Megarbane-Loiselet neonatal progeroid syndrome; a rare cause for neonatal progeroid syndrome

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Abstract

Premature ageing syndromes are a rare group of disorders. True premature ageing, skin atrophy/ lipoatrophy, cutis laxa or simply an older appearance than would be expected for the person's age lead to a this phenotype. Many of these conditions share common clinical manifestations which can make difficulty in precise diagnosis[1]. Conditions with lipoatrophy are also described as neonatal progeroid syndromes. In this case report we describe a 2 month old child with manifestations of Megarbane-Loiselet neonatal progeroid (MLNP) syndrome, a neonatal progeroid syndrome for its rareness.

Introduction

Neonatal progeroid syndrome is described under premature ageing syndromes. Weidemann Rautenstrauch, DeBarsy syndrome, Acrometageria, Mandibuloacral dysplasia, Megarbane-Loiselet neonatal progeroid syndrome, Penttinen progeroid disorder, Lenz-Majewski syndrome, Mulvihill-Smith syndrome and Lenaerts syndrome are described under Neonatal progeroid syndrome[1]. Wiedemann-Rautenstrauch syndrome is commonly described in literature[2,3,4]. DeBarsy syndrome and Megarbane-Loiselet neonatal progeroid syndrome are two other conditions described under neonatal progeroid syndrome[1,5,6,7]. The child we report here has more clinical features suggestive of Megarbane-Loiselet neonatal progeroid syndrome with DeBarsy syndrome as a differential diagnosis.

Case report

A two month old baby boy was referred from a pediatric clinic with subcutaneous lipoatrophy and joint contractures. He was the second child born to non-consanguinous parents. His elder sibling was healthy. Antenatal scan revealed mesomelic shortening of long bones. His birth weight was 2.750 kg but he had severe failure to thrive postnataly.

Clinical examination revealed facial dysmorphic features including frontal bossing, hypertelorism, low set ears, pinched nose with flat nasal bridge and micrognathia. He also had sparse hair, prominent abdominal veins with loose pendulous skin (cutis laxa) mainly in extremities (Figure 1). He also had bilateral joint contractures at wrist joints, 3rd and 4th metacarpophalengeal joints and bilateral thumb adduction (Figure 2).

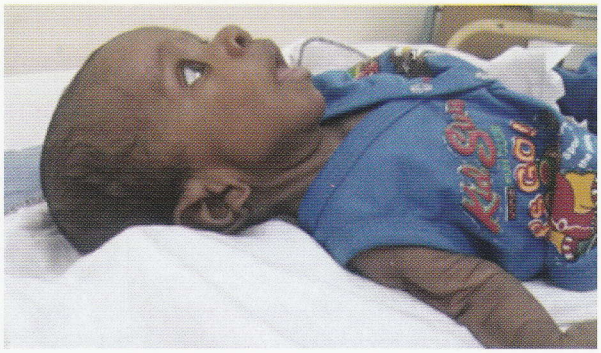

Figure 1.

Figure 2.

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Discussion

Megarbane-Loiselet neonatal progeroid syndrome is a rare premature ageing syndrome. It is defined as a condition characterized by neonatal progeroid phenotype, joint contractures, thumb adduction, inguinal hernia and early death. Inheritance is probably autosomal recessive and basic gene defect is unknown[1]. Insufficient production of type I and III procollagens are demonstrated in patients with this condition. Affected individuals have pre- and postnatal growth retardation; a characteristic face with hypertelorism, pinched nose, small mouth and micrognathia; sparse hair; thin skin on face and scalp; prominent occiput, cataracts, adduction of thumb and cardiac defects. The child presented to us had similar facial dysmorphic features, cardiac defects and thumb adduction which are features of MLNP syndrome. Among those clinical manifestations thumb adduction highly resembles MLNP syndrome. The life span is usually less than one year in patients with MLNP syndrome. In most cases cause of death was related to cardiac defects[1]. Intrauterine growth retardation is usually found in neonatal progeroid syndromes[1,2,7]. The patient described had a normal birth weight which is not described in patients with MLNP syndrome.

Facial dysmorphism in MLNP syndrome and presence of cutis laxa in extremities are also seen in DeBarsy syndrome. Corneal clouding and athetoid movements are two characteristic features of DeBarsy syndrome[1,5]. In the absence of those characteristic features, DeBarsy syndrome was excluded.

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