CASE REPORT

Cerebral multicystic lesions in a child with neurofibromatosis

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SUMMARY

Neurofibromatosis type 1 (NF-1) is an autosomal dominant neurocutaneous syndrome, with frequent involvement of the central nervous system (CNS). As well as abnormal cellular differentiation, disordered cell migration during development is the most common cause of the various brain lesions. Cystic lesions are rarely observed in neurocutaneous diseases, and the origin of the cysts is not known. This paper presents a rare case, a child at the age of 3, who was diagnosed as NF-1 and was observed to have asymptomatic cystic lesions in right temporal lobe in radiological examination of CNS. This study draws attention to the relationship between these rare cystic lesions of unknown origin and neurocutaneous diseases.

BACKGROUND

Radiological evaluation of the central nervous system (CNS) is important for both diagnosis of neurocutaneous diseases and the follow-up of possibly developing pathologies. MRI of CNS in neurofibromatosis type 1 (NF-1) shows different findings, varying from unidentified bright objects (UBO) to astrocytoma and optic glioma.[1] The literature includes studies reporting the presence of multiple cerebral cystic lesions in cases diagnosed as tuberous sclerosis and hypomelanosis,[2 3] and a sole case diagnosed as NF-1.[4] Due to its rare incidence, this study presents only the second NF-1 case having asymptomatic cerebral cystic lesions.

CASE PRESENTATION

A 3-year-old boy was referred to our clinic for the evaluation of NF-1 due to the brown spots on his body. The patient's medical history indicated the presence of these spots from birth by his family (NF-1 diagnosis). The clinical diagnosis was based on many cafe-au-lait spots and axillary-pubic freckling and Lisch nodules in eyes. Neurological examination gave normal findings. Vital findings were normal. The chromosome analysis was not performed. The patient had one healthy female sibling, and there was no family history of NF-1. In addition to his motor and mental development, his birth history and neonatal milestones were normal.

INVESTIGATIONS

At the age of 3 years, his cranial MRI identified multiple cysts in the right temporal areas, isointense to cerebrospinal fluid in all sequences, with a maximal diameter of 2–14 mm. Appearing cortically and subcortically, the cysts were found to cause a mass effect on the adjacent cortex; however, no surrounding oedema was found (figures 1 and 2). Especially in the globus pallidus, internal capsule, thalamus, cerebellum and brain stem regions, basal ganglia were observed to have few lesions that were hyperintense on T2-weighted images. This state was typical for unidentified bright objects of NF-1 (figure 2). They are generally asymmetrical and diffused. The patient had normal spinal MRI.

DIFFERENTIAL DIAGNOSIS

- Genetic and metabolic disorders, including mucopolysaccharidoses, mannosidosis, Lowe syndrome, megalencephaly and ectodermal dysplasia.
- Neurocutaneous diseases such as tuberous sclerosis complex and NF-1.

TREATMENT

Conservative treatment.

OUTCOME AND FOLLOW-UP

In the 1-year follow-up, the patient showed no neuromotor deterioration and radiological progression.

DISCUSSION

The evaluation of basal cerebral and spinal MRI during the diagnosis of neurocutaneous diseases is necessary for the verification of diagnosis and determination and pursuit of developing pathologies. The evaluation of CNS in NF-1 may help heterotopia, cortical dysplasia, cortical malformations like pachygyria and polymicrogryia, UBOs, optic glioma, encephalocele, Arnold-Chiari malformation, aqueductal stenosis and spinal meningocele. Hyperintense lesions on T2-weighted brain MRI in specific places are frequently observed and pathognomonic of NF-1. Similar with the case of our study, basal ganglia, cerebellum and brain-stem regions are the places they are commonly seen. The diagnosis of NF-1 can be supported by the presence of the lesions.[1]

Cohen *et al*[4] reported the first case to have multiple cysts and NF-1 diagnosis. Radiological findings of this case are similar to that of our case, except for the localisation of lesions. The cysts of both cases were asymptomatic, of different sizes, multiple, cortical-located and subcortical-located, with mass effect on adjacent structures, and with no oedema presence around the cysts. The cause and pathogenesis of these cystic lesions determined in NF-1 are not known. However, these cysts are thought to develop due to cystic degeneration of dysplastic lesions in white substances, focal

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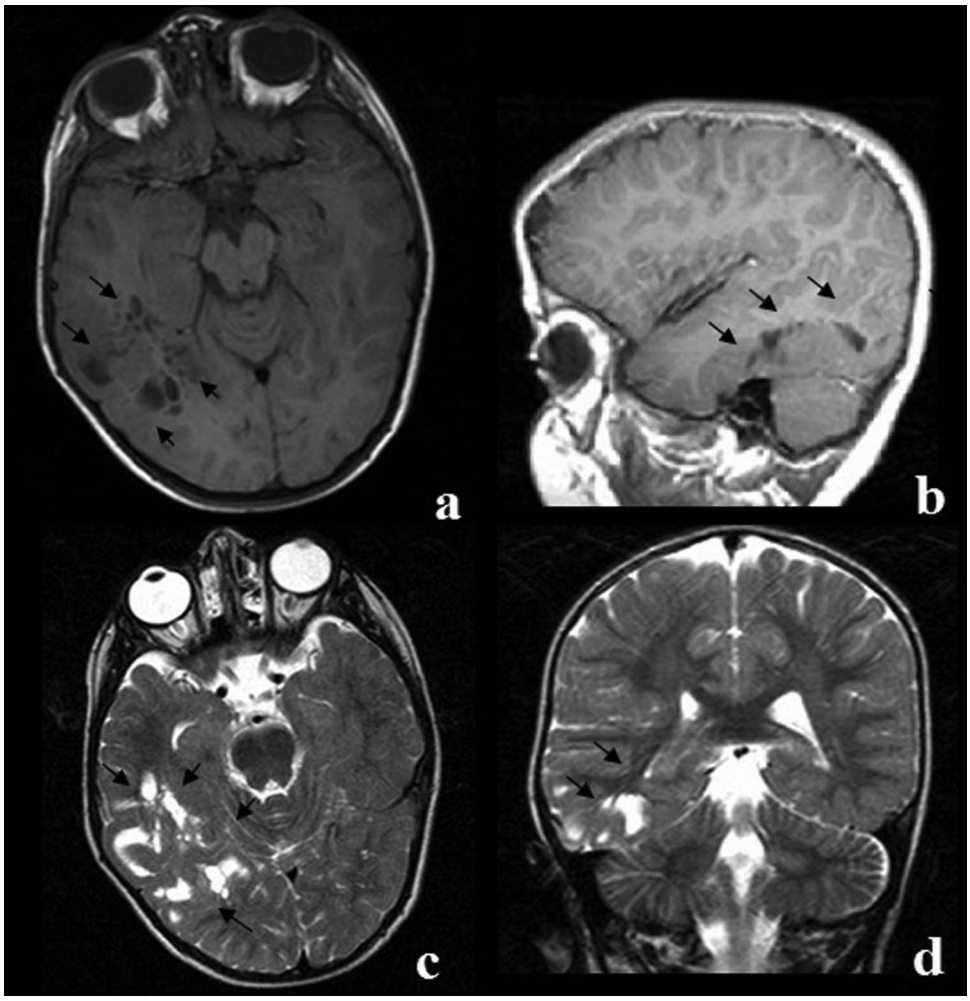

Figure 1 Axial (A) and sagittal (B) T1-weighted MRIs show a cluster of small cysts in the right temporal white matter near the lateral ventricle. Axial (C) and coronal (D) T2-weighted MRIs. Multiple cortical and subcortical cysts are seen. Note pressure on, and distortion of, the adjacent cortex, without any surrounding oedema (black arrows).

extension of perivascular areas or parenchymal neuroepithelial cysts.[4]

Another rare radiological finding determined in neurocutaneous diseases is intracerebral cysts. Tassel *et al*[2] determined cerebral cysts, 44% of which were in different locations, in 18 patients with tuberculosis diagnosis. Rott *et al*[5] described large cyst-like brain lesions in the subcortical area of three patients, and assumed the lesions to represent cellular degeneration of cortical tuber. Jurkiewicz *et al*[6] reported 17 patients having cyst-like cortical lesions after examining MRI scans of 73 patients with tuberous sclerosis complex. The cysts of this case had different appearance on MRI than the patients with

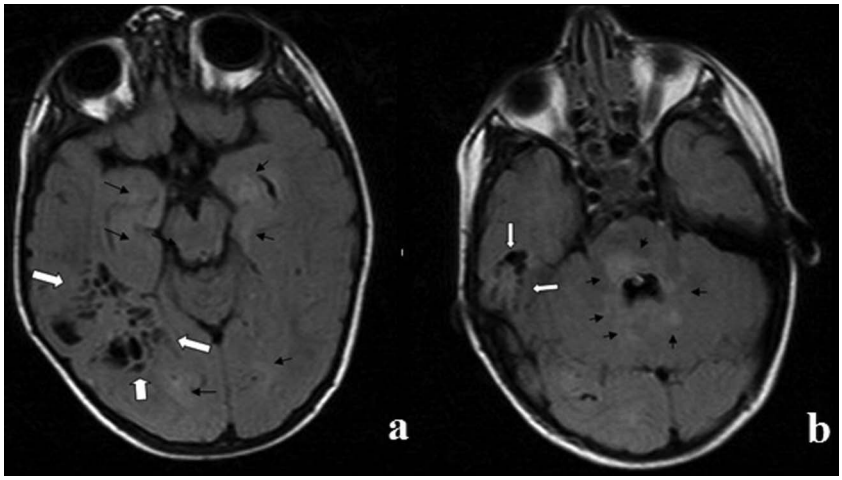

Figure 2 Axial fluid-attenuated inversion-recovery MRIs (A) and (B) show that the cysts are isointense with cerebrospinal fluid (white arrows) and unidentified bright objects (black arrows).

tuberous sclerosis complex. The constant existence of a peripheral hyperintense signal on T2-weighted images and on fluid-attenuated inversion-recovery sequences results from the fact that the location of the cysts in tuberous sclerosis complex are inside cortical tubers.[6] The cystic spaces in the present case are irrelevant to the unidentified bright objects of NF, since the latter does not usually undergo cystic degeneration, and since our patient's cystic spaces are not located in the area where unidentified bright objects are generally found.[1]

In conclusion, cerebral cystic lesions in neurocutaneous diseases are rarely observed. These lesions with unknown causes may be rarely observed in cases diagnosed as NF-1.

Learning points

- The nature of these cysts is unknown.
- These lesions must be differentiated from other cystic lesions of the brain.
- The radiological assessment is the critical point in the diagnosis.
- These lesions may be rarely observed in cases diagnosed as neurofibromatosis type 1.

Competing interests None.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

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