



Case Report

Weismann-Netter-Stuhl Syndrome: A rare form of skeletal dysplasia

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ABSTRACT: A case of Weismann-Netter-Stuhl syndrome was described because of its rare entity in pediatric age group. The clinical presentation was short stature with bowing of lower legs with characteristic radiological findings. It was differentiated from congenital syphilis and rickets by negative laboratory results and the characteristic radiographic features of anterior and medial bowing of the mid to distal third of the tibiae and fibulae and cortical hyperostosis of the concave margins.

KEY WORDS: Weismann-Netter-Stuhl syndrome; Skeletal dysplasia

INTRODUCTION

Weismann-Netter-Stuhl Syndrome is a rare heritable skeletal dysplasia, which often presents as asymptomatic bowing of lower leg or short stature.¹ This condition is usually bilateral and not related to healed rickets. Although more than 40 cases are described, there is mention of only 8 cases in pediatric age group who are younger than 16 years.¹⁻¹² It was first described in 1954 by Weismann-Netter and Stuhl.

CASE REPORT

This is a case report of a 14 year old female child who presented in the out patient department with the chief complaint of short stature. She was a product of uncomplicated pregnancy delivered by normal vaginal delivery at home. According to her mother all milestones were normal as per the age. There was no teratogenic exposure or consanguinity. Ambulation was delayed until 3 year of age but other developmental milestones were normal. Bowing of the lower extremities was noted at the age of 5 years. There had been no fractures or other significant medical history. The

mother was 42 years of age and 162 cm tall. The father was 48 years of age and 181 cm tall. There were 4 siblings who were all of normal stature and without clinical features of this disorder.

Physical examination revealed a proportionate height of 149 cm (5th percentile), a weight of 40 kg (10th percentile). The height and arm span were equal. The upper segment-lower segment ratio (1.12) was normal. Physical examination results were normal except for the presence of mild medial bowing of both thighs and lower legs, and mild anterior bowing of both lower legs (**Figure 1**). There were no dysmorphic facial features.



Figure 1: Showing medial bowing of both thighs and lower legs; mid anterior bowing of both lower legs

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Radiographic features (**Figure 2**) confirmed the anterior and medial bowing of the mid to distal third of the tibiae and fibulae and showed cortical hyperostosis of the concave margins. Additional features included accentuation of the bony trabeculae in the medullary cavity of the tibiae. The anterior tibial cortex was of normal thickness. The hands had a normal bone age (according to the method of Greulich and Pyle).¹⁰ Laboratory studies disclosed normal serum calcium, phosphorus, total protein, and 25-OH-cholecalciferol values. Serologic studies for syphilis (rapid plasma reagin) were negative. Serum alkaline phosphatase was also normal. This case served to heighten our awareness of this condition, which should be recognizable in the pediatric age range.



Figure 2: Radiograph showing the anterior and medial bowing of the mid to distal third of the tibiae and fibulae with cortical hyperostosis of the concave margins

DISCUSSION

The Weismann-Netter-Stuhl syndrome was first described in 1954.¹ Since that time, there have been more than 40 additional reports, but mention of only 8 children^{1,3,9}. Only 1 report of an affected child has appeared in the English literature³. Most of the adults with Weismann-Netter-Stuhl syndrome reported a delay in ambulation and bowing of the lower extremities for which there was no diagnosis during early life.³ This latter observation suggested that, in addition to ourselves, others may be unfamiliar with this syndrome. The thickening of the fibula is true "tibialisation" and is the main feature and the only feature confirming diagnosis.¹³

In the present case, the diagnosis of Weismann-Netter-Stuhl syndrome was suggested by the presence of two reliable diagnostic features. The first of these was

symmetric anterior and medial bowing of the tibiae and fibulae, located at the junction of the middle and lower thirds of the diaphyses. The second was cortical hyperostosis of the posterior concave surface with disruption of the trabecular pattern in the region of curvature. In the present case, growth velocity was normal. Antero-posterior radiograph of both lower extremities showed medial bowing of lower one third of the tibiae. There was medial cortical thickening. Lateral radiographs of right and left lower extremities demonstrated anterior bowing of lower one third of tibiae with posterior cortical thickening. Autosomal dominant inheritance has been proposed for this syndrome and would be consistent with the pedigree of the present case. This child had 3 unaffected siblings and a father of relatively advanced age, both of which suggest a new autosomal dominant mutation.

Cases of Weismann-Netter-Stuhl syndrome involving the upper extremities and affecting siblings have rarely been reported. In the literature there is a case report of the presence of radiological findings of Weismann-Netter-Stuhl syndrome in two siblings; with upper extremity involvement in one of them.¹⁴ Most of the reported cases had normal IQ levels. There is an unusual case report of a child with clinical and radiological features of this syndrome with complete occipitoatlantal fusion, C2-C3 fusion, spina bifida occulta of L5 and absent coccyx.¹⁵ There is also a case report of patient with Weismann-Netter-Stuhl syndrome with an unusual manifestation of communicant hydrocephalus and arachnoid cyst.

Table 1 shows the summary of clinical and radiological features present in our case and other studies^{1,3,9,11}.

CONCLUSION

The Weismann-Netter-Stuhl syndrome is a rare, heritable skeletal dysplasia which may present in the pediatric age range with bowing of the extremities, a delay in ambulation, or short stature. It should be differentiated from congenital syphilis and rickets by negative laboratory results and the characteristic radiographic features. The persistently elevated alkaline phosphatase levels, abnormal lower leg radiographs, exaggeration of bowing throughout the pubertal growth spurt, and more pronounced short stature in adult life all suggest that the Weismann-Netter-Stuhl syndrome is an ongoing generalized skeletal dysplasia during childhood.

Table-1 Clinical and Radiological features of Weismann Netter Stuhl Syndrome

Clinical features	Present Case 14yrs/F	Francis et al ¹¹ 4yrs/M	Roninow et al ³ 8yrs/M	Weismann et al ^{1,9} (5 cases)				
				16yrs F	10yrs M	15yrs M	10yrs F	8yrs F
Bowed lower extremity	+	+	+	+	+	+		+
Short stature	+	-	+	+	-	-	+	+
Ambulation delay	+	+	+	+	+	+	+	+
Kyphoscoliosis	-	-	-	-	-	+	+	-
Family history	-	-	+	+	+	+	+	+
Mental retardation	-	-	+	-	-	-	+	+
Goiter	-	-	-	-	-	-		
Pernicious anemia	-	-	-	-	-	-		
Language delay	-	-	+	-	-	-		
Hyperphosphatasemia	-	+	+	-	-	-		
(N) fracture healing		+	+					
(N) growth velocity	+							
Aminoaciduria		+/-	+/-					
Delayed dental eruption	-	+	+	-	-	-		
Radiographic								
Bilateral bowing of tibia and fibula	+	+	+	+	+	+	-	-
Unilateral bowing of tibia and fibula	-	-	-	-	-	-	+	+
Squared pelvis	-	-		-	-	-		
Bowed femora	-	-	-	-	-	-		
Normal skull	+		+	+	+	+		
Bowed radii ulni	-		+	-	-	-		
Bowed humeri	-		+	-	-	-		
Exaggerated trabeculation	-	+	+	-	-	-		
Thin posterior ribs	-	-	+	-	-	-		
Coxa vara	-	+		-	-	-		
Horizontal sacrum	-			-	-	-		
Sacralization of L5	-	+		-	-	-		
Low set L5	-	+		-	-	-		

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