

Porencephalic cyst in adult

Stefano Tambuzzi¹ , Guendalina Gentile¹ , Riccardo Zoja¹ 

How to cite: Tambuzzi S, Gentile G, Zoja R. Porencephalic cyst in adult. *Autops Case Rep* [Internet]. 2022;12:e2021351. <https://doi.org/10.4322/acr.2021.351>

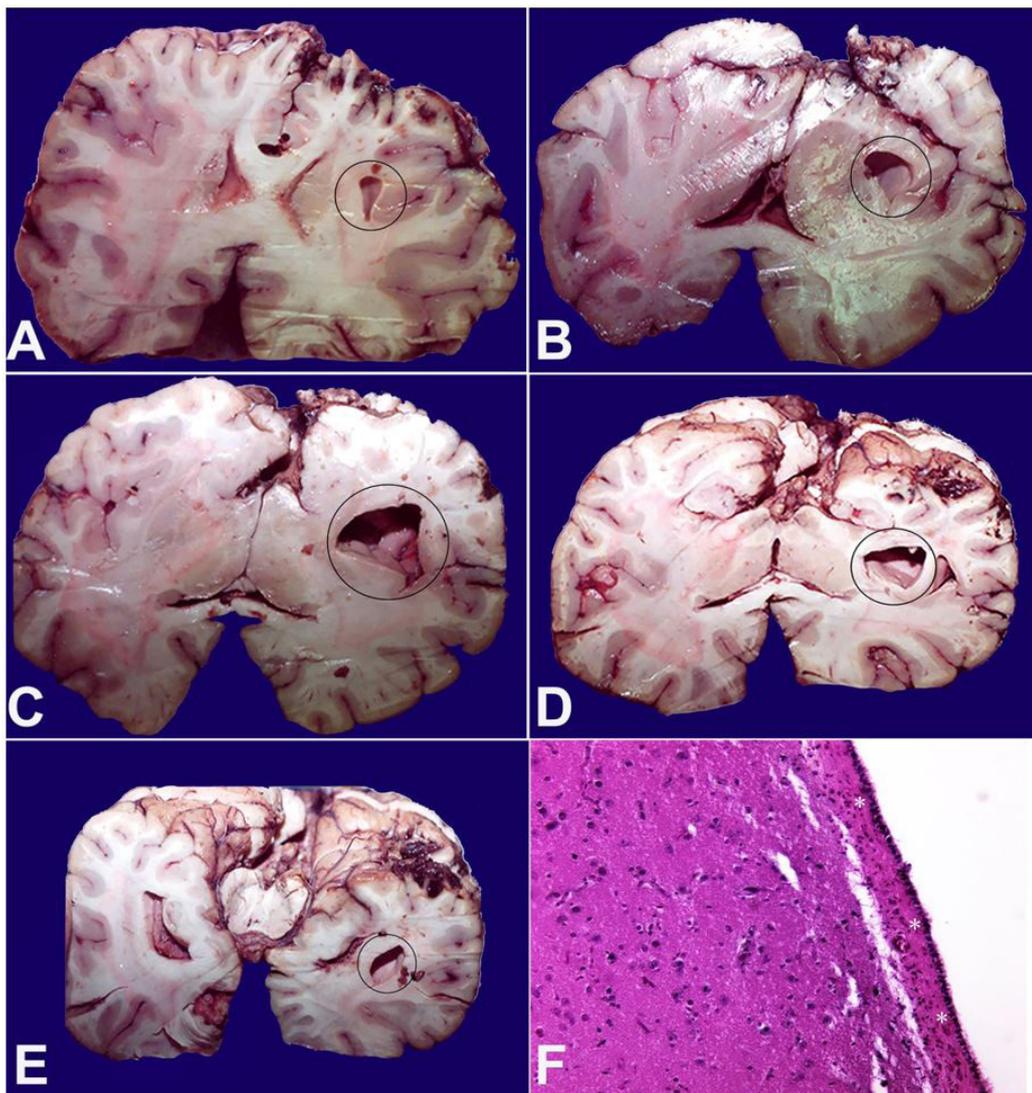


Figure 1. A-E – Macroscopic view of left craniocaudal coronal encephalic sections with evidence of the porencephalic cyst within the cerebral white matter (marked with circles); F – Microscopic view of the cystic cavitation at the left temporal lobe, which wall appears regular and smooth with sub ependymal lining (asterisk) (H&E, 200x).

¹Università degli Studi di Milano, Dipartimento di Scienze Biomediche per la Salute, Sezione di Medicina Legale e delle Assicurazioni – Laboratorio di Istopatologia Forense e Microbiologia Medico Legale, Milano, Italy

Porencephalic cyst is a rare entity affecting the central nervous system, whose etiopathogenesis has not yet been clearly defined. It is a cavity within the cerebral hemisphere¹ with smooth wall lined by gliotic or spongiotic white matter,² which contains cerebrospinal fluid.³ The cavity usually communicates directly with the ventricular system and is often seen in territories supplied by the cerebral arteries.² In other cases it may be separated by a thin layer of brain tissue and be covered externally by the arachnoid.⁴ The cavities may vary greatly in size and location, being cortical or sub-cortical, unilateral or bilateral, single or multiple.⁵ Porencephalic cysts can be classified as congenital or acquired: it has been suggested that they are caused by a disturbance of vascular supply leading to cerebral degeneration.⁶ Congenital porencephalic cysts result from intra-uterine vascular injury leading to cerebral ischemia or intra-parenchymal hemorrhage. Intra-uterine infectious injury by a virus like cytomegalovirus can also give rise to congenital porencephalic cysts.⁷ A further cause has been recognized in alcohol taken during pregnancy, as distortions in the formation of the brain with excessive plication and invaginations of the fetuses' brain walls in alcoholic mothers have been demonstrated.⁸ Acquired porencephalic cysts are secondary to injury later in life due to trauma, surgery, ischemia, or infection.⁹ De novo or inherited heterozygous mutations in COL4A1, which encodes the type IV a1 collagen chain, have been reported in individuals with porencephaly.⁶ Finally, more recently, mutations in the tubulin alpha 1a gene (TUBA1A) have also been related to the onset of this pathological condition.¹⁰

From an epidemiological point of view, porencephalic cysts are described in newborns with an estimated incidence of 3.5 per 100,000 live births,³ and only very rarely they are reported in adults.

The symptoms that characterize porencephaly are numerous¹¹ and vary with the neuronal loss that precedes the formation of cysts, or to the mass effect of the cyst itself. In the first case, cerebral palsy or convulsions, mental retardation, and learning difficulties are observed, while in the second case, the mass effect occurs with increased intracranial pressure or focal deficits attributable to localization in the involved lobe.^{5,12,13} In this case, epilepsy, visual, speech, and hearing impairment, rhinorrhea, or otorrhea of the cerebrospinal fluid⁴ typically occur and, last but

not least, may be also observed psychiatric disorders (schizophrenia, psychosis).¹⁴

For the diagnosis of porencephaly, the electroencephalogram may help but the findings are not specific, and radiological techniques are necessary. A cerebral CT scan shows a hypodense intracranial cyst, with a well-defined border and central attenuation due to the cerebrospinal fluid. Mass effect on the adjacent parenchyma is not usually observed, but very large cysts can cause it locally. Brain MRI shows a well-defined, white matter-coated brain cyst with or without gliosis, with the cerebrospinal fluid signal. Fetal MRI is also helpful to differentiate this entity from extra-axial lesions such as arachnoid cysts but also rare tumors.¹³ Neurosurgical treatment is reserved for symptomatic patients, especially those suffering from drug-resistant neurological complications, such as epilepsy.¹⁵ In these cases, the therapeutic approaches may consist of: fenestration of the cysts in the lateral ventricle with permanent endovascular occlusion with a balloon,¹⁶ cortical resections,¹⁷ lobectomies,¹⁸ subtotal or total hemispherectomy,¹⁹ as well as a ventriculoperitoneal shunt in the case of hydrocephalus.²⁰ There is no literature discussing therapy for asymptomatic porencephaly which only requires monitoring and surveillance. In some exceptional asymptomatic or pauci-symptomatic cases, the first diagnosis may only be reached upon the autopsy examination.

Figures 1A-1E refer to the case of a 45-year-old man found dead in his home, prone on the bathroom floor, with no visible signs of violence on the body. To clarify the causes of death, he was subjected to a clinical autopsy. During the interview with the deceased's relatives, it emerged that the man had been suffering for years from recurrent episodes of mild headache that he had never wanted to investigate from a medical point of view. No other pathological conditions were documented, nor the drugs or alcohol abuse.

Upon dissection of the brain (weight 1214 g [mean reference range 1366 g]), a slight asymmetry of the cerebral hemispheres was noted, with a prevalence of the left one. The leptomeninges were smooth and shiny, with no focal alterations; the vessels were marked dilated and congested. At serial coronal sections, a large cavitated formation was present in the white matter of the entire left cerebral hemisphere, in the superolateral position to the ventricle. It extended from

the middle third of the frontal lobe to the middle third of the occipital lobe, reaching a maximum diameter of 2.5 cm in the left temporal area. In addition, the occipital lobe of the left cerebral hemisphere showed an abnormal supernumerary convolution. In Figure 1F, the surface of the cystic cavitation wall, which appears regular and smooth with a sub ependymal coating, can be observed.

At the end of the autopsy, the cause of death was identified in acute circulatory failure in a subject suffering from severe coronary artery disease and porencephaly. Therefore, the finding of the porencephalic cyst was incidental, and the origin of this disease was likely attributed to a congenital cause. Indeed, the man had always had good health, except only for recurrent episodes of mild headache.

We deemed this finding of interest both for the rarity of the detected cerebral anomaly and for the man's survival until adulthood in the absence of severe symptoms. Indeed, the first diagnosis occurred only *postmortem* and the histopathological analysis confirmed the porencephalic nature of the large cerebral cavitated defect.

Keywords

Autopsy; Porencephaly.

REFERENCES

1. Wynne D, Abdul Jalil MF, Dhillon R. Endoscopic fenestration of a symptomatic porencephalic cyst in an adult. *World Neurosurg*. 2020;141:245-6. <http://dx.doi.org/10.1016/j.wneu.2020.06.092>. PMID:32569761.
2. Oommen AT, Sethy G, Minz NT, Patra J, Panda SS. Unusual presentation of porencephalic cyst in an adult. *J Clin Diagn Res*. 2017;11(2):OD12-3. <http://dx.doi.org/10.7860/JCDR/2017/22654.9374>. PMID:28384919.
3. Qureshi A, Jehangir A, York EP. Porencephalic cyst: a rare cause of new-onset seizure in an adult. *J Community Hosp Intern Med Perspect*. 2018;8(2):92-3. <http://dx.doi.org/10.1080/20009666.2018.1454788>. PMID:29686797.
4. Ryzenman JM, Rothholtz VS, Wiet RJ. Porencephalic cyst: a review of the literature and management of a rare cause of cerebrospinal fluid otorrhea. *Otol Neurotol*. 2007;28(3):381-6. <http://dx.doi.org/10.1097/mao.0b013e31802ead9e>. PMID:17325618.
5. Ho SS, Kuzniecky RI, Gilliam F, Faught E, Bebin M, Morawetz R. Congenital porencephaly: MR features and relationship to hippocampal sclerosis. *AJNR Am J Neuroradiol*. 1998;19(1):135-41. PMID:9432171.
6. Yoneda Y, Haginoya K, Arai H, et al. De novo and inherited mutations in COL4A2, encoding the type IV collagen $\alpha 2$ chain cause porencephaly. *Am J Hum Genet*. 2012;90(1):86-90. <http://dx.doi.org/10.1016/j.ajhg.2011.11.016>. PMID:22209246.
7. Kokkinos V, Garganis K, Kontogiannis K, Zountsas B. Hemispherotomy or lobectomy? The role of presurgical neuroimaging in a young case of a large porencephalic cyst with intractable epilepsy. *Pediatr Neurosurg*. 2011;47(3):204-9. <http://dx.doi.org/10.1159/000330546>. PMID:22122950.
8. Kovetskiĭ NS. Etiology of porencephalia and other developmental disorders of the brain in alcoholic embryopathy. *Zh Nevropatol Psikhiatr Im S S Korsakova*. 1989;89(2):117-22. PMID:2728712.
9. Osborn AG. *Brain*. Vol. 1. Salt Lake City, Utah: Amirsys; 2004. p. 7-36. Porencephalic cyst - Choroid plexus cyst. Diagnostic imaging.
10. Sato T, Kato M, Moriyama K, et al. A case of tubulinopathy presenting with porencephaly caused by a novel missense mutation in the TUBA1A gene. *Brain Dev*. 2018;40(9):819-23. <http://dx.doi.org/10.1016/j.braindev.2018.05.012>. PMID:29907476.
11. Hino-Fukuyo N, Togashi N, Takahashi R, et al. Neuroepidemiology of porencephaly, schizencephaly, and hydranencephaly in Miyagi Prefecture, Japan. *Pediatr Neurol*. 2016;54:39-42.e1. <http://dx.doi.org/10.1016/j.pediatrneurol.2015.08.016>. PMID:26545857.
12. Park HJ, Lee SH, Park HJ, Shin SH. A porencephalic cyst formation in a 6-year-old female with a functioning ventriculoperitoneal shunt: a case-based review. *Childs Nerv Syst*. 2018;34(4):611-6. <http://dx.doi.org/10.1007/s00381-018-3725-x>. PMID:29380111.
13. Abergel A, Lacalm A, Massoud M, Massardier J, des Portes V, Guibaud L. Expanding porencephalic cysts: prenatal imaging and differential diagnosis. *Fetal Diagn Ther*. 2017;41(3):226-33. <http://dx.doi.org/10.1159/000447740>. PMID:27409647.
14. Noyan OC, Salcini C, Talu BS, Eryilmaz G. Porencephalic cyst and late onset brief psychotic disorder. *BMJ Case Rep*. 2016;bcr2016215098. <http://dx.doi.org/10.1136/bcr-2016-215098>. PMID:27053544.
15. Curry CJ, Lammer EJ, Nelson V, Shaw GM. Schizencephaly: heterogeneous etiologies in a population of 4 million California births. *Am J Med Genet A*. 2005;137(2):181-9. <http://dx.doi.org/10.1002/ajmg.a.30862>. PMID:16059942.

16. Phillips J, Roberts G, Brennan P, Staunton H. Minimally-invasive endovascular occlusion therapy in two adults for seizures associated with congenital porencephalic cysts. *J Epilepsy*. 1998;11(6):374-82. [http://dx.doi.org/10.1016/S0896-6974\(98\)00042-5](http://dx.doi.org/10.1016/S0896-6974(98)00042-5).
17. Iida K, Otsubo H, Arita K, Andermann F, Olivier A. Cortical resection with electrocorticography for intractable porencephaly-related partial epilepsy. *Epilepsia*. 2005;46(1):76-83. <http://dx.doi.org/10.1111/j.0013-9580.2005.28704.x>. PMID:15660771.
18. Burneo JG, Faught E, Knowlton RC, et al. Temporal lobectomy in congenital porencephaly associated with hippocampal sclerosis. *Arch Neurol*. 2003;60(6):830-4. <http://dx.doi.org/10.1001/archneur.60.6.830>. PMID:12810487.
19. Chugani HT, Asano E, Juhász C, Kumar A, Kupsky WJ, Sood S. "Subtotal" hemispherectomy in children with intractable focal epilepsy. *Epilepsia*. 2014;55(12):1926-33. <http://dx.doi.org/10.1111/epi.12845>. PMID:25366422.
20. Ramasamy SP, Chandrasekaran S, Jayakumar CR, Mallick AS. A case of Dandy Walker cyst with porencephaly. *Singapore Med J*. 1992;33(2):196-7. PMID:1621130.

This study was carried out at the Institute of Forensic Medicine of Milan, Italy

Authors' contributions: Stefano Tambuzzi and Guendalina Gentile equally contributed to this work. They devised the project and the main conceptual idea of the article, collected data, drafted the manuscript and performed literature research. Riccardo Zoja guarantor of the project and directed the study, devised the main conceptual idea of the article.

Ethics statement: The subject involved in this study underwent a judicial autopsy at the Institute of Legal Medicine of Milan in order to identify the cause of death. Data collecting, sampling and subsequent forensic analysis were authorized by the inquiring magistrate. Consequently, in accordance with the Italian law, ethical approval is not required in these cases. Publication of data is allowed when the case has been closed, but the anonymity of the subject must be guaranteed.

Conflict of interest: none

Financial support: none

Submitted on: September 24th, 2021

Accepted on: December 9th, 21021

Correspondence

Guendalina Gentile

Università degli Studi di Milano, Sezione di Medicina Legale e delle Assicurazioni – Laboratorio di Istopatologia Forense e Microbiologia Medico Legale

via Luigi Mangiagalli, 37, 20133 Milano, Italy

Phone: +3 (90) 250315721 / Fax: +3 (90) 250315724

guendalina.gentile@unimi.it