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Facial Dysmorphism, Mental Retardation, Triphalangeal Toes and Unilateral Renal Agenesis: A New Syndrome

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Abstract

A congenital syndrome is suspected when there are more than three minor anomalies which are variations of normal morphological features that are considered of little or no known medical, surgical, or cosmetic significance. More than one major anomaly which is an abnormality that has major medical, surgical or cosmetic significance, and one major anomaly with two or more minor anomalies are also suggestive of congenital syndrome. There are a large number of syndromes associated with various combination of facial dysmorphism, mental retardation, limb abnormalities, and unilateral renal agenesis. The aim of this paper is to report the occurrence of a new mental retardation dysmorphic syndrome with the novel occurrence of unique triphalangeal toes abnormalities in association with left renal agenesis and distinctive facial features including hypertelorism and absent naso-frontal angle.

Key Words: Mental retardation, triphalangeal toes, left renal agenesis, hypertelorism, absent naso-frontal angle.

Introduction

A congenital syndrome is suspected when there are more than three minor anomalies which are variations of normal morphological features that are considered of little or no known medical, surgical, or cosmetic significance. More than one major anomaly which is an abnormality that has major medical, surgical or cosmetic significance, and one major anomaly with two or more minor anomalies are also suggestive of congenital syndrome [1,2,3,4,5]. There are a large number of syndromes associated with various combination of facial dysmorphism, mental retardation, limb abnormalities, and unilateral renal agenesis [5,6,7]. The aim of this paper is to report the occurrence of a new mental retardation dysmorphic syndrome with the novel occurrence of unique triphalangeal toes abnormalities in association with left renal agenesis and distinctive facial features including hypertelorism and absent naso-frontal angle.

Case report

M.A was first seen at the age of two years and seven month during August, 2019. The parents brought him to the pediatric neuropsychiatric clinic because he was not saying any word and unable to walk. The mother also complained that the boy was drooling and unable to feed himself with a spoon.

The boy had distinctive facial features characterized by hypertelorism, broad nasal bridge, and absent naso-frontal angle [Figure-1].

Figure-1: The boy had distinctive facial features characterized by hypertelorism, broad nasal bridge, and absent naso-frontal angle

His social interaction was and eye contact was good but he was not saying any word and was unable to walk. The father helped him to stand holding furniture but he couldn't walk (Figure-2).

Figure-2: The father helped him to stand holding furniture, but he couldn't walk

The also boy had triphalangeal toes (Figure-3) and long spindle fingers (Figure-4).

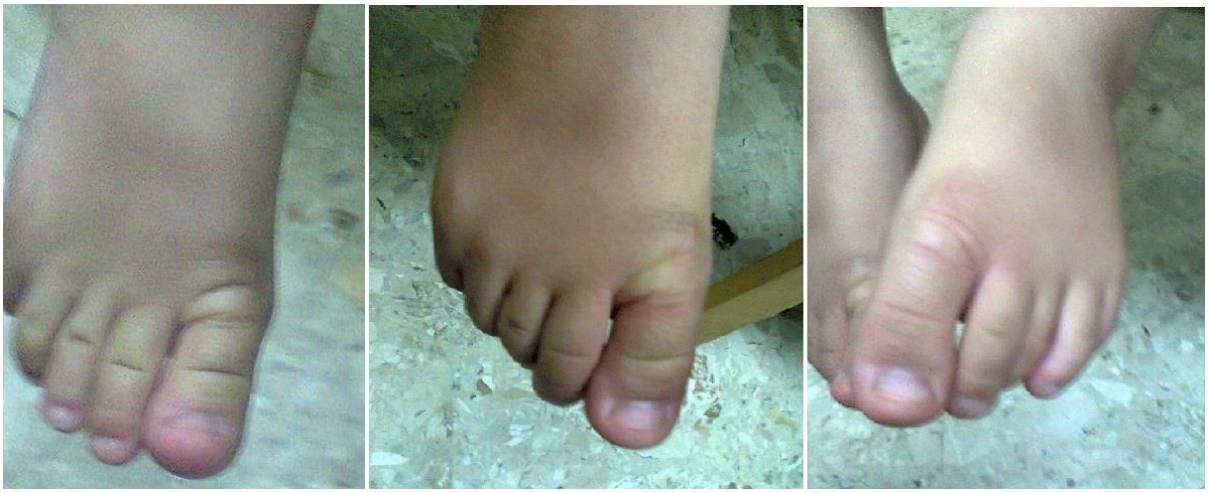

Figure-3: The boy had triphalangeal toes

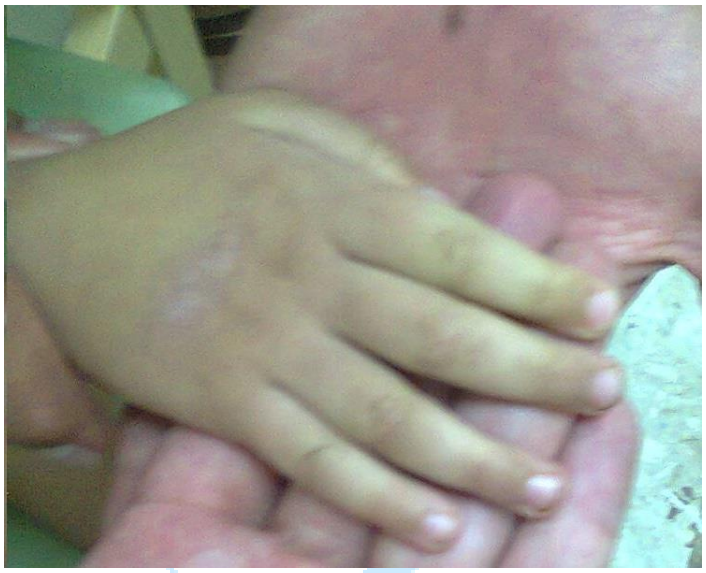

Figure-4: The boy had long spindle fingers

Family history was negative for any similar disorder, but the parents were consanguineous, and both had long toes. As the mother reported that she had oligohydramnios during pregnancy, the boy was sent for abdominal ultrasound which showed left renal agenesis.

Discussion

The increasing number of congenital syndromes demanded the evolution of approaches for their clinical recognition and diagnosis. It is generally recommended to make a list of the anomalies in the patient that are likely to be more specific, followed by listing the possible syndromes. Finally, the most probable diagnosis can be reached by narrowing of the diagnostic possibilities depending on the combination of anomalies the patient has [3,4]. There are a large number of syndromes associated with various combination of facial dysmorphism, mental retardation, limb abnormalities, and unilateral renal agenesis. However, in this paper a patient with novel association of facial dysmorphism with absent naso-frontal angle, mental retardation, triphalangeal toes, and left renal agenesis is reported.

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