

Marfanoid habitus associated with hydrocephalus: report of two cases

Hábito marfanóide e hidrocefalia sem cranioestenose: Relato de dois casos

José Aloysio Costa Val¹

Paulo Mallard Scaldaferrí²

ABSTRACT

Marfan syndrome is characterized by manifestations in three systems: skeletal, ocular and cardiovascular. There are no publications of typical Marfan syndrome patients associated with craniosynostosis or hydrocephalus. There are a series of publications of the association between craniosynostosis and marfanoid habitus, which led to the description of new syndromes: Loeys-Dietz types IA, IIA, IB e IIB and Shprintzen-Goldberg. Among those syndromes, there are cases of hydrocephalus, but they are typically associated with craniosynostosis. We describe two cases of patients with marfanoid habitus associated with hydrocephalus without craniosynostosis. The first patient presented with skeletal malformations with arachnodactyly, camptodactyly, malar hypoplasia in association with interatrial shunt, gastroparesis, mild mental retardation and symptomatic hydrocephalus. The other patient presented with marfanoid habitus associated with mild cognitive deficit and symptomatic hydrocephalus. None of those patients had craniosynostosis. Many phenotypic characteristics our patients presented resemble the craniosynostosis-marfanoid habitus syndromes. The absence of craniosynostosis raises suspicion on the possibility of occurrence of those syndromes with atypical characteristics or a new syndrome not yet described. We believe that the description of these cases may enlighten this discussion.

Keywords: Marfan syndrome; Hydrocephalus; Craniosynostosis; Marfanoid habitus

RESUMO

A Síndrome de Marfan é marcada por manifestações em três sistemas: esquelético, ocular e cardiovascular. Não há relatos de casos de Marfan típicos associados a cranioestenose e hidrocefalia. Uma série de relatos de associação entre cranioestenose e hábito marfanóide foram publicadas, levando à descrição de novas síndromes: Loeys-Dietz tipos IA, IIA, IB e IIB e Shprintzen-Goldberg. Entre as síndromes descritas há casos de hidrocefalia, mas todas estão associadas à ocorrência de cranioestenose. Descrevemos aqui dois casos de pacientes com hábitos marfanóides e hidrocefalia sem cranioestenose. Um dos pacientes apresentou malformações esqueléticas marfanóides, com aracnodactilia, camptodactilia, hipoplasia malar associadas a comunicação interatrial, gastroparesia, retardo mental leve e hidrocefalia sintomática. O segundo paciente apresentou hábito marfanóide associado a déficit cognitivo leve e hidrocefalia sintomática. Nenhum dos dois pacientes apresentou cranioestenose. Várias características fenotípicas destes pacientes os assemelham às síndromes de cranioestenose-hábito marfanóide. A ausência de cranioestenose e a presença de hidrocefalia levantam a suspeita de ocorrência de tais síndromes com características atípicas ou nova síndrome ainda não descrita. Acreditamos que a descrição destes casos possa levar à luz esta discussão.

Palavras-chave: Marfan; Hidrocefalia; Cranioestenose; Hábito marfanóide

1. Division of Neurosurgery of Santa Casa de Londrina, Paraná, Brazil
2. Division of Neurosurgery of Londrina State University, Paraná, Brazil
3. Division of Neurology of Santa Paula Hospital, São Paulo, Brazil
4. Division of Neurosurgery of Tulane University, New Orleans, Louisiana, USA
5. Division of Neurosurgery of Santa Paula Hospital, São Paulo, Brazil
6. Department of Surgery, Post Graduation Section, Federal University of Rio Grande do Sul, Rio Grande do Sul, Brazil

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INTRODUCTION

Marfan syndrome is one of the most common disorders affecting the connective tissue, being classically branded by involving three systems: skeletal, ocular and cardiovascular. Among the most common manifestations there are aortic aneurisms, eye lens subluxation and excessive growth of long bones^{3, 7, 10}. Most of the typical cases of Marfan are associated to a mutation on the fibrilin-1 gene (FBN1)⁶.

There are no reports of typical Marfan cases associated with craniostenosis and hydrocephalus. The first report of a patient with marfanoid habitus and craniostenosis was published in 1987⁴. Since then, a series of reports of associations between craniostenosis and marfanoid habitus have been published^{2, 9, 11, 12}. New syndromes associated with marfanoid and craniostenosis characteristics have been described: Loeys-Dietz types IA, IIA, IB and IIB⁹ and Shprintzen-Goldberg^{5, 8, 13-16}. Among the new syndromes associated with marfanoid habitus there were also cases of hydrocephaly^{1, 5, 9}.

We hereby describe two cases of patients with marfanoid habitus associated with hydrocephalus without craniostenosis.

CASE REPORT

Case 1. BSS, female patient, being followed since childhood, has marfanoid habitus, with the upper body being larger than her lower body. She also has other skeletal malformations, including arachnodactyly, camptodactyly and important scoliosis, with complications which were treated in the past with thoracolumbar surgical arthrodesis. Among its craniofacial manifestations we noticed malar hypoplasia and hypertelorism. She also had an interatrial cardiac shunt, which was surgically corrected.

Later on, the patient developed bloating and dyspepsia refractory to regular treatment. A barium study of her esophagus-stomach-duodenum transit showed longer gastric transit time due to gastroparesis.

In the follow-up, brain MRI showed supratentorial hydrocephalus, cerebral aqueduct stenosis and underdevelopment of the corpus callosum and hippocampus. Later on, the patient developed clinical symptoms of hydrocephalus and was submitted to an endoscopic third-ventriculostomy, with satisfactory control of the intracranial hypertension.

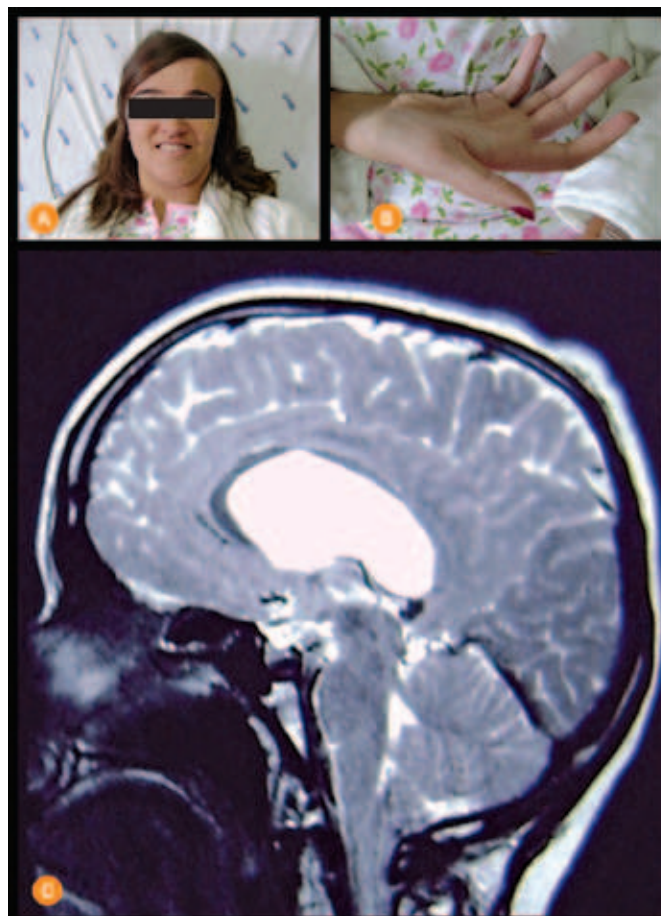


Figure 1. A. Hipertelorism, malar hipoplasia. B. Arachnodactily, camptodactily. C. MRI T2W (sagittal cut) , T2: supratentorial hidrocephalus due to aqueduct stenosis.

Case 2. Since birth, JDS, male patient, has had an atypical biophysical profile when compared to the rest of the family. He was unexpectedly slim and with an important scoliosis. We also noticed arachnodactyly and enlarged upper body when compared to his lower body. Among his craniofacial manifestations, we noticed hypertelorism, malar hypoplasia and low implantation of the ears.

At 6 years of age, his mother took him to the physician for an evaluation: he had mild mental retardation, with clear difficulties to learn exact sciences, especially mathematics. She also reported the child had urinary incontinence and progressive spastic paraparesis.

Brain MRI showed supratentorial hydrocephalus with aqueduct stenosis. Since symptoms matched MRI findings, we indicated endoscopic third-ventriculostomy. He improved his symptoms and continues to being followed up : his school performance improved and there were also improvements in gait and urinary incontinence.

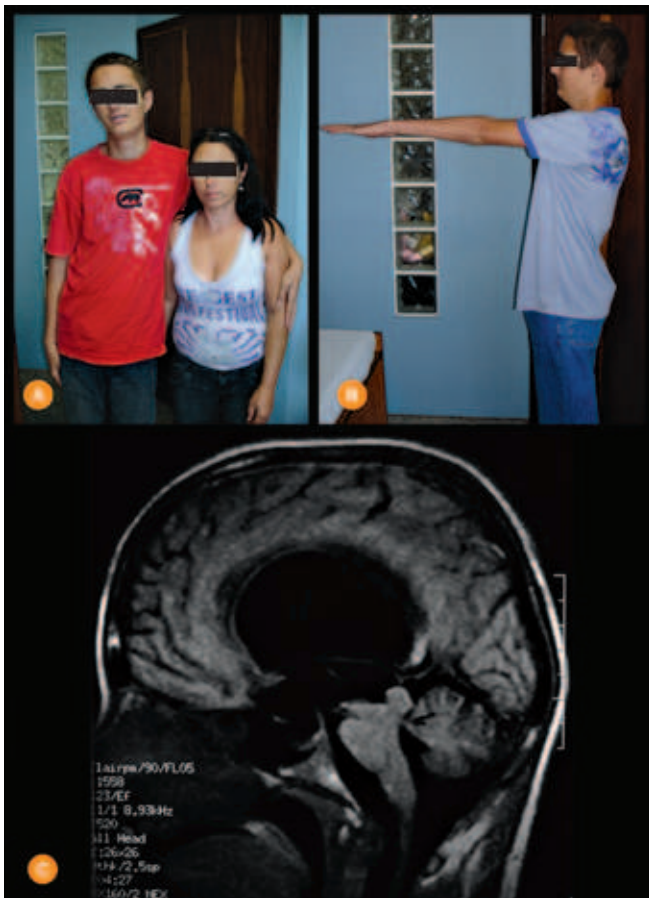


Figure 2. Upper segment/trunk disproportion, low ear implantation, arachnoidactyly. B. Patient and mother's height. C. MRI, sagittal cut, without contrast: supratentorial hydrocephalus caused by aqueduct stenosis.

DISCUSSION

There are no reports of patients with typical Marfan disease associated with hydrocephalus or craniostenosis. The findings of patients with marfanoid habitus and craniostenosis have led to the identification of new syndromes: Loeys-Dietz, Shprintzen-Goldberg and their subtypes.

Among the patients described, there are reports of hydrocephalus^{1,5,9}. The patients were divided in new syndromes according to key characteristics, such as slit uvula, abdominal hernias, mental retardation, chest aneurisms, and other manifestations.

Nonetheless, the presence of craniostenosis in the patients hereby described has led to new diseases be nicknamed "marfanoid craniostenosis-habitus syndromes"². The cases hereby ex-

posed present numerous phenotypic characteristics matching the ones from the previously described syndromes.

The presence of marfanoid habitus, with scoliosis, malar hypoplasia, arachnoidactyly and camptodactyly associated with hydrocephalus has been described in patients with craniostenosis. The phenotypic findings of the patients hereby presented, especially the congenital association of marfanoid habitus and hydrocephalus suggest overlapping with the marfanoid craniostenosis-habitus syndromes. However, the lack of craniostenosis distinguishes our patients here from the other cases described in the literature, raising the suspicion of the occurrence of such syndromes with atypical characteristics or another syndrome still not described in the literature. We believe the report of these cases may show some light on this discussion.

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CORRESPONDING AUTHOR

José Aloysio da Costa Val
Instituto Mineiro de Neurocirurgia
R. da Paisagem, 280 – Nova Lima
Belo Horizonte, MG
costaval.bh@terra.com.br