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Research Article

Mowat Wilson Syndrome Associated With Pseudo Rocker Bottom Feet Deformity

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Abstract

Background: Mowat Wilson syndrome was probably first described in a 2.5-year old boy by Lurie et al (1994) from Baltimore School of Medicine. However, the syndrome was named after Mowat et al (1998) who provided a detailed description of six children with syndrome and discussed the occurrence of similar syndromes in the literature.
Mowat Wilson syndrome is a mental and growth retardation syndrome associated with distinctive facial dysmorphism consisting of deep set large eyes, hypertelorism, and open mouthed expression most of the time, low set ears, and other congenital abnormalities including agenesis of the corpus callosum and skeletal deformities. The syndrome is commonly associated with Hirschsprung disease or chronic constipation. In the hands of experts, the distinctive facial features allow the clinical diagnosis of Mowat-Wilson syndrome.
The aim of this paper to report a study of the first case of Mowat syndrome in Iraqi boy which was associated with pseudo rocker bottom deformity of the feet.
Materials and methods: A sixteen months old boy with Mowat syndrome is studied and the literature reporting the syndrome was studied.
Results: The boy was the first child of a non-consanguineous healthy parents. He had psychomotor retardation, poor feeding and poor growth, and chronic constipation during infancy. Distinctive facial dysmorphism included deep set large eyes, hypertelorism, open mouthed expression most of the time, low set ears, and also high arched palate. Skeletal deformities included congenital dislocation of the right hip and pseudo rocker bottom feet deformity. Brain MRI showed agenesis of the corpus
Conclusion: The 383 case of Mowat syndrome is an Iraqi boy who had an unusual deformity of the feet "Pseudo rocker bottom feet" which has not been reported before with this syndrome.

Keywords: Mowat Wilson syndrome; hip dislocation; pseudo rocker bottom feet deformity

Introduction

Mowat Wilson syndrome was probably first described in a 2.5 year old boy by Lurie et al (1994) from Baltimore School of Medicine. However, the syndrome was named after Mowat et al (1998) who provided a detailed description of six children with syndrome and discussed the occurrence of similar syndromes in the literature.

Mowat Wilson syndrome is a mental and growth retardation syndrome associated with distinctive facial dysmorphism consisting of deep set large eyes, hypertelorism, open mouthed expression most of the time, low set ears, and other congenital abnormalities including agenesis of the corpus callosum and skeletal deformities. The syndrome is commonly associated with Hirschsprung disease or chronic constipation [1,2,3].

In the hands of experts, the distinctive facial features allow the clinical diagnosis of Mowat Wilson syndrome[4].

The aim of this paper to report a study of the first case of Mowat syndrome in Iraqi boy which was associated with pseudo rocker bottom deformity of the feet.

Materials and methods

A sixteen months old boy with Mowat syndrome is studied and the literature reporting the syndrome was studied.

Results

HZ was first seen at the age of sixteen months during September 2019, because of psychomotor retardation, poor feeding and poor growth (weight 7.5 kilograms). He was not controlling his neck when sitting on the chair and was unable to crawl nor was saying any word. The mother was told after the birth of her boy that he experienced birth asphyxia. In addition, the child was well-known to have congenital dislocation of the hip that has not been treated for no satisfactory reasons. The boy suffered from chronic constipation during early infancy which was improved before the end of the first year. The boy was the first child of a non-consanguineous healthy parents. He had a younger brother aged two months and was considered by the mother to be healthy. The mother like the previously consulted doctor didn't suspect a dysmorphic or a genetic disorder.

At the clinic the boy was unable to control his head when sitting on the chair and showed poor awareness to the surroundings (Figure-1). He had

obvious facial dysmorphism consisting of deep set large eyes, hypertelorism, open mouthed expression most of the time, low set ears, and also high arched palate (Figure-2). The clinically apparent dislocation of the right hip was confirmed by radiographs (Figure-3).The boy also had slender, tapered fingers, and bilateral foot deformity similar to the rocker bottom feet which is commonly associated with Edwards' syndrome (Figure-4).

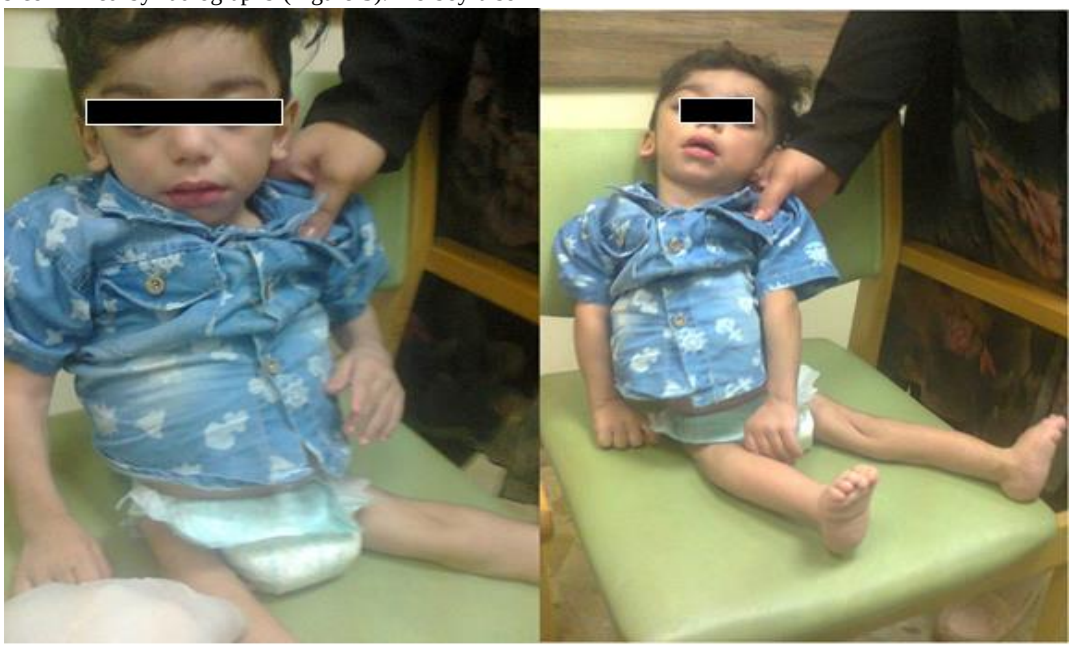

Figure-1: At the clinic the boy was unable to control his head when sitting on the chair

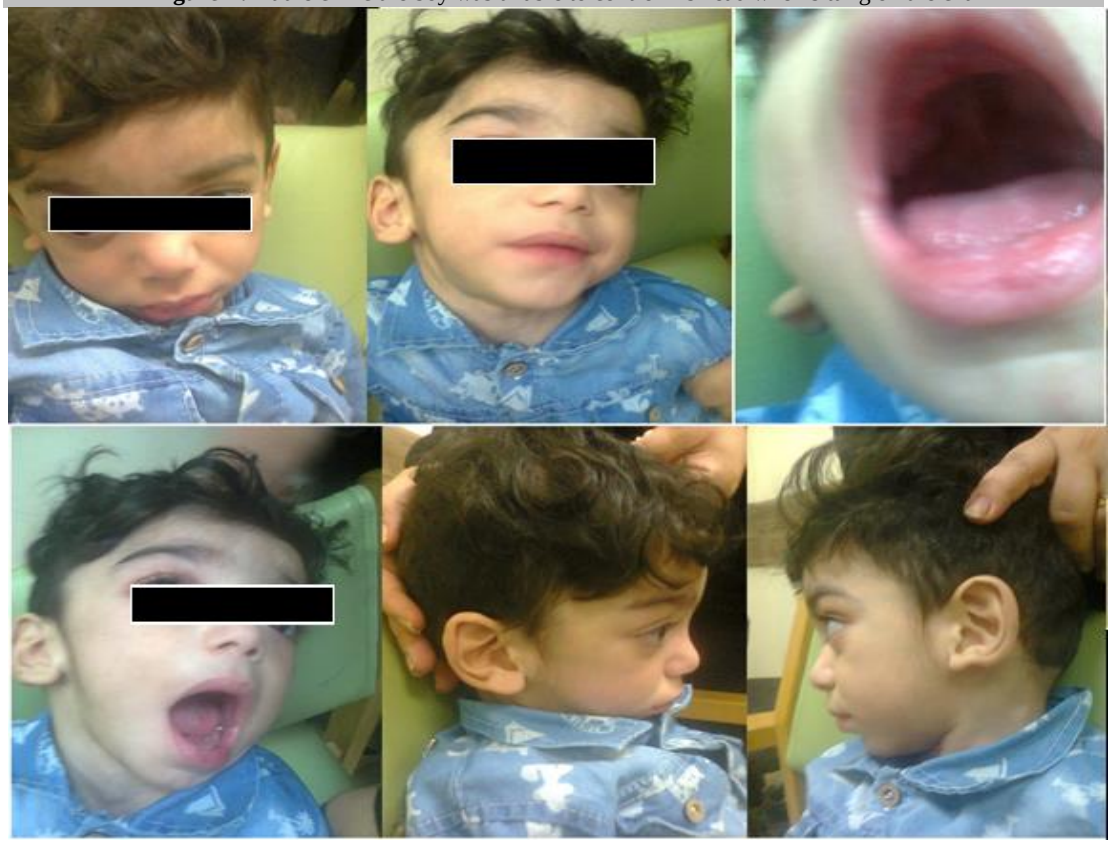

Figure-2: The boy had obvious facial dysmorphism consisting of deep set large eyes, hypertelorism, open mouthed expression most of the time, low set ears, and also high arched palate

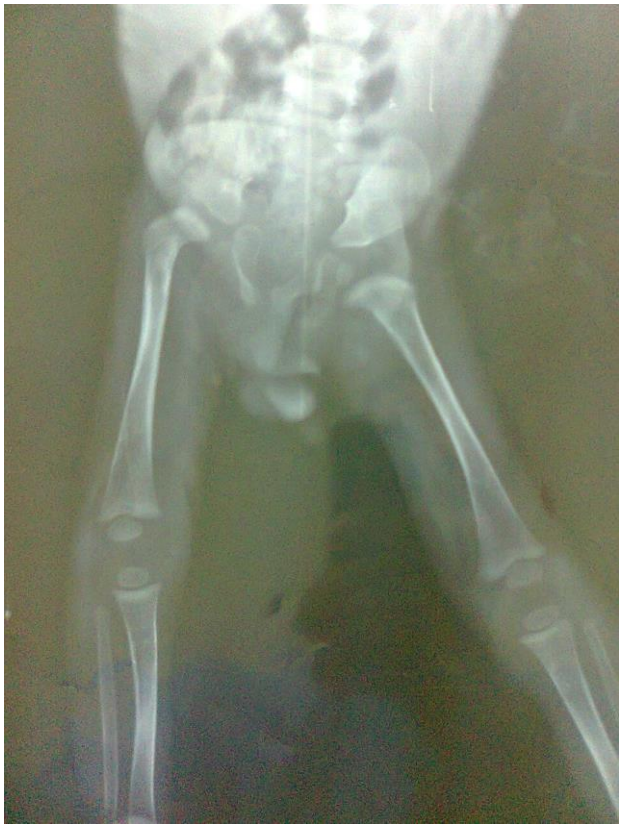

Figure-3: Advanced developmental dislocation of the right hip with superior lateral displacement and pseudo articulation of ileum

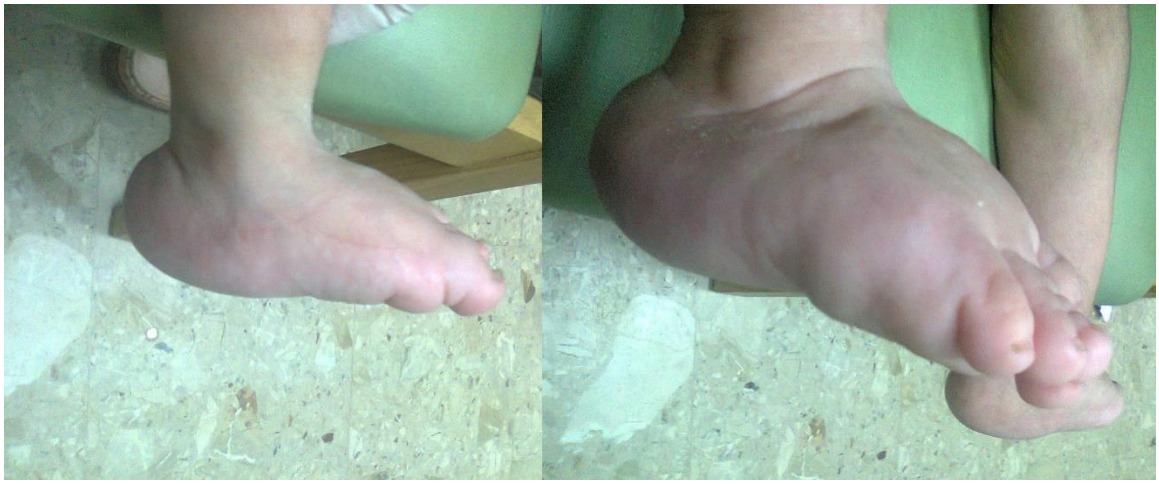

Figure-4: The patient had bilateral foot deformity similar to the rocker bottom feet which is commonly associated with Edward syndrome

Rocker bottom foot which is also called congenital vertical talus, is characterized by a prominent calcaneus (heel bone) and a convex rounded bottom of the foot making the foot similar to the bottom of a rocking chair. However, radiographs showed normal positioning of talus (Figure-5), and the deformity represents a pseudo rocker bottom feet

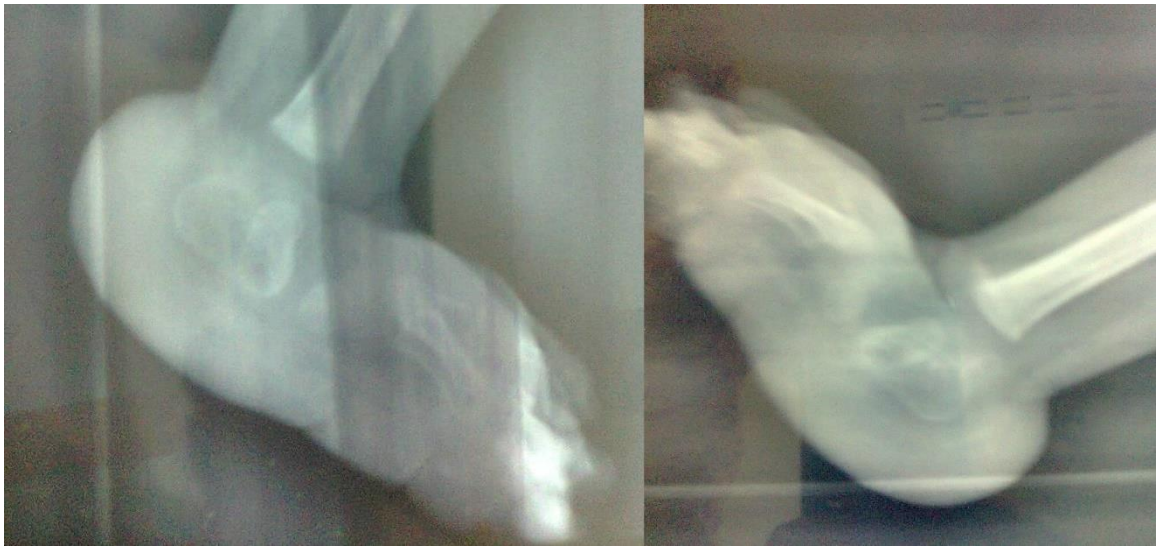

Figure-5: Radiographs showing normal positioning of talus, and thus the deformity represents a pseudo rocker bottom feet

Brain MRI performed at the age of to 70 days showed changes suggestive of diffuse hypoxic ischemic encephalopathy including periventricular leukomalacia with multiple brain hemorrhages mainly in the right occipital lobe, right parietal lobe, and both thalmai. The MRI also showed evidence of diffuse brain atrophy with abnormal signals in the thalami and basal ganglia.

Brain MRI performed before about one year showed agenesis of the corpus callosum and multiple parenchymal hemorrhages on the basal ganglia and right parietal, temporal lobes.

Discussion

Mowat et al (1998) from Australia described six children with a distinctive facial features with deep set large eyes, hypertelorism, open mouthed expression most of the time, mental and growth retardation. Five of the children reported by Mowat et al had Hirschsprung disease including four with short segment disease and one had chronic constipation but a normal rectal biopsy. The patients also had musculoskeletal features including slender, tapered fingers, and bilateral calcaneovalgus foot deformity. Three of the six patients had congenital heart defect and three had renal structural abnormalities. Four patients had cerebral ultrasound in early infancy suggesting complete or partial agenesis of the corpus callosum, but later cerebral CT was normal in two patients and MRI showed complete agenesis of the corpus callosum in the other two patients.

Zweier et al (2002) from Germany reported four patients with Mowat Wilson syndrome, making the total number of the reported cases eleven, and suggested the emergence of a specific clinical dysmorphic mental retardation syndrome. Two of the patients of Zweier et al had Hirschsprung disease.

The syndrome has been reported from many countries including USA (Lurie et al, 1994) , Australia (Mowat et al, 1998), Germany (Horn et al,2004), France (Cacheux et al,2001), Italy (Cerruti Mainardi et al ,2004) , Japan (Ishihara et al,2005), Croatia (Sasso et al,2008), Denmark (Engenheiro,2008), Serbia (Cuturilo et al,2009), Poland (Smigiel,2019), United Kingdom (Leong, 2010), Malaysia (Balasubramaniam et al, 2010), Indonesia (Mundhofir et al,2012), Turkey (Meral et al,2012), South Korea (Park et al,2013), Brazil (Valera et al,2013), Cyprus (Tanteles and Christophidou-Anastasiadou (2014), Egypt (Abdalla and Zayed , 2014), Spain (Ferris Villanueva and colleagues ,2015), Ireland (Coyle and Puri ,2015), India (Deshmukh et al, 2016), China (Jiang et al,2016), Slovenia (Rogac and colleagues, 2017), Colombia (Cano Sierra and colleages, 2018), Argentina (Gosso et al,2018) [1-27].

Balasubramaniam et al (2008) from Malaysia [13] and Nissen et al (2011) from Denmark [14] estimated that about 180 cases of Mowat Wilson syndrome have been reported in the medical literature .Coyle and Puri (2015) from Ireland estimated 256 patients with Mowat Wilson syndrome were reported in fifty-two published articles [22]. Therefore, cases of Mowat Wilson syndrome have been reported including 35 cases reported by Evans et al (2016) [28], Six cases reported by Kilic et al (2016) [29], three cases reported by Moore et al (2016) [30], one case reported by Deshmukh et al (2016) [23], one case reported by Packiasabapathy et al (2016} [31], 34 cases reported by Niemczyk et al (2017) [32], seven patients reported by Bonanni et al (2017) [33], one patient reported by Baxter et al (2017) [34], two cases reported by Seo et al (2017) [35], one patient reported by Rogac and colleagues (2017) [25], 11 patients reported by Spunton et al (2018) [36], one patient reported by Cano Sierra and colleages (2018) [26], two patients reported by Gosso et al (2018) [27], one patient reported by Nevarez Flores and colleagues (2019) [37], one patient reported by Wag et al (2019) [38], Three cases reported by Wang et al (May,2019) [39], and 16 cases reported by Di Pisa et al (2019) [40].

The total number of the previously reported patients with Mowat Wilson syndrome in the medical literature is 382.

Conclusion: The 383 case of Mowat syndrome is an Iraqi boy who had an unusual deformity of the feet "Pseudo rocker bottom feet" which has not been reported before with this syndrome.

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