Porencephaly

Porencephaly its a intracranial cysts

It is a rare congenital disorder that results in cystic degeneration and encephalomalacia and the formation of porencephalic cysts. The term is used variably among radiologists with its broadest definition being a cleft or cystic cavity within the brain, and its more narrow definition being a focal cystic area of encephalomalacia that communicates with the ventricular system and/or the subarachnoid space.

Etiology

Exon mutations of the COL4A1 genes are responsible for a broad spectrum of cerebral, ocular, and systemic manifestations. We describe here the phenotype of a likely pathogenic gene variant, p.Gly743Val, which is responsible for a missense mutation in the COL4A1 gene exon 30 in a three generation family with severe hypermetropia and highly penetrant porencephaly in the absence of systemic manifestations. This report highlights both the broad spectrum of COL4A1 mutations and the yield of testing the COL4A1 gene in familial ophthalmological and brain disorders [1)].

Direct injury to neural tissue from hematoma: once hemorrhage resorbs may leave patient with porencephaly or cystic lesions.

Ecephaloclastic insult (e.g. intrauterine infections and ischemia) often caused by vascular infarcts or following intracerebral hemorrhage or penetrating trauma (including repeated ventricular punctures).

Serial ventricular taps for periventricular-intraventricular hemorrhage (PIVH).: May be a viable short-term option for those infants who cannot tolerate LPs or in whom there is obstruction to CSF flow in the lumbar subarachnoid space (e.g. due to spinal subdural hematoma from previous LP). However it is not desirable for long-term use because of repeated trauma to brain (risk of porencephaly) and risk of intracerebral, intraventricular, or subdural hemorrhage.

If continued taps are likely (i.e., large hemorrhage, or rapid recurrence of intracranial hypertension as determined by palpation of fullness of anterior fontanelle (AF) following several taps), the acceptable options include:

1. continuing serial LPs

2. percutaneous ventricular taps: not recommended for more than a few treatments as it causes porencephaly

Types

There are two types of porencephaly:

Type I: unilateral, due to hemorrhage or ischemia.

Type II: bilateral, due to neuronal migration disruption.

Sometimes associated with Dandy Walker malformation, Agenesis of the corpus callosum.

Diagnosis

Porencephalic cysts are diagnosed radiologically

Differential diagnosis

Schizencephaly: cleft lined with cortical grey matter (often abnormal, may have polymicrogyria). This distinguishes it from porencephaly

Treatment

Management depends on the clinical manifestation.

Endoscopic fenestration of the cyst membrane into the lateral ventricle can be successful in reducing cyst volume and improving mass effect [2) 3)].

Case reports

A case of co-existence of colpocephaly with porencephaly diagnosed incidentally in a 54-year-old male presenting with subtle cognitive and neurologic abnormalities. Neuropsychological assessment revealed weaknesses in executive functions, processing speed, and language. This is the only reported case of dual incidental findings of porencephaly and colpocephaly in an adult [4)].

Porencephaly and Intracranial Calcifications in a Neonate [5)].

A porencephalic cyst presenting with nondominant parietal lobe symptoms in adulthood. Wynne et al. hypothesized that a membrane between the cyst and ventricle allowed formation of a 1-way valve that led to slowly progressive cyst enlargement, eventually causing mass effect and nondominant parietal lobe symptoms. Endoscopic fenestration of the cyst membrane into the lateral ventricle was successful in reducing cyst volume and improving mass effect [6)].

1)
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