



## **Craniovertebral malformation complex in a child with Weismann-Netter-Stuhl syndrome**

**Ali Al Kaissi,<sup>1</sup> Farid Ben Chehida,<sup>2</sup> Hassan Gharbi,<sup>2</sup> Mourad Jinziri,<sup>3</sup> Hatem Safi,<sup>3</sup>  
Maher Ben Ghachem,<sup>3</sup> Franz Grill,<sup>4</sup> Franz Varga,<sup>1</sup> Klaus Klaushofer<sup>1</sup>**

### **Abstract**

**Objective:** Bowing of the legs is usually thrown into the basket of vitamin D deficiency rickets; therefore, a significant number of affected children can be misdiagnosed and improperly managed. This case illustrates how the careful clinical and radiological assessment of such a case can lead to the adequate understanding of its etiology.

**Description:** We report a sporadic case of a 2-year-old male child who presented with radiological features that were compatible with Weismann-Netter-Stuhl syndrome. In addition, we observed craniovertebral malformation complex. He was of normal intelligence. To our knowledge, the combination of Weismann-Netter-Stuhl syndrome and presence of a hypoplastic occipitalized atlas and further C2-C3 fusion has not been reported before. The diagnosis of Weismann-Netter-Stuhl is discussed. Classically, Weismann-Netter-Stuhl syndrome is characterized by short stature, mental retardation (in some individuals), dural calcification, and anterior bowing of the tibiae. However, we believe that careful clinical and radiological examinations can reveal more striking data which might positively reflect on the whole process of management.

**Comments:** We postulate that the congenital limitations in neck movements in our patient developed because of the marked fusion of the hypoplastic and occipitalized atlas and simultaneous C2-C3 fusion. Therefore, if this form of malformation is disregarded, there may be involvement of the atlantoaxial structure, and this can possibly lead to serious neurological and even life-threatening complications. The use of CT scanning for the detection of such abnormalities can be remarkably important.

*J Pediatr (Rio J). 2006;82(3):236-9: Weismann-Netter-Stuhl syndrome, bowing of the tibiae and fibulae, hypoplasia of the anterior arch of the atlas, occipitalization of the atlas.*

### **Introduction**

Bowing of the tibiae and fibulae is an unusual clinical finding, especially in the absence of predisposing biochemical disorders. According to the London Dysmorphology Database,<sup>1</sup> this feature is found in 99 syndromes, but it rarely occurs without any other skeletal involvement, such as bowing of the femora, as occurs in different camptomelic syndromes. It is an important feature of Weismann-Netter-Stuhl (WNS) syndrome, which was first reported as tibioperoneal diaphyseal toxopachyosteosis by Weismann-Netter and

Stuhl in 1954.<sup>2</sup> This syndrome is defined as an anomaly of the diaphyseal part of both tibiae and fibulae with posterior cortical thickening and anteroposterior bowing. The anomaly is usually bilateral and symmetrical, and patients have short stature. The thickening of the fibula is a true "tibialization" and is an important diagnostic feature of this rare syndrome. The original cases were mentally handicapped, but this feature has been unusual in subsequent case reports.

We report on a case of a 2-year-old male child who presented with a clinical picture of this disorder. In addition to bowing of the tibiae and fibulae he had complete occipitoatlantal fusion and C2-C3 fusion. Moreover, he had spina bifida occulta of L5 and absent coccyx. The overall malformation complex of the craniovertebral and pelvic abnormalities was elicited by means of three-dimensional CT scans, as plain X rays were not clear enough and could be misleading to orthopedic surgeons.

1. Ludwig-Boltzmann Institute of Osteology, Hanusch Hospital of WGKK and AUVA Trauma Centre Meidling, 4th Medical Department, Vienna, Austria.
2. Centre de Radiologie Ibn Zohr, Tunis.
3. Service d'Orthopédie Infantile, Tunis.
4. Orthopaedic Hospital of Speising, Vienna, Austria.

Manuscript received May 30 2005, accepted for publication Mar 20 2006.

**Suggested citation:** Al Kaissi A, Ben Chehida F, Gharbi H, Jinziri M, Safi H, Ben Ghachem M, et al. Craniovertebral malformation complex in a child with Weismann-Netter-Stuhl syndrome. *J Pediatr (Rio J)*. 2006;82:236-9.

We consider that our findings might represent a novel, unreported association or an unusual case of WNS syndrome, a disorder that should be differentiated from vitamin D deficiency rickets by its normal biochemical tests and the specific clinical and radiological features which were encountered.

### Clinical report

A 24-month-old male child (Figure 1) was referred to our department because of congenital, bilateral bowing of the tibia. He was born at full term, delivered by Cesarean section, with a birth weight of 3,250 g and a length of 49 cm. He was the product of an uneventful gestation, born to a 35-year-old mother who already had four children and had experienced no abortions, married to a 45-year-old unrelated man. There were no significant events in the child's early life. His developmental milestones were all within normal limits and there was no history of any significant illnesses.

At the age of two years, his height was 74 cm (below the 3rd percentile), his occipitofrontal circumference (OFC) was 50 cm (75th percentile) and his weight was 14 kg (75th percentile). Craniofacially, there was frontal bossing, a depressed nasal bridge, full cheeks, and protruding ears. His neck was short and there was a relative restriction of neck movements because of the cervical fusions. There was mild ligamentous laxity, but neither scoliosis, kyphosis, nor abnormalities of the upper limbs were noted. His neurological examination was normal, and there were no skin stigmata suggestive of a neurogenetic disorder. The child had normal genitalia. All other investigations including an abdominal ultrasound, karyotyping, and metabolic tests, which aimed to test calcium, phosphorus, and vitamin D metabolism, were normal.

### Radiographic features

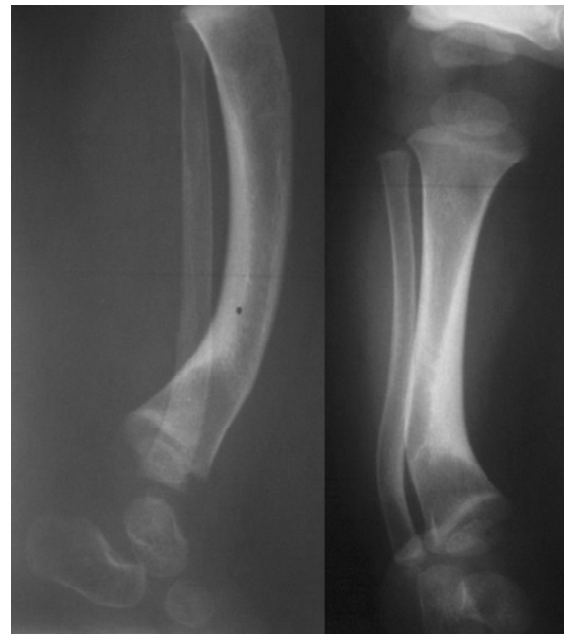
Lower limb X rays (Figure 2) showed bowing of both tibiae and fibulae with an anterior convexity and the apex of the curve was at the junction of the middle and lower thirds. The trabecular pattern of the medullary cavity of the midshaft was distorted, and posterior cortical thickening was seen. Other long bones did not show similar bowing as that seen in the tibiae and fibulae.

Cervical X rays showed overlapping of cervical vertebrae, which was extremely difficult to assess, and were not considered appropriate.

The brain CT scan revealed normal cerebral structures and no calcification was noted. Cervical spine imaging using three-dimensional reconstruction CT scanning revealed total fusion of the hypoplastic atlas to the occiput (Figures 3 and 4). The latter cervical malformation was aggravated by the simultaneous fusion of C2 and C3

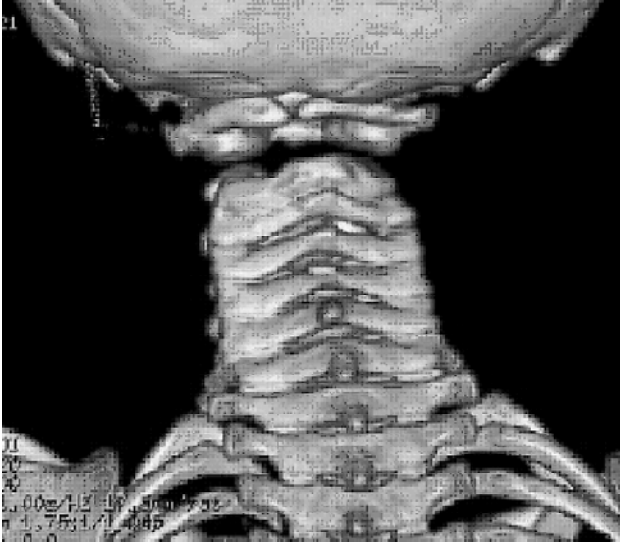


**Figure 1** - Bilateral bowing of the legs



**Figure 2** - Tibiae and fibulae showing anterior convexity and posterior cortical thickening

vertebrae, respectively. The three-dimensional CT scan of the pelvic bones showed spina bifida occulta of L5 and absent coccyx.



**Figure 3** - 3D reconstruction CT scan at the age of 3.5 years (posterior view) showing occipitalization of the hypoplastic posterior arch of the atlas, odontoid hypoplasia and further fusion of C2-C3, and congenital absence of the normal anatomical bifidity of spinous processes of C4-C6 (a sign of malsegmentation)



**Figure 4** - CT scan reconstruction (anterior view) confirming the occipitalization of the hypoplastic anterior arch of the atlas and odontoid hypoplasia and fusion of C2-C3

## Discussion

WNS syndrome usually manifests as short stature, mental retardation (in some individuals), dural calcification and anterior bowing of the tibiae. There may be bowing of long bones and a generalized diaphyseal dysplasia. The condition can be misdiagnosed as rickets.<sup>3</sup> Robinow & Johnson<sup>4</sup> described the first child with WNS syndrome in the Anglo-American literature, and Pieron et al.<sup>5</sup> reported

a child with WNS syndrome who presented with scoliosis and a more pronounced horizontalization of the sacrum. Many cases described in the literature are characterized by the presence of tibiofibular dysmorphism, most often, as the only abnormality.<sup>6</sup> Of a total of 82 cases, 14 are pediatric.<sup>7</sup> Other reported features have included communicating hydrocephalus,<sup>8</sup> marked kyphoscoliosis, costal deformity, flaring of the iliac wings and a horizontal sacrum.<sup>9</sup> Yekeler et al.<sup>10</sup> described the radiological findings of WNS syndrome in two siblings, with upper extremity involvement in one of them. No spinal involvement, as seen in our patient, was observed.

Congenital bowing of the tibia, pseudoarthrosis, and/or subperiosteal bone proliferation, can be seen in children with neurofibromatosis; however, its occurrence is mostly unilateral. Moreover, appendicular skeletal abnormalities might occur and be associated with localized gigantism. When present, intraosseous cystic lesions can progress and cause destruction of the bone shaft, with consequent deformity or instability.<sup>11</sup> In neurofibromatosis, the lateral view of the spinal column can show scalloping of the posterior borders of one or more vertebral bodies and sometimes also of the anterior borders, giving rise to a Diablo or Capstan appearance, in which a two headed top is thrown up and caught with a string stretched between two sticks, and could also be similar to the twisted ribbon appearance, in which a narrow long strip of fabric is tying something; this may be seen in the first two years of life.<sup>12</sup> The majority of these features were not present in our case. Both normal skin examination and negative family history of neuroectoderm and neural crest involvement can be sufficiently conclusive in ruling out a neurogenetic disorder.

The craniovertebral malformation complex (CVMC) can occur in a number of known syndromes (e.g.: craniosynostosis syndromes), and in association with Klippel-Feil syndrome, or as complications of Down's syndrome, and in mucopolysaccharidoses (e.g.: Morquio's syndrome). It can occur in isolation or as part of a syndromic association,<sup>13</sup> and no association with WNS has been reported. The CVMC encountered in our patient can be considered a hazardous orthopedic abnormality, and for the purpose of a comprehensive management, careful assessment should take place.

The child in this case report had the congenital, symmetrical bowing of the legs that was compatible with that one seen in WNS syndrome, but in addition, we observed congenital fusion of the occiput and the hypoplastic atlas and further fusion of C2 and C3 vertebrae, which might be the cause of the development of atlantoaxial complications. This malformation has not yet been reported in WNS syndrome. For proper management, imaging demonstration of the suboccipital region is obligatory in all patients with WNS syndrome.

## Acknowledgement

We wish to thank Dr Michael Baraitser, former consultant in Clinical and Molecular Genetics at the Institute of Child Health - University College London, for his valuable help.

## References

1. Baraitser M, Winter RM. London dysmorphology database, London medical databases, Version 1.0.4 [CD-ROM]. Oxford: Oxford University Press; 2005.
2. Weismann-Netter R, Stuhl L. D'une osteopathie congenitale eventuellement familiale. *Presse Med.* 1954;62:1618-22.
3. Caksen H, Kurtoglu S. Do not overlook Weismann-Netter syndrome in differential diagnosis of skeletal dysplasias. *Genet Couns.* 2004;15:81-5.
4. Robinow M, Johnson GF. The Weismann-Netter syndrome. *Am J Med Genet.* 1988;29:573-9.
5. Pieron R, Mafart Y, Rybojad M, Noblinsky B, Dreyfus F. Toxopachyosteosis of the tibial and fibular diaphyses (Weismann-Netter and Stuhl syndrome). *Sem Hop.* 1982;58:1043-5.
6. Zakarian H, Ceccaldi M, Acquaviva PC, Sarrat P, Maestracci D. Weismann-Netter and Stuhl disease. A review of the literature. Apropos of a case with uncommon radiologic signs. *J Radiol.* 1989;70:133-7.
7. Coimbra AV, Filardi S, Fernandes SR, Marques-Neto JF, Samara AM. Weismann-Netter-Stuhl syndrome: first Brazilian case reports. *Joint Bone Spine.* 2000;67:539-43.
8. Kurtoglu S, Dundar M, Kumandas S, Gnduz Z, Uzum K, Durak AC, et al. Patient with Weismann-Netter and Stuhl (toxopachyosteosis) syndrome with communicant hydrocephalus and arachnoid cyst. *J Pediatr Endocrinol Metab.* 2000;13:211-5.
9. Leone G. A case of Weismann-Netter and Stuhl syndrome ("toxopachyosteose diaphysaire tibio-peroniere"). *Radiol Med (Torino).* 1981;67:463-6.
10. Yekeler E, Ozdemir C, Gokalp S, Yildirim A, Bas F, Gunoz H, et al. Weismann-Netter-Stuhl syndrome in two siblings. *Skeletal Radiol.* 2005;34:176-9.
11. Boero S, Catagni M, Donzelli O, Facchini R, Frediani PV. Congenital pseudoarthrosis of the tibia associated with neurofibromatosis-1: treatment with Ilizarov's device. *J Pediatr Orthop* 1997;17:675-84.
12. Biot L, Fauchet R, Stagnara P. Les lésions vertébrales de la neurofibromatose. *Rev Chir Orthop.* 1974;60:607-21.
13. Kaissi AA, Chehida FB, Safi H, Nassib N, Ghachem MB, Gharbi H, et al. Progressive congenital torticollis in VATER association syndrome. *Spine.* 2006;31:376-8.

## Correspondence:

Dr. Ali Al Kaissi  
 Hanusch Hospital – 4th Medical Department  
 Heinrich Collin – Str. 30, A-1140 – Vienna – Austria  
 Tel.: +43-0-1-91021-86724  
 Fax: +43-0-1-91021-86929  
 E-mail: ali.alkaissi@osteologie.at