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Familial Syringomyelia in Two Siblings

İki Kardeşi Etkileyen Familyal Siringomiyeli

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ABSTRACT

We report on syringomyelia of the thoracic spinal cord in two sisters without a Chiari or any other malformation. In both cases syringomyelia was determined by magnetic resonance imaging (MRI). Spinal MRI showed a syringomyelia between T6-T8 levels in the first sibling and between T7- T9 in the second sibling. The cranio-cervical junction was normal and tonsiller ectopia was not present. Neurological examination of both sisters was normal. They did not undergo surgery as they had mild symptoms without neurological deficit and the size of the syrinx was small. Familial syringomyelia is a very rare finding and extremely rare at only the thoracic level. Genetic and environmental factors seem to be involved in familial syringomyelia.

KEY WORDS: Familial Syringomyelia, Magnetic resonance imaging, Thoracic spinal cord

ÖZ

Bu yazıda iki kızkardeşte görülen, chiari ya da başka bir malformasyon olmaksızın torakal spinal kordda lokalize siringomyeli olgularını sunduk. Her iki vakada siringomyeli MRG ile tespit edildi. Spinal MRG'de birinci kızkardeşte T6-T8 arasında ve ikinci kızkardeşte T7-T9 arasında lokalize siringomyeli tespit edildi. Kranyoservikal bileşke normaldi ve tonsiller ektopi yoktu. Her iki kardeşin de nörolojik muayenesi normaldi. Orta derecede semptomları olması ve sirinks boyutlarının küçük olması nedeniyle cerrahi tedavi uygulanmadı.Familyal siringomyeli nadir görülmekle birlikte, torakal yerleşimli olgulara daha da nadir rastlanılmaktadır. Genetik ve çevresel faktörler, familiyal siringomyeli oluşumunda etkili gibi görünmektedir.

ANAHTAR SÖZCÜKLER: Familyal Siringomyeli, Manyetik Rezonans Görüntüleme, Torasik Spinal Kord

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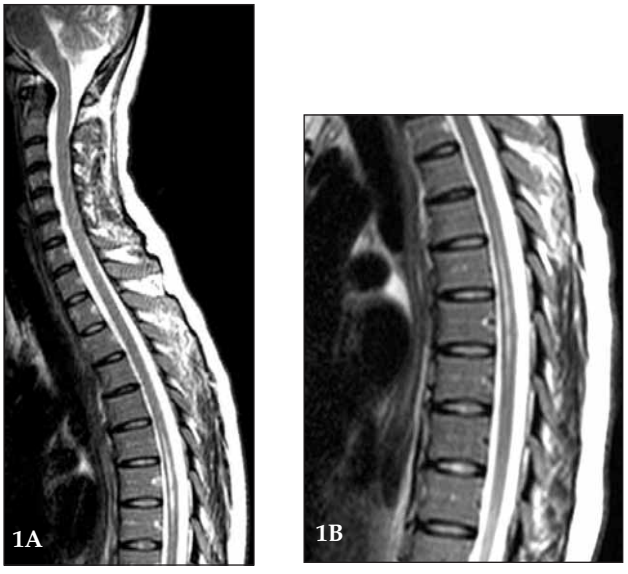
INTRODUCTION

Syringomyelia is a disease demonstrating formation of cystic cavities inside the spinal cord, with characteristic clinical signs and symptoms. It is generally accompanied by malformations such as the Chiari malformation or by trauma. Although it occurs sporadically, extremely rarely familial cases have been reported (1-19). We report syringomyelia of the thoracic spinal cord in two sisters without any other malformation.

CASE REPORTS

Sibling 1: The older sister- 25 years old- had a slight numbness on her left shoulder and back. Cranial nerve functions were intact. Muscle tone and the deep tendon reflexes were normal in all extremities. There was no sensory or motor loss on her extremities. Vibratory sensation was normal. Spinal thoracal MRI showed syringomyelia between T6-T8 levels (Figure 1A,B,C). Cranial, cervical and lumbosacral MRI revealed normal findings. The cranio-cervical junction was normal. Tonsiller ectopia was not present.

Sibling 2: The younger sister - 24 years old - had mild headache with slight numbness on her both shoulders and back. The neurological examination was normal. Cranial and spinal MRI was performed. Syringomyelia was detected between the T7- T9 levels on spinal thoracal MRI (Figure 2A,B,C). Cranial, cervical and lumbosacral MRI findings were normal. Like her sister, the cranio-cervical junction was normal and tonsillar ectopia was not present.



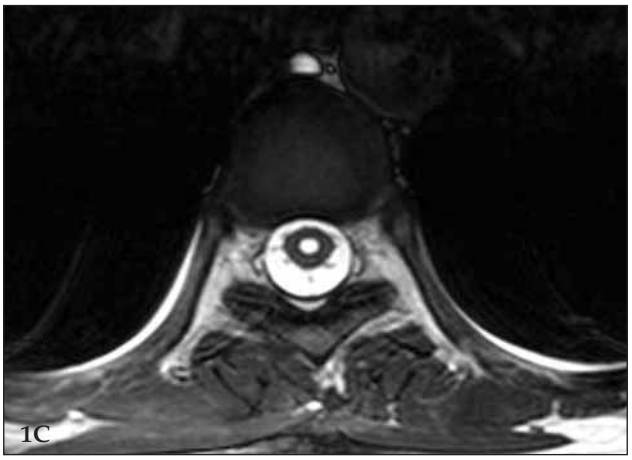
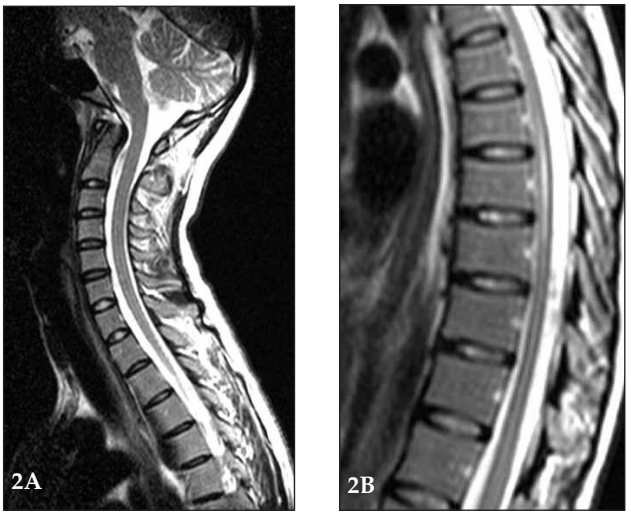


Figure 1: : MRI of the older sister **A,B.** Sagittal and **C**. axial T2-weighted images show syringomyelia between the T6-T8 levels. The cranio-cervical junction is normal. Tonsiller ectopia is not present.



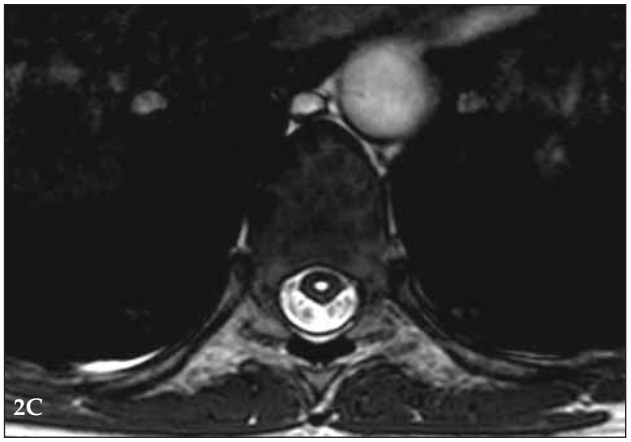


Figure 2: : MRI of the younger sister **A B**. sagittal and **C**. axial T2-weighted spinal MRI reveals syringomyelia between the T7- T9 levels. Note that the cranio-cervical junction is normal and tonsiller ectopia is not seen.

The cases were diagnosed as familial syringomyelia. They did not undergo surgery as both had mild symptoms without neurological deficit and the size of the syrinx was small.

Control MRI obtained a year after the diagnosis revealed that the syringomyelia in both patients remained the same.

DISCUSSION

Syringomyelia is generally a progressive disorder resulting from the formation of cystic cavities inside the spinal cord, with characteristic clinical signs and symptoms (2). It can be classified into communicating and non-communicating groups (17). The non-communicating group has been described after trauma, in association with compression by a tumor and arachnoiditis. In the communicating group, CSF pathways communicate with the syrinx in the spinal cord (1,17).

Although syringomyelia is an uncommon disease of the spinal cord and occurs sporadically, familial cases continue to be reported (6,10,16,19). Twenty-three families with syringomyelia have been reported up to date to our knowledge in literature (1-19). The existence of these families and review of the literature certainly suggests that genetic factors may play role in the pathogenesis of syringomyelia related to abnormalities of the hindbrain structures, although the number of cases is too small for the exact nature of transmission to be recognized (6,18). Rare familial cases with a mendelian transmission of syringomyelia (autosomal dominant or recessive inheritance patterns) have been reported and their incidence estimated as approximately 2% (12,19). It has been suggested that familial syringomyelia may originate from aberrant genes influencing spinal cord development, although a distinct gene location has not been reported yet. The assumption that genetic factors may play a role in the pathogenesis of syringomyelia is supported by the frequent association of syringomyelia with other malformations, especially with Chiari type I malformation. There is a high coincidence of Chiari I malformation (2,10,16,19) up to 80% (12) with other skeletal abnormalities especially at the skull base (1,12) and with basilar impression with an incidence of 18-38% (15). The degree of genetic relation of Chiari type I malformation and syringomyelia is not yet clarified, but the report of Mavinkurve et al. (10) suggests a genetic component to the pathogenesis of this condition. Chiari malformation or other causes of partial obstructions at the cervicomedullary junction, with spontaneous disruption of the wall of a cervical syrinx and formation of a communication between the cavity and the subarachnoid space explains the coincidence and these findings can be shown on MRI (2). Our cases did not have any skeletal anomaly or Chiari malformation.

The association of syringomyelia with hereditary diseases has been reported (14). A case of syringomyelia secondary to arachnoiditis associated with arachnoid telangiectasia was reported in a female patient with no other stigmata of hereditary haemorrhagic telangiectasia (4). Colombo et al reported a case of familial syringomyelia in two sisters, one of whom had syringomyelia and Chiari type I malformation and the other had syringomyelia and Klippel-Feil syndrome (6). A woman with syringobulbia and syringomyelia and her sister with syringomyelia were described by Busis et al (3) and the diagnosis was confirmed radiologically and surgically. Koga et al (8) reported a case of non-hereditary hypopituitarism with Chiari malformation and syringomyelia not associated with perinatal injury, namely a midline anomaly syndrome and MRI-clarified hypoplasia of the anterior pituitary lobe, Arnold-Chiari malformation and cervical syringomyelia. Syringomyelia is also reported in one of monozygotic twins and their brother (9). Monozygotic triplets, each exhibiting variable degrees of tonsillar ectopia and one with a Chiari I malformation and associated syringomyelia were reported and the presence of a common hereditary factor in the aetiology of these malformations was suggested (5). Recently, an article on twin brothers with syringomyelia (16) and another on two sisters each presenting with scoliosis demonstrated a Chiari malformation with syringomyelia (10) and all were treated by cervicomedullary decompression.

In 1982, Newman et al noted a significant association of HLA-A9 with syringomyelia (11). This finding was also mentioned in a report of familial syringomyelia in a brother and sister by Zakeri et al (19). We were not able to study HLA in our patients since they refused.

In addition to genetic predisposition, external environmental influences are reported to play a role in developing syringomyelia (3,7). Malessa et al (9)

reported that heavy work or excessive strain may precipitate the formation of a syrinx. The older sister in our report is a technician in a radiology department and the younger sister is a student, both of which were not involved in a heavy work.

Zakeri et al (19) recommended that close relatives of patients affected with familial syringomyelia undergo routine neurological and radiological surveys. We invited the parents and other sister and brother for further neurological and radiological evaluations. All family members except our cases were normal both on clinical and radiological examinations.

Both genetic and environmental factors have been attributed to be involved in familial syringomyelia. We believe that in both of our cases the main factor leading to syringomyelia was the genetic origin, but environmental factors may also have contributed since both live in Kocaeli, an industrial city. Further research should be performed to explain the exact etiology and pathogenesis.

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