

Porencephaly may Present even in Elderly

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Sir,

Porencephaly is a rare disorder with a cyst or cavity formation in the cerebral hemispheres of the brain. No reliable data are available to date regarding its prevalence. It is commonly detected in early childhood. It can be detected prenatally by sonography. However, porencephaly may also be detected first time, even in elderly patients, either during the investigation of seizure disorders or may be detected incidentally.

A 60-year-old male was admitted to the emergency medical ward with a focal seizure with secondary generalization. His seizure was controlled with lorazepam and a loading dose of phenytoin (800 mg), and the patient was subsequently maintained on phenytoin with a daily dose of 300 mg/day. He had no history of neurological insults since childhood except one similar attack of seizure 2 years back. He was a manual laborer and had no formal education in school. His physical and mental development was normal. He was nonalcoholic, nondiabetic, and nonhypertensive. Family history did not reveal any neurological disorders. During the examination, the patient was alert, conscious, and cooperative. His speech and visions were normal. No neurodeficits were detected except asymmetric plantar responses. His computerized tomography (CT) scan of the brain revealed cystic encephalomalacia lined with the gliotic white matter in a left parietal region extending from the inner surface of the skull to the left lateral ventricle, and his

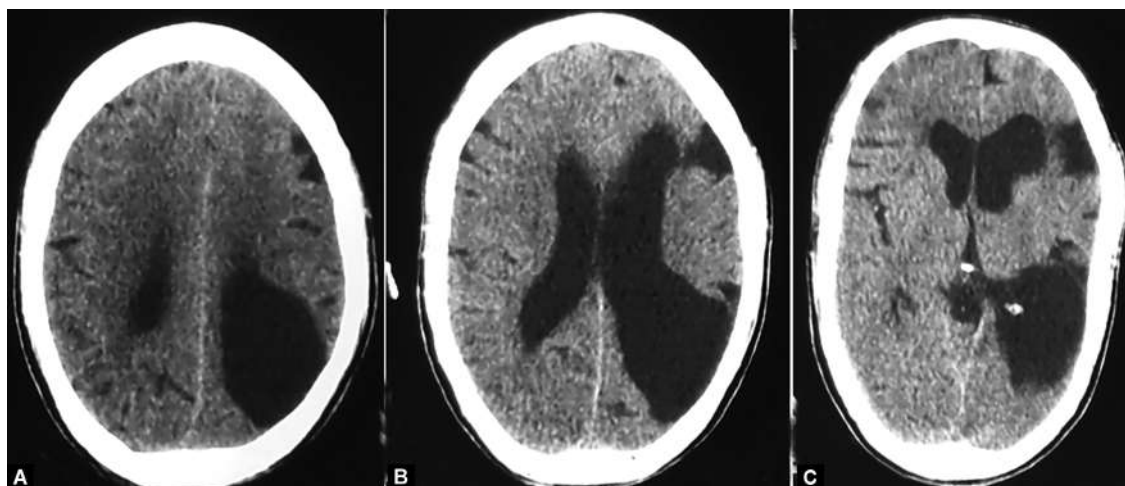
left lateral ventricle was comparatively dilated (Figs 1A to C). The patient had porencephaly likely of prenatal origin, as he had no major neurological insults since infancy.

Porencephaly is associated with a zone of encephalomalacia with the formation of a cerebrospinal fluid (CSF) filled cavity or cyst, which is lined by gliotic white matter. It can be located in any lobe and communicates with either subarachnoid space or ventricles, or both. Richard L Heschl coined the term 'porencephaly' in 1859 (Greek origin: "poros" meaning opening or passage) to describe a full thickness defect in the cerebral hemisphere forming a channel between subarachnoid space and lateral ventricle. Porencephaly develops prenatally during the second half of pregnancy or postnatally. It is either a developmental defect or may be acquired due to brain injury from infarction, hemorrhage, infection, or trauma. Different risk factors associated with porencephaly include thrombophilia like, protein C or protein S deficiency, factor V Leiden mutations, von Willebrand disease, neonatal alloimmune thrombocytopenia, maternal use of warfarin and other drugs like cocaine, congenital infections, the trauma of fetus during amniocentesis, antenatal abdominal trauma of mother, etc. Familial cases of porencephaly are associated with genetic mutations encoding α -1 and α -2 chains of type IV collagen (COL4A1 and 2).¹ COL4A1 mutation can also cause kidney, eyes, and cardiac or skeletal muscle defects, and both COL4A1 and COL4A2 mutations can cause a stroke. So, the affected infants of porencephaly and at-risk family members need neurologic, ophthalmologic, renal, and cardiac screening.

Porencephaly may remain asymptomatic throughout life or may have mild symptoms. Severe cases have hemiplegia, quadriplegia,

speech abnormalities, hearing and visual disturbances, seizure disorders, mental retardation, and other neurodeficits. While symptomatic cases are easily detected in early childhood, asymptomatic cases may remain undetected throughout life. Porencephaly may be detected in adults or the elderly with the onset of seizures or may be detected incidentally or during post-mortem examination.²

Differential diagnosis of porencephaly includes schizencephaly, hydranencephaly, multicystic encephalopathy, arachnoid cyst, ependymal cyst, focal encephalomalacia, mega cisterna magna (MCM), Dandy-Walker syndrome (DWS), etc. Schizencephaly is a CSF-filled cavity extending from the ventricle to the brain surface and is lined by gray matter (heterotopic), while porencephaly is lined by white matter (gliotic). Schizencephaly is usually unilateral but may be bilateral also. It is either "open-lipped" or "closed-lipped." In severe cases of bilateral open-lipped schizencephaly, both lateral ventricles communicate widely with extra-axial space. Other malformations which may be associated with schizencephaly include corpus callosum dysgenesis, absent septum pellucidum, septo-optic dysplasia, polymicrogyria, and microcephaly. For porencephaly, brain insult is in the mid or later part of gestation or postnatal, while for schizencephaly, insult is earlier. A relatively mature brain helps to develop glial scarring around the cyst in porencephaly. In hydranencephaly (bubble brain), brain hemispheres are replaced by fluid-filled sacs covered with thin membranes representing leptomeninges. It may be considered porencephaly in its extreme form. Rarely hydranencephaly is seen in Fowler syndrome, also known as proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome,



Figs 1A to C: Computerized tomography (CT) scan brain showing cystic encephalomalacia in the left parietal region extending from the inner surface of the skull to the left lateral ventricle

and is caused by mutations in Feline leukemia virus subgroup C cellular receptor family, member 2. Multicystic encephalopathy is associated with numerous loculated lacy pseudocysts within the white matter and cortex due to an extensive brain insult (e.g., hypoxic ischemic encephalopathy) in the perinatal period. Hydranencephaly, Proliferative vasculopathy and hydranencephaly-hydrocephalus syndrome, and multicystic encephalopathy patients rarely survive beyond infancy.

Encephalomalacia can also occur in adults due to brain infarction, hemorrhage, etc. Focal encephalomalacia may not communicate with a CSF space. An arachnoid cyst is a CSF containing an intra-arachnoid cyst (due to splitting) without ventricular communication.

Arachnoid cysts are extra-axial and may have a mass effect. They are commonly supratentorial and usually seen in the middle cranial fossa and Sylvian fissure. Other places of arachnoid cysts include the cerebellopontine angle, cisterna magna, quadrigeminal cisterns, suprasellar regions, cerebral convexities, and spinal cord. Ependymal cysts are usually seen deep in the parenchyma and in intraventricular, periventricular, and subarachnoid spaces, and they are lined by ependymal cells. MCM and DWS are cystic lesions in the posterior cranial fossa. MCM is characterized by intact cerebellar vermis and absence of hydrocephalus, while DWS is associated with dysgenesis or agenesis of the cerebellar vermis, hydrocephalus, and agenesis of the corpus callosum.

Management of porencephaly includes antiepileptics, treatment of underlying disorders, physiotherapy, and rehabilitation therapy. Prognosis is dependent on the size, extent, and anatomical distribution of the lesion, underlying aetiologies, and associated abnormalities. Awareness regarding porencephaly, its presentation, and differential diagnosis will help in better management of the disorder.

REFERENCES

1. Maurice P, Guilbaud L, Garel J, et al. Prevalence of COL4A1 and COL4A2 mutations in severe fetal multifocal hemorrhagic and/or ischemic cerebral lesions. *Ultrasound Obstet Gynecol* 2021;57(5):783–789.
2. Tambuzzi S, Gentile G, Zoja R. Porencephalic cyst in adult. *Autops Case Rep* 2022;12:e2021351.