Indian J Pediatr
DOI 10.1007/s12098-016-2182-3

SCIENTIFIC LETTER

Williams-Beuren Syndrome with Mirror Movements

Sedat Işıkay[1] · **Sadettin Sezer**[2] · **Serkan Kırık**[3] · **Olcay Güngör**[3]

Received: 25 January 2016 / Accepted: 9 June 2016

To the Editor: Williams-Beuren Syndrome (WBS) is a rare neurodevelopmental disorder caused by deletion of chromosome 7 at q11.23. It is characterized by distinctive facies, congenital cardiovascular malformations, intellectual disabilities, and various other manifestations [1, 2]. Here, we present a child with WBS who presented with abnormal involuntary hand movements.

An 8-y-old boy presented with history of simultaneous, involuntary movements of left hand along with voluntary movements of the opposite hand for about 7 y. He had difficulty in writing, drawing, pushing buttons, using scissors and eating because of involuntarily movements of the left hand. The perinatal history was unremarkable. The parents described developmental delay in his motor, social and language skills since infancy. He was diagnosed with supravalvular aortic stenosis at the age of 3. His weight, height, and head circumference were on 50th percentile. Dysmorphic facial features including puffy eyes, full cheeks, flat midface, epicanthic folds, narrow and high palate, wide mouth with thick lips, and long philtrum were noted in physical examination (Fig. 1). During examination, involuntary, simultaneous movement of the left hand whenever the right hand was moved were noted. Routine blood and metabolic tests and brain and cervical vertebra magnetic resonance imaging were normal. Fluorescent in situ hybridization study revealed deletion of 7q11.23 region and he was diagnosed with WBS.

Neurologic problems in WBS vary and include mental retardation, delayed developmental milestones, and visual spatial deficits [1, 2]. To the best of our knowledge, only two cases about the co-existence of mirror movements with

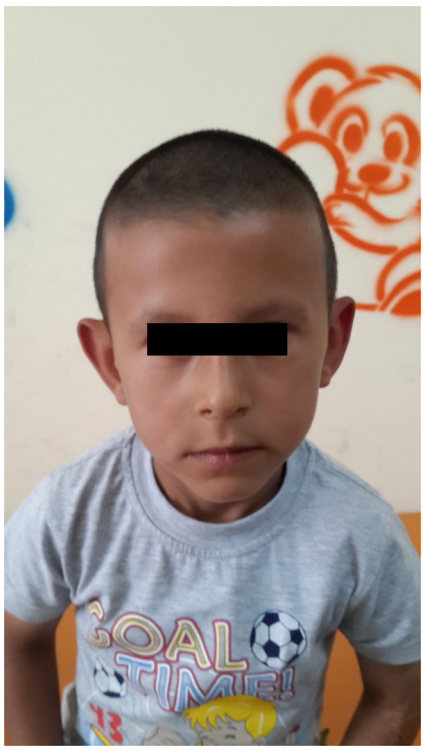

Fig. 1 The facial appearance of the patient

✉ Sedat Işıkay
dr.sedatisikay@mynet.com

[1] Department of Pediatric Neurology, Hasan Kalyoncu University, 27000 Gaziantep, Turkey

[2] Department of Pediatric Cardiology, Gaziantep Children's Hospital, Gaziantep, Turkey

[3] Department of Pediatrics, Kahramanmaraş Sütçü İmam University, Kahramanmaraş, Turkey

Published online: 15 June 2016

WBS have been reported before and this is the third one [1, 3]. Infrequently, mirror movements may accompany WBS.

Compliance with Ethical Standards

Ethical Statement Parents' consent for publication is obtained.

Conflict of Interest None.

Source of Funding None.

References

1. Glós J, Bzdúch V, Lisy L, Jariabková K. Large mirror movements of upper extremities in Williams syndrome. Pediatr Neurol. 1993;9: 414.
2. Pober BR. Williams-Beuren syndrome. N Engl J Med. 2010;362: 239–52.
3. Revol P, Jacquin-Courtois S, Delporte L, et al. Kinematics analysis of mirror movements in a case of Williams and Beuren syndrome. FENS Abstr. 2008;4:119.33.