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Corpus Callosum Cysts in Mucopolysaccharidosis: A Case Report

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Abstract | **Case Report**

Mucopolysaccharidoses are rare, progressive, multisystem lysosomal storage disorders caused by enzymatic deficiencies leading to the accumulation of glycosaminoglycans in tissues. Neurological involvement is particularly prominent in neuronopathic forms, especially severe MPS I, severe MPS II, MPS III, and MPS VII, although neuroradiological abnormalities may also be observed in forms classically considered non-neuronopathic or only mildly neuronopathic. Among these abnormalities, cystic lesions of the corpus callosum most often correspond to dilated Virchow–Robin perivascular spaces, sometimes associated with white matter signal abnormalities, ventriculomegaly, cerebral atrophy, or communicating hydrocephalus. Brain MRI, particularly sagittal T1- and T2-weighted sequences, axial T2, FLAIR, and diffusion-weighted imaging, is the modality of choice for characterizing these lesions, differentiating them from leukodystrophy, cystic encephalomalacia, or primary callosal malformations, and ensuring follow-up. Recognition of these lesions has diagnostic value, because their presence in the corpus callosum is unusual in healthy individuals and should suggest a metabolic disorder, especially when associated with dysostosis multiplex, macrocephaly, ENT involvement, hepatosplenomegaly, or neurodevelopmental delay. Management is based on a multidisciplinary approach combining specific therapy when available, treatment of neurological complications, and regular radioclinical follow-up.

Keywords: Mucopolysaccharidoses, Glycosaminoglycans, Corpus callosum, Dilated Virchow–Robin spaces, Magnetic resonance imaging, Lysosomal storage disorders.

INTRODUCTION

Mucopolysaccharidoses (MPS) are a group of inherited lysosomal storage disorders characterized by defective degradation of glycosaminoglycans, formerly known as mucopolysaccharides, resulting in progressive accumulation within lysosomes and the extracellular space [1–4]. They are classically divided into several clinical types — MPS I, II, III, IV, VI, VII, and IX — corresponding to different enzymatic deficiencies, with autosomal recessive inheritance in most cases, except for MPS II, or Hunter syndrome, which is X-linked [1,3,4]. Clinically, MPS combines, to varying degrees, coarse facial features, dysostosis multiplex, short stature, joint stiffness or laxity depending on the type, ENT and respiratory involvement, valvular heart disease, hepatosplenomegaly, ophthalmological abnormalities, and neurological impairment [1,4,11,12]. Central nervous system involvement is not uniform: it is prominent in MPS III, or Sanfilippo syndrome, in severe MPS I, or Hurler syndrome, in severe MPS II, or neuronopathic Hunter syndrome, and in some forms of MPS VII, whereas MPS IV and VI are more commonly dominated by osteoarticular and spinal cord involvement, with usually preserved intelligence but a risk of cervicomedullary compression [1,3,4,12]. Medical imaging plays a central role in the evaluation of MPS. Skeletal radiographs allow recognition of dysostosis multiplex, whereas MRI is the reference modality for assessing the brain, corpus callosum, posterior fossa, perivascular spaces, ventricles, and spinal axis [3–5,9]. The most frequently described cerebral abnormalities include dilated perivascular spaces, white matter signal abnormalities, ventriculomegaly or hydrocephalus, cortical and subcortical atrophy, corpus callosum abnormalities, and occasionally basal ganglia lesions producing a cribriform or “honeycomb-like” appearance [2–7]. In this context, “corpus callosum cysts” must be interpreted with caution: they do not always represent true malformative cysts, but most often correspond to fluid-filled cavities related to dilated perivascular spaces within the corpus callosum, best seen on midline or paramedian sagittal images [3–5].

CASE REPORT

6-year-old girl followed for mucopolysaccharidosis, with metabolic keratopathy on ophthalmological examination and headache, A brain MRI was performed, showing multiple cystic and microcystic lesions involving the corpus callosum and the para-occipital periventricular white matter, showing CSF-like signal intensity, with no contrast enhancement after gadolinium administration and no diffusion restriction. Some of these lesions are centered by a small enhancing venule, supporting the diagnosis of dilated Virchow–Robin spaces (Fig 1).

The diagnosis of Mucopolysaccharidosis type 1 was established based on increased urinary excretion of dermatan sulfate and heparan sulfate, markedly reduced alpha-L-iduronidase activity, and the identification of pathogenic variants in the IDUA gene. These findings were consistent with a suggestive clinical picture combining facial dysmorphism, corneal clouding, and ENT involvement.

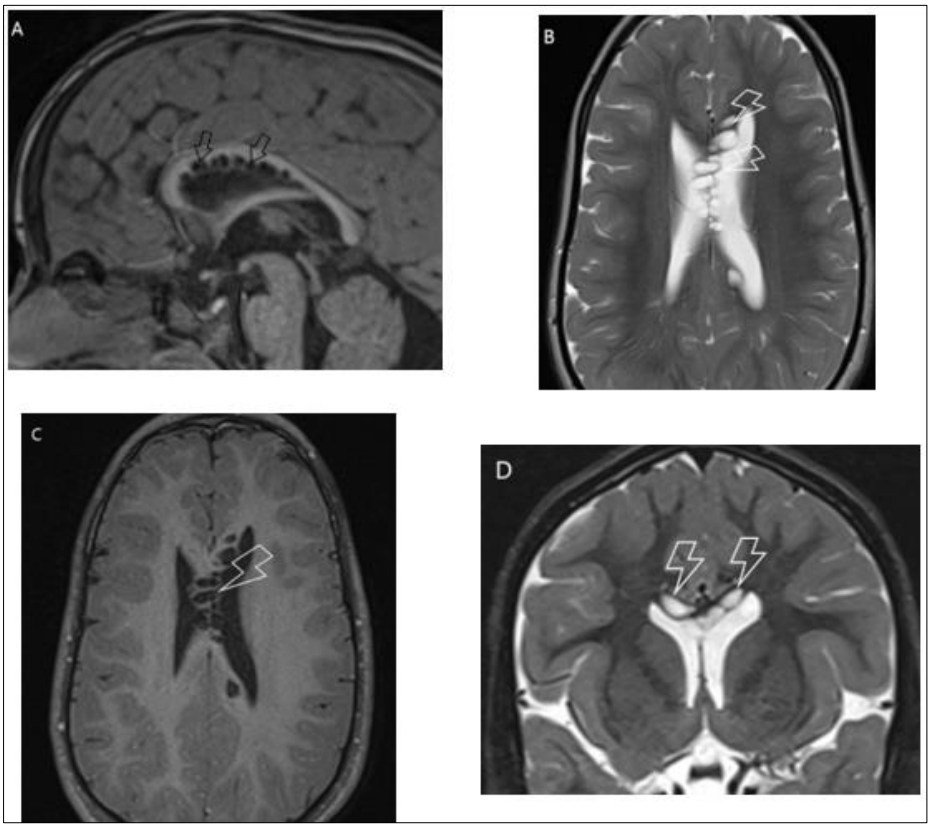

Figure 1:
A: Sagittal T1-weighted MRI image showing adjacent thin-walled cystic lesions within the corpus callosum, with CSF-like signal intensity (arrows).
B: Bilateral cystic lesions of the corpus callosum showing marked T2 hyperintensity (arrows).
C: No contrast enhancement after gadolinium administration, with a few small central venular structures (arrows).
D: Coronal fat-saturated T2-weighted sequence.

DISCUSSION

From a pathophysiological perspective, the accumulation of glycosaminoglycans within the meninges, arachnoid villi, perivascular spaces, and cerebral parenchyma explains many of the abnormalities observed on MRI [1,3,4]. GAG deposits may impair cerebrospinal fluid circulation and resorption, promote enlargement of the subarachnoid spaces and ventricles, and lead to dilatation of the perivascular spaces [1,3]. Reichert *et al.,* emphasized that these dilated perivascular spaces contain a mixture of interstitial fluid, CSF, and probably storage material, and that they may represent an early marker of abnormal CSF circulation before more obvious ventriculomegaly develops [3]. On MRI, these lesions strictly follow CSF signal intensity on all sequences: they are hypointense on T1-weighted images, hyperintense on T2-weighted images, suppressed on FLAIR when purely fluid-filled, and show neither diffusion restriction nor contrast enhancement. They appear as small, round, fusiform, or cribriform cavities, often multiple [3–5]. Their size is usually between 2 and 8 mm, although larger forms may be observed [3]. Corpus callosum involvement is of particular interest because dilated perivascular spaces in this location are uncommon in healthy subjects; their presence, especially when multiple and associated with white matter abnormalities, should therefore draw the

radiologist's attention to MPS or another metabolic disease [3]. Palmucci *et al.,* indicated that the corpus callosum, well evaluated on sagittal images, may sometimes be the isolated site of these lesions, although they may also involve the parieto-occipital white matter, basal ganglia, thalami, and the gray–white matter junction [4]. In the series by Calleja Gero et al., corpus callosum abnormalities were found in several patients, including focal lesions in one patient with Hurler syndrome, dilated callosal perivascular spaces in two children with Hunter syndrome, and callosal atrophy in one patient with MPS III and in one patient with a moderate form of MPS II [2]. These data show that callosal involvement is not specific to a single type of MPS but belongs to a broader neuroradiological spectrum. The associated clinical features strongly depend on the type of MPS. In MPS I, alpha-L-iduronidase deficiency leads to accumulation of dermatan sulfate and heparan sulfate; Hurler syndrome is the most severe form, with progressive dysmorphism, macrocephaly, hernias, recurrent ENT infections, dysostosis multiplex, cardiorespiratory involvement, and progressive neurodevelopmental delay [1,3,11]. Hurler–Scheie and Scheie forms are more attenuated, with absent or less marked cognitive involvement, although osteoarticular, ophthalmological, and cardiac abnormalities may be significant [1,11]. In MPS II, or Hunter syndrome, involvement is related to iduronate-2-sulfatase deficiency; it affects almost exclusively boys and includes a severe neuronopathic form, with developmental delay, behavioral disorders, cognitive regression, and marked MRI abnormalities, and an attenuated form in which intelligence may be preserved despite sometimes significant neuroradiological lesions [3,7,8,14]. MPS III, or Sanfilippo syndrome, is dominated by neurobehavioral involvement, with hyperactivity, sleep disorders, delayed then regressive language development, progressive childhood dementia, and more subtle somatic signs, explaining why diagnosis is often delayed [1,3]. MPS IV and VI differ by less typical central neurological involvement: MPS IV, or Morquio syndrome, is mainly characterized by skeletal dysplasia, atlantoaxial instability, and risk of cervical myelopathy, whereas MPS VI, or Maroteaux–Lamy syndrome, combines dysostosis multiplex, cardiorespiratory involvement, corneal clouding, and a high risk of spinal cord compression, with generally preserved intelligence [1,4,12]. MPS VII, a rarer disorder, may combine visceral involvement, dysostosis multiplex, developmental delay, and sometimes fetal hydrops [3,4]. Brain imaging findings in MPS must be interpreted in light of this clinical heterogeneity. Dilated perivascular spaces are particularly described in MPS I and II but may also be observed in MPS III and VI [3,4,6]. White matter abnormalities appear as T2/FLAIR hyperintense areas, often periventricular and sometimes subcortical, which may mimic leukodystrophy [3,4,6]. They may correspond to dysmyelination, progressive demyelination, gliosis, or an indirect effect of lysosomal storage on oligodendrocytes and neurons [3,4,7]. Ventriculomegaly may reflect cerebral atrophy, communicating hydrocephalus, or a combination of both; therefore, analysis of the subarachnoid spaces, head circumference, clinical evolution, signs of intracranial hypertension, and CSF dynamics is essential [3,13]. Cortical and subcortical atrophy, when present, generally reflects more advanced neurodegenerative involvement, although the correlation between MRI severity and cognitive level remains imperfect [2,6,7]. In the series by Matheus et al., patients with MPS I or II and mild clinical presentation could show severe cerebral MRI abnormalities without intellectual disability, confirming that imaging should not be interpreted in isolation [6]. Similarly, Calleja Gero *et al.,* found no clear correlation between neuroradiological abnormalities, MPS type, and clinical severity [2]. The differential diagnosis of corpus callosum cysts in this setting includes nonspecific dilated perivascular spaces, sequelae of ischemia or infection, cystic periventricular leukomalacia, certain leukodystrophies, malformative abnormalities of the corpus callosum, neuroepithelial cysts, porencephalic cavities, and some phakomatoses [3,5]. Several arguments favor MPS: multiplicity of lesions, strictly fluid-like signal without edema or enhancement, cribriform or fusiform arrangement, association with white matter hyperintensities, ventriculomegaly, macrocephaly, thickening of the cranial vault, sellar abnormalities, dysostosis multiplex, cervical canal stenosis, hypertrophy of retro-odontoid tissues, and suggestive systemic signs [3,4]. Absence of diffusion restriction helps exclude certain ischemic or infectious lesions, whereas the absence of significant mass effect and enhancement makes an active tumoral or inflammatory lesion unlikely [3]. The MRI protocol should be adapted to the patient's age, anesthetic risk, and the multisystem nature of the disease. A brain protocol including 3D T1, T2, FLAIR, diffusion-weighted imaging, T2*/SWI, and high-quality sagittal reconstructions allows assessment of the corpus callosum, ventricles, perivascular spaces, white matter, and basal ganglia [3,9]. Gadolinium injection is not systematically required to characterize perivascular spaces but may be useful in cases of diagnostic uncertainty or when searching for an associated complication [3]. Spinal MRI, particularly of the craniocervical junction, is essential in MPS I, IV, VI, and VII, but also in MPS II, in order to detect cervical canal stenosis, odontoid hypoplasia, atlantoaxial instability, ligamentous or dural thickening, and spinal cord injury [3,4,8,12]. The frequent need for anesthesia in children requires shortening the examination while preserving the essential sequences, because patients with MPS often have macroglossia, airway infiltration, limited mouth opening, cervical instability, and a high risk of difficult intubation [3,4].

Management of MPS is based on a multidisciplinary strategy. Biological diagnosis relies on urinary GAG measurement, specific enzymatic testing, and genetic confirmation [1,3,11]. Symptomatic

treatment includes ENT and respiratory care, cardiac surveillance, ophthalmological follow-up, physiotherapy, occupational therapy, treatment of sleep disorders, neuropsychological management, orthopedic surgery, spinal decompression in cases of symptomatic stenosis, and ventriculoperitoneal shunting or other neurosurgical approaches in symptomatic communicating hydrocephalus [1,12,13]. Aliabadi *et al.,* showed that CSF shunting could be effective in communicating hydrocephalus associated with MPS I, II, and III, but the indication must be discussed in an expert center, taking into account anesthetic risk and overall neurological status [13]. Specific treatments vary according to the MPS type: intravenous enzyme replacement therapy is available particularly for MPS I, II, and VI, improving several visceral and functional manifestations, but its effect on cerebral involvement is limited by poor penetration across the blood–brain barrier [1,3,10–12]. Hematopoietic stem cell transplantation is indicated in selected severe forms of MPS I, especially when performed early before advanced neurological involvement, but it carries significant morbidity and does not completely correct established skeletal or corneal abnormalities [1,10,11]. Emerging approaches — intrathecal or intracerebroventricular enzyme replacement therapy, gene therapy, substrate reduction therapy, and strategies designed to cross the blood–brain barrier — specifically aim to better control neurocognitive involvement, which remains the main therapeutic challenge in neuronopathic forms [1,9,10].

In radiological practice, corpus callosum cysts in MPS should therefore be described precisely: number, size, callosal topography — genu, body, splenium — signal on T1, T2, and FLAIR, strictly fluid-like nature or otherwise, diffusion characteristics, possible enhancement, association with white matter lesions, ventriculomegaly, atrophy, basal ganglia abnormalities, posterior fossa findings, and spinal signs [3–5]. The radiological conclusion should avoid describing these findings as a "cystic tumor" or "indeterminate cystic lesion" when the appearance is typical of dilated perivascular spaces. A useful formulation would be: "multiple fluid-like cystic formations of the corpus callosum, without diffusion restriction or enhancement, compatible with dilated perivascular spaces in the context of mucopolysaccharidosis." When the diagnosis of MPS is not yet known, the association of these lesions with craniospinal dysostosis, macrocephaly, ventriculomegaly, or cervical stenosis should prompt recommendation of a specialized metabolic work-up [3,4].

Conclusion

Cystic lesions of the corpus callosum observed in mucopolysaccharidoses most often correspond to dilated Virchow–Robin perivascular spaces, representing a neuroradiological expression of glycosaminoglycan storage and impaired cerebrospinal fluid circulation. Their callosal location is important because it is unusual in common variants of perivascular spaces and should prompt a search for associated signs of MPS. MRI is the key examination for characterizing these lesions and assessing the white matter, ventricles, cerebral atrophy, and especially the craniocervical axis. The isolated prognostic value of callosal cysts remains limited, because the correlation between MRI abnormalities and cognitive severity is variable; however, their recognition helps guide diagnosis, organize appropriate surveillance, and direct multidisciplinary management. In MPS, the radiologist's role is therefore not limited to describing cerebral lesions: it also contributes to early diagnosis, detection of neurosurgical and spinal complications, and follow-up of the effects and limitations of specific treatments.

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