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First Turkish Patient with Floating Harbor Syndrome with Additional Findings: Cryptorchidim and Microcephaly

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We report the first Turkish patient with Floating Harbor Syndrome (FHS). The 12-year old male patient exhibited classical dysmorphic features of FHS, mental retardation, celiac disease and additional undescribed findings: microcephaly and cryptorchidism.

Key Words: Cryptorchidism, floating-harbor syndrome, and microcephaly

INTRODUCTION

Floating-Harbor Syndrome (FHS) is a very rare disorder characterized by short stature, delayed language skills, and a triangular shaped face. A prominent nose, deep-set eyes and a wide mouth with thin lips give the affected patient a distinct appearance.[1] The described features of thirty patients with FHS reported in the literature are shown in Table 1.[1-10] We present here the first Turkish patient with FHS, who exhibited additional features - cryptorchidism and microcephaly- not previously reported.

CASE REPORT

A 12-year-old boy was admitted to the hospital with complaints of chronic diarrhea for five years and growth retardation. He was the fifth child of

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Table 1. The Features of Floating Harbor Syndrome

Described features	Present case
Short stature	+
Triangular face	+
Posteriorly rotated/placed ears	+
Deep-set eyes, long eyelashes	+
Strabismus	-
Prominent/bulbous nose	+
Wide columella, smooth philtrum, thin lips	+
Downturned mouth	-
Supernumerary upper incisor	-
Short neck	-
Low posterior hairline	-
Celiac disease	+
Delayed bone age	+
Joint laxity	-
Fifth finger clinodactyly	+
Brachydactyly	-
Clubbed fingers	-
Wide thumbs	-
Sprengel deformity	+
Pseudoarthrosis clavicle	-
Hirsutism	-
Expressive language delay	-
High-pitched voice	-
Hypernasal speech	+
Mental retardation	+
Normal motor development	+
Cardiac septal defect	-
Genitourinary defect	+

a consanguineous marriage. No reliable history about his natal and postnatal period was available. He had a history of normal motor development and had undergone a surgical operation for cryptorchidism. On examination, his weight and height were below the bottom 3rd percentile (- 3.9 and -4.7 S.D, respectively), and his occipitofrontal circumference (OFC) was below 3 S.D. for his age and sex: he had microcephaly. He had a triangular face, deep-set eyes, long lashes, prominent nose, wide columella, smooth philtrum and thin lips (Fig. 1). Hypernasal speech, posteriorly placed ears, fifth finger clinodactyly, broad elbows and ankles, and Sprengel deformity were also detected. The results of neurological and ophtalmological examinations were normal. His bone age was equivalent to that of an 8 year-old. Complete blood count, serum electrolytes, transaminases, albumin, thyroid function tests and urinalysis were all normal. Anti-gliadin IgA, anti-gliadin IgG and anti-endomysial IgA antibodies were positive. Upper gastrointestinal endoscopy revealed antral gastritis with positive rapid urease test for Helicobacter pylori. In duodenal biopsy, total villous atrophy, crypt hyperplasia and increased intraepithelial lymphocytes were observed; these findings were consistent with celiac disease. Magnetic Resonance Imaging of the brain and echocardiography were normal. The patient's IQ was 64.

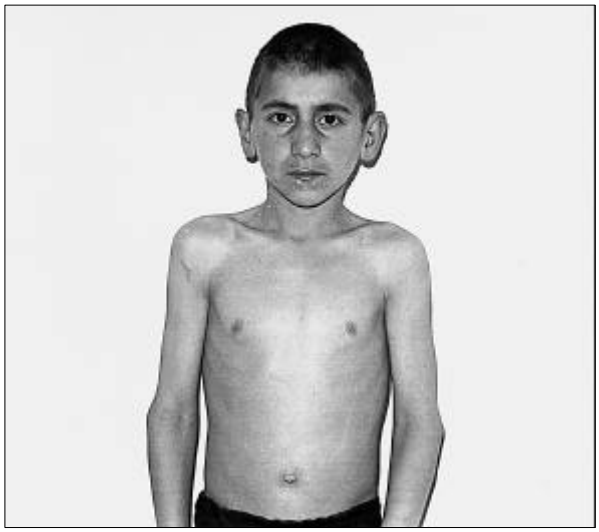

Fig. 1. Frontal view of the patient with FHS. Note the triangular face, deep-set eyes, prominent nose and smooth philtrum.

DISCUSSION

Most of the characteristic findings described for FHS were present in our case. The peculiar faces-triangular in shape, with deep-set eyes, long lashes, prominent nose, smooth philtrum, wide columella, and thin lips - were striking. The short stature associated with delayed bone age, hypernasal speech and celiac disease all permitted us to establish the diagnosis of FHS. So far, thirty patients have been reported in the literature. The complaint that had taken our patient to the doctor was the chronic diarrhea from celiac disease. Chudley and Moroz[2] first reported the case of a 17-year-old girl with celiac disease, whose features were consistent with FHS. Ala-Mello and Peippo[11] suggested that all patients with this syndrome should be examined for celiac disease.

The additional features we detected were microcephaly and cryptorchidism. Since these two features are also elements of Rubinstein-Taybi Syndrome (RTS), differential diagnosis with the mentioned syndrome was very important.[5] RTS is a rare genetic multisystem disorder that affects many organ systems of the body. The findings associated with this syndrome include growth retardation and delayed bone age; mental retardation; craniofacial dysmorphism, including hypertelorism, a broad nasal bridge, and an abnormally large or "beak-shaped" nose; abnormally broad thumbs and great toes. In addition, most affected children experience motor development, which is not a described feature of FHS.[1] In our case, no history of motor development delay was available and craniofacial dysmorphism, other than microcephaly, was suggestive of FHS. Our patient had no highly-arched palate, micrognathia, strabismus, ptosis, downwardly slanting eyelid folds or epicanthal folds. Furthermore, no malformations of the heart or kidneys were detected. While GIS manifestation of RTS is constipation, in FHS, celiac disease is a common disorder.[2] The OFCs of the patients reported in the literature were at or below the 5th percentile, but their heights were as well.[2,12] In our case, OFC was small for his height; thus, he had microcephaly.

Undescended testicles in males is a common feature in RTS, but has not been identified as an element of FHS. However, since genitourinary

defects such as small penis and renal abnormalities were reported in FHS, cryptorchidism may be seen as well.[8]

Consequently, to the best of our knowledge, this is the first FHS case presented with cryptorchidism and microcephaly. Since FHS is not a common genetic syndrome and there are only 30 cases that have been reported in the literature, it is reasonable to anticipate that further dysmorphic features will be described in the future.

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