD (National Organization ...
UCH is a rare finding encompassing a spectrum ranging from complete aplasia (unilateral absence of one cerebellar hemisphere) through subtotal asymmetrical hypoplasia to mild asymmetry in the size of the cerebellar hemispheres. 1 In our experience, minor asymmetry of the cerebellar hemispheres is occasionally seen as an incidental finding without clinical significance whereas severe UCH is expected to be of clinical relevance.

Outcome of severe unilateral cerebellar hypoplasia ...
Ulnar hypoplasia–split foot syndrome is characterised by the association of severe ulnar hypoplasia, absence of fingers two to five, and split-foot. ... Orphanet is a European reference portal for information on rare diseases and orphan drugs. Access to this database is free of charge.

Ulnar hypoplasia lobster claw deformity of feet - Rare disease
A rare case of a 14-year-old child with congenital mitral insufficiency secondary to hypoplasia of the posterior leaflet is reported. Echocardiography revealed the almost complete absence of the posterior mitral leaflet, which determined massive regurgitation. At surgical inspection the posterior leaflet was almost completely absent, represented only by tags of fibrous tissue that strictly ...

Severe Hypoplasia of the Posterior Mitral Leaflet - The ...
NORD, a 501(c)(3) organization, is the leading patient advocacy organization dedicated to improving the lives of individuals and families living with rare diseases.

Home - NORD (National Organization for Rare Disorders)
The rare association of pontocerebellar hypoplasia with anterior horn cell involvement has been classified as pontocerebellar hypoplasia type 1. Its classic phenotype is usually severe. However, the pontocerebellar hypoplasia type 1 may have wider variability in clinical and radiological features. There may be a genetic heterogeneity as well.

Pontocerebellar hypoplasia type 1 with a milder phenotype ...
In conclusion, severe hypoplasia of the posterior mitral leaflet is a rare cause of mitral regurgitation. It can be successfully corrected by a reductive ring annuloplasty in presence of a sufficiently long and mobile anterior leaflet. Nonetheless, the long-term durability of this repair remains uncertain.

Severe Hypoplasia of the Posterior Mitral Leaflet ...
Hypoplasia of the thoracic and abdominal aorta is an extremely rare vascular pathology. The most common clinical manifestation is severe uncontrolled hypertension in adolescents and young adults. Medical treatment alone can decrease blood pressure, but often very high doses of antihypertensive drugs are needed.

Severe diffuse hypoplasia of the aorta associated with ...
Pulmonary hypoplasia is incomplete development of the lungs, resulting in an abnormally low number or size of bronchopulmonary segments or alveoli.A congenital malformation, it most often occurs secondary to other fetal abnormalities that interfere with normal development of the lungs. Primary pulmonary hypoplasia is rare and usually not associated with other maternal or fetal abnormalities.

Pulmonary hypoplasia - Wikipedia
Pontocerebellar hypoplasia type 2 (PCH2) is a rare condition that affects the development of the brain.Signs and symptoms vary but may include microcephaly, developmental delay with lack of voluntary motor development, intellectual disability and movement disorders (i.e. chorea, dystonia, and spasticity).Affected people may also experience dysphagia (difficulty swallowing), impaired vision ...