Alagille syndrome

Alagille syndrome (AGS) is an autosomal dominant arteriodysplastic syndrome with multiple organ system involvement caused by a mutation in the Jagged1 gene.

Intracranial hemorrhage is one of the many complications observed in this patient population. While there are multiple case reports in the literature reviewing the spectrum of cerebrovascular events and abnormalities.

Although intracranial hemorrhage has frequently been found responsible for mortality in adult patients with Alagille syndrome (AGS), no specific underlying cause has been identified.

The genomic abnormalities are the Jagged1 gene and the Notch receptor signaling pathway which may reveal elements of the pathophysiology involved in aneurysm formation and rupture.

In light of the increased incidence of intracranial hemorrhage in AGS and the possible link to aneurysmal subarachnoid hemorrhage, establishing the incidence of intracranial aneurysms in AGS and the role of screening these patients is indicated

Case reports

2012

O'Connell et al. describe the first case of superior cerebellar aneurysm rupture causing WFNS grade 1 subarachnoid haemorrhage in a 17-year-old girl. The clinical condition and management of this rare occurrence is discussed with a review of literature. [1].

2006

A 21-year-old female with a known history of Alagille syndrome (AGS) who was found to have a basilar terminus aneurysm without evidence of rupture. Prior to intervention, the patient's hospital course became complicated by multiple medical problems associated with AGS. Subsequently, the patient had an acute neurological decline. An unenhanced CT of the head demonstrated diffuse subarachnoid hemorrhage, intraparenchymal hematoma and intraventricular hemorrhage. This is the third reported case of documented aneurysmal subarachnoid hemorrhage in a patient with AGS. The authors present a brief review of the vascular abnormalities both intracranial and systemic seen in AGS. [2].

2004

Schlosser et al. describe the case of severe subarachnoid hemorrhage in a 30-year-old woman harboring five intracranial aneurysms and multiple peripheral vascular anomalies. To evaluate a possible higher incidence of intracranial aneurysms, a study of the cerebral vasculature in all AGS patients by using noninvasive imaging techniques should be considered [3].

Cowan et al. present a case in which a posterior communicating artery (PCoA) infundibulum progressed into an aneurysm in a patient with Alagille syndrome (arteriohepatic dysplasia). The 3-mm PCoA infundibulum had been noted on angiography studies obtained 5 years earlier, prior to clip occlusion of a basilar tip aneurysm. Recently, the patient presented to the emergency department with the sudden onset of headache and decreased mental status. A computerized tomography scan of the head with three-dimensional angiography revealed no gross subarachnoid hemorrhage, but did demonstrate a 5-mm PCoA aneurysm. Lumbar puncture demonstrated xanthochromia and a large quantity of red blood cells. The patient underwent open surgery for aneurysm clip occlusion and obtained a good recovery. This case illustrates the small but growing number of examples of infundibulum progression. It also indicates the need for a close follow up in patients with congenital abnormalities that may pose an increased risk for what has traditionally been considered a benign lesion [4].

1)
O'Connell D, Kaliaperumal C, Fanning N, Wyse G, Kaar G. Superior cerebellar aneurysm causing subarachnoid haemorrhage in a 17-year-old with alagille syndrome. Br J Neurosurg. 2012 Apr;26(2):287-9. doi: 10.3109/02688697.2011.614022. Epub 2011 Oct 25. PubMed PMID: 22026469.
2)
Tumialán LM, Dhall SS, Tomak PR, Barrow DL. Alagille syndrome and aneurysmal subarachnoid hemorrhage. Case report and review of the literature. Pediatr Neurosurg. 2006;42(1):57-61. Review. PubMed PMID: 16357504.
3)
Schlosser HG, Kerner T, Woiciechowsky C, Benndorf G. Multiple cerebral aneurysms and subarachnoid hemorrhage in a patient with Alagille syndrome. AJNR Am J Neuroradiol. 2004 Sep;25(8):1366-7. PubMed PMID: 15466333.
4)
Cowan JA Jr, Barkhoudarian G, Yang LJ, Thompson BG. Progression of a posterior communicating artery infundibulum into an aneurysm in a patient with Alagille syndrome. Case report. J Neurosurg. 2004 Oct;101(4):694-6. PubMed PMID: 15481729.