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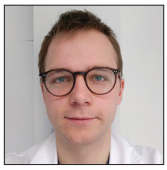
Case Report

Brain overgrowth associated with megalencephaly-capillary malformation syndrome causing progressive Chiari and syringomyelia

Tom Deleu[1], Katrien Jansen[2], Frank Van Calenbergh[1]

Departments of [1]Neurosurgery and [2]Pediatrics, University Hospitals Leuven, Leuven, Belgium.

E-mail: *Tom Deleu - deleu.tom@uzleuven.be; Katrien Jansen - katrien.jansen@uzleuven.be; Frank Van Calenbergh - frank.vancalenbergh@uzleuven.be



***Corresponding author:**
Tom Deleu,
Department of Neurosurgery,
University Hospitals Leuven,
Leuven, Belgium.

deleu.tom@uzleuven.be

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ABSTRACT

Background: Megalencephaly-capillary malformation (M-CM) syndrome is a rare overgrowth syndrome characterized by macrocephaly, port-wine stains, asymmetric brain growth, hydrocephalus, and developmental delay. Cerebellar tonsil herniation is often seen, but rarely with syringomyelia.

Case Description: A newborn with M-CM syndrome developed a progressive Chiari malformation type I (CM-I) with syringomyelia. At 4 months, he was treated for subdural hematomas, while at 10 months, he required a shunt for hydrocephalus. At 16 years of age, he newly presented a left hemiparesis and ataxia. Notably, successive volumetric measurements of the posterior fossa/cerebellum showed disproportionate cerebellar growth over time that correlated with the appearance of a CM-I. Following a suboccipital craniectomy with C1-laminectomy and duraplasty, he neurologically improved.

Conclusion: M-CM with CM-I and syringomyelia rarely present together. Here, we treated an infant with M-CM who developed a progressive CM-I malformation and syringomyelia reflecting disproportionate growth of the cerebellum/posterior fossa over a 16-year period.

Keywords: Chiari I malformation, Megalencephaly-capillary malformation, Overgrowth syndromes, Syringomyelia

INTRODUCTION

Macrocephaly-cutis marmorata telangiectatica congenita (M-CMTC) is a rare condition. It is characterized by macrocephaly, port-wine stains, asymmetric growth, polydactyly, joint hyperlaxity, and central nervous system abnormalities (i.e., including hydrocephalus and developmental delay).[1-3] It was first described in 1997 by Clayton-Smith *et al.*,[2] but has since been renamed macrocephaly-capillary malformation (M-CM).[9] A small group of these patients has a genetic defect at the PIK3CA site.[7] Neuroimaging findings include: ventriculomegaly (76%), polymicrogyria or cortical dysplasia (23%), brain asymmetry (52%), white matter abnormalities (90%), and cerebellar tonsil herniation (70%).[3] Here, we report an infant with M-CM who, over a 16-year period, progressively developed a CM-I and an extensive syringomyelia.

CASE REPORT

A male infant, diagnosed with M-CMTC/M-CM, presented with diffuse capillary malformations on his face, neck, shoulders, and limbs at birth. At 4 months of age, the MRI showed progressive macrocrania with large subdural hematomas requiring evacuation. Volumetric measurements of the posterior fossa and cerebellum on sequential MRI's (i.e., using the iPlan™ software [Brainlab AG, Munich, Germany]) were performed. They showed a steep rise in the ratio of cerebellar to posterior fossa volume between the MRI at 4 months and at 10 months that coincided with the appearance of a CM-I [Table 1]. Further, at 10 months, ventriculomegaly required the placement of a ventriculoperitoneal shunt. At 1 year, he developed ventriculitis, a left-sided subdural empyema, and bilateral internal jugular vein thrombosis (i.e., Lemierre syndrome: due to anaerobic infection of the head/neck with pharyngeal bacteria).[6] Despite the evacuation of the subdural empyema and IV antibiotics, he had a residual left hemiparesis and hemianopic defects. At 9 years of age, although asymptomatic, the MRI demonstrated progressive tonsillar herniation and a syrinx [Figure 1]. The syrinx expanded slowly between ages 9 and 15, but remained asymptomatic. However, at age 16, he presented with increased nausea, a denser left hemiparesis, and plus ataxia. The MRI, now, documented a holocord syrinx. During a suboccipital craniectomy and C1 laminectomy, with duraplasty, the enormously dilated midline occipital veins were encountered (i.e., vascular malformations are highly correlated with the M-CM syndrome). Three, 6, and 9 months postoperatively, the patient's only residual deficit was a mild gait disturbance.

DISCUSSION

Syringomyelia

Seventy percent of M-CM patients have a CM-I,[3] with associated syringomyelia (23% and 65%).[1,8] There was a steep rise in the ratio of cerebellar volume to posterior fossa volume on the MRI scans obtained at 4 months and 10 months, likely due to brain overgrowth and a more crowded posterior fossa. Three other mechanisms could have contributed to this finding: hydrocephalus, the VP-shunt, and/or the internal jugular vein thrombosis [Table 2]. The CM-I also led to the formation of a holocord syrinx that slowly increased over a 16-year period.

Table 1: Volume of the posterior fossa and cerebellum as measured on MRI.

Age	Cerebellum (cm^3)	Posterior fossa (cm^3)	Volume ratio (cerebellum/posterior fossa)
4 months	58	85	0.68
10 months	89	108	0.82
9 years	130	159	0.81
15 years	129	159	0.81
16 years	132	157	0.84

Vascular malformations

Vascular malformations are one of the two main features of the M-CM syndrome.[4,5] This patient had diffuse capillary

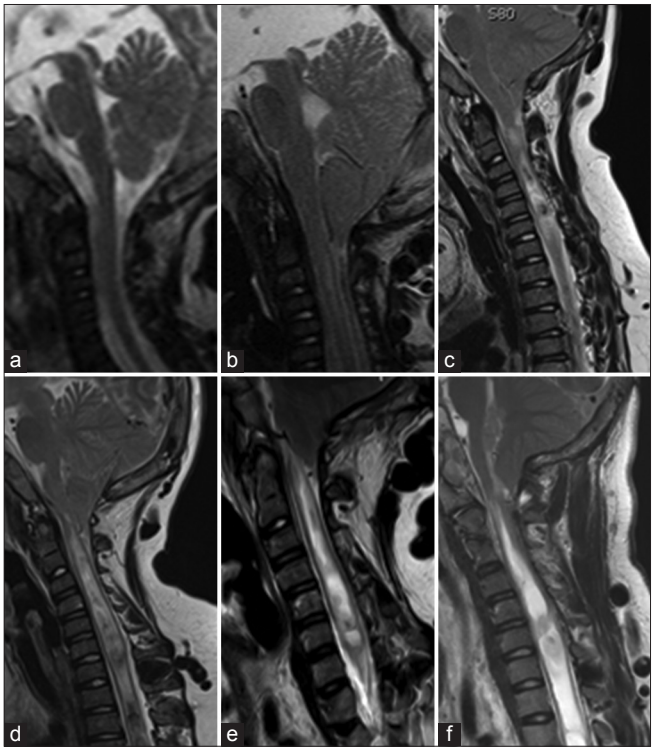

Figure 1: Progression of the Chiari malformation type I and syringomyelia on sagittal T2-weighted MRI (a-e). Images taken at 4 months (a), 10 months (b), 9 years (c), 11 years (d), 16 years (e). Postoperative sagittal T2-weighted MRI (f).

Table 2: Other mechanisms potentially contributing to the development of syringomyelia.

Mechanism contributing to syringomyelia	Effect size and explanation
Hydrocephalus	No effect. The hydrocephalus is controlled with the VP-shunt, with imaging indicating a normal shunt function.
VP-shunt/ craniocerebral disproportion	Unlikely this plays an important role. The shunt was only implanted after the appearance of the CM-I.
Internal jugular vein thrombosis	Minor effect. The thrombosis occurred after the appearance of the CM-I but could contribute to the slow progression.

CM-I: Chiari malformation type I

malformations on his face, neck, shoulders, and limbs, and hugely dilated midline veins in the posterior fossa. Great care was taken during the suboccipital craniotomy to preserve these venous structures and avoid uncontrollable intraoperative hemorrhaging.

CONCLUSION

An infant presented with M-CM syndrome; at 4 months, he had subdural hematomas, and at 10 months, hydrocephalus requiring shunt placement. Over the subsequent 16-year follow-up, and due to disproportionate growth of the cerebellum and posterior fossa, he developed a progressive CM-I malformation and holocord syringomyelia warranting a suboccipital craniectomy and C1 laminectomy, with duraplasty.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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