

Plantar Lipomatosis, Unusual Facial Phenotype and Developmental Delay: A New MCA/MR Syndrome

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We describe two boys with global developmental delay and a phenotype of microcephaly, midface hypoplasia, enlarged fleshy ears, depressed nasal bridge, anteverted nostrils, central palatal ridge, and high forehead. Bilateral congenital fat pads are present anteromedial to the heels. Fetal finger and toe pads are present and palmar and plantar grooves are deeper than normal with "pillowing" of the areas between the grooves. No patients with similar clinical findings have been located, but these two children have a remarkably similar clinical presentation which we consider a "new" syndrome. *Am. J. Med. Genet.* 75:18-21, 1998. © 1998 Wiley-Liss, Inc.

KEY WORDS: plantar lipomatosis; fetal finger and toe fat pads; plantar fat pads; mental retardation

INTRODUCTION

We have studied two boys with global developmental delay and similar facial appearance, plantar fat pads, fetal finger and toe pads, and increased internipple distance. No reports of similar individuals have been located. This appears to represent a new syndrome.

CLINICAL REPORTS

Patient I

A 9-year-old boy was first seen in Minneapolis at the age of 4 months. He was born at term after a normal pregnancy by cesarean section because of breech position to a 35-year-old mother and 58-year-old father. There is no parental consanguinity. He has no sibs, although his father has two normal sons and one normal daughter from his first marriage. There is no family history of any other birth defects or mental retardation.

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Developmental delay was evident at 6 months. He sat up at 9-10 months and walked at 4 years. He has no words at 9 1/2 years. A seizure disorder with grand mal and myoclonic seizures began at age 5 years and is controlled with Tegretol. An MRI scan of the brain at 5 years was normal. He has undergone repair of inguinal herniae and a right hydrocele and had two operations for subluxation of the hip at ages 7 and 8 years. A renal ultrasound at 9 years was normal. He is followed for intermittent exotropia and hyperopia. His chromosomes are normal (46,XY).

His present weight is 21.9 kg (<5th centile) and height is 113.7 cm (<5th centile). Head circumference is 49.5 cm (<5th centile). He has microcephaly, frontal bossing with high forehead, prominent chin, deep-set eyes, narrow palpebral fissures and mild bilateral ptosis (Fig. 1A,B). Marked midface hypoplasia has become more pronounced with time. Also evident are depressed nasal bridge, long upper lip with flat philtrum, everted lower lip and tongue fasciculations. He has curly blond hair. The palate has a central ridge. Teeth are extremely widely spaced and the few permanent erupted teeth show reduction in crown form such that the incisal edge is narrower than the cervix of the various anterior teeth (Fig. 2). The pinnae are large, posteriorly rotated and apparently low-set with fleshy lobes. There are pretragal pits anterior to both ears.

The neck is short and the chest is narrow with marked pectus excavatum. The internipple distance is increased and the areolae are reduced in diameter (Fig. 3). The heart, lungs, abdomen, and genitalia are normal. There is excess skin over the hands and feet, causing them to appear puffy. Fat pads are located on the medial border of both feet just anterior to the calcanei (Fig. 4). Fetal finger pads involving most digits and some fetal toe pads are seen (Fig. 5). Dermatoglyphic analysis revealed seven ulnar loops, one radial loop, and two whorls on the hands. There were digital and axial triradii. On the left hand is a single palmar transverse crease and clinodactyly of both fifth fingers. On the palms and soles, there are pillowed areas of skin separated by grooves with puffing out of the intergroove regions. The limbs are cool to touch and show livedo reticularis which has been present since birth. There are stiff elbows and knees and decreased range of motion at the hips. He is unable to pronate and/or

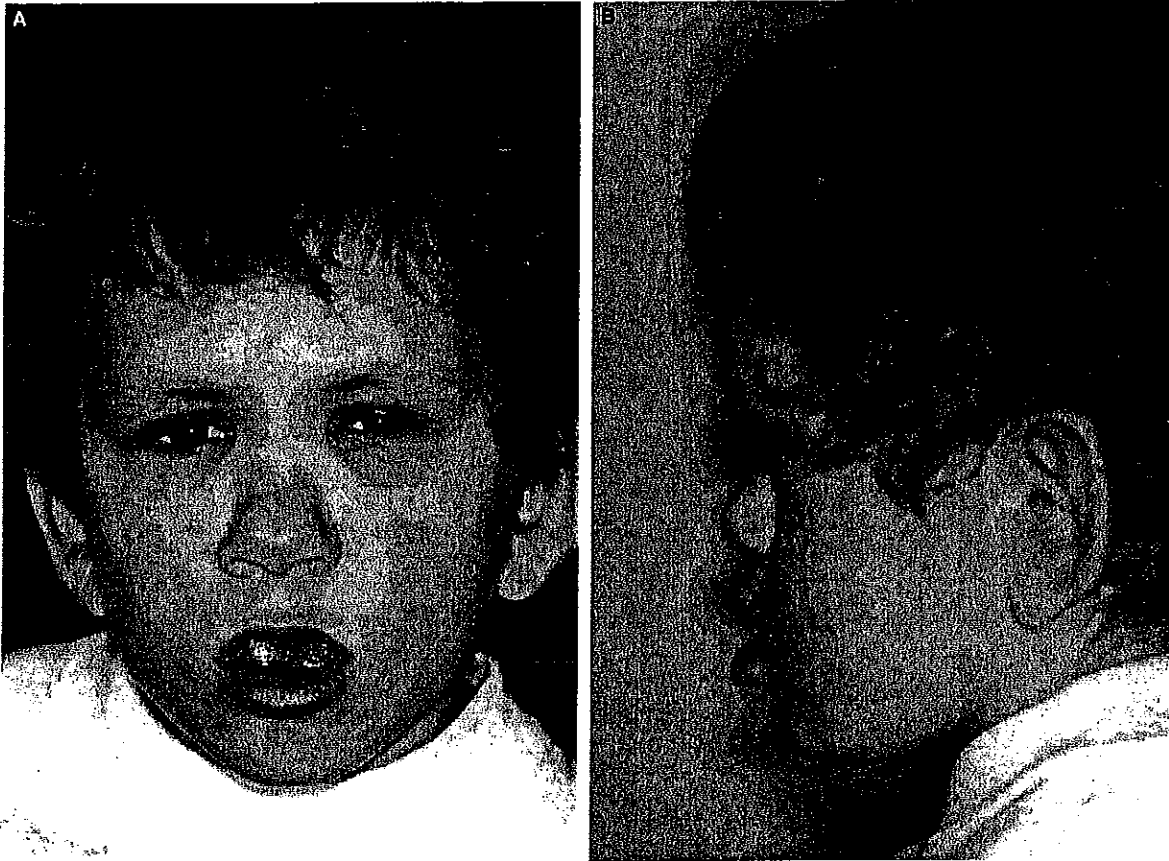


Fig. 1. A, B: Patient 1. Note midface hypoplasia, everted lower lip and large pinnae.

supinate his right forearm. There is significant thoracic scoliosis. Neurological exam shows hypertonia, hyperreflexia, and Babinski signs without clonus.

Patient 2

The patient is a 2 1/2-year-old boy born at 41 weeks of gestation in Northern Ireland to a 36-year-old

mother and a 37-year-old father. Birth weight was 3.62 kg. There are no other sibs and no parental consanguinity. There is no family history of birth defects or mental retardation.

He has global developmental delay. He has no speech and can walk only with significant assistance. Herniorrhaphy and repair of massive hydroceles was performed at age 20 months. His teeth were delayed in

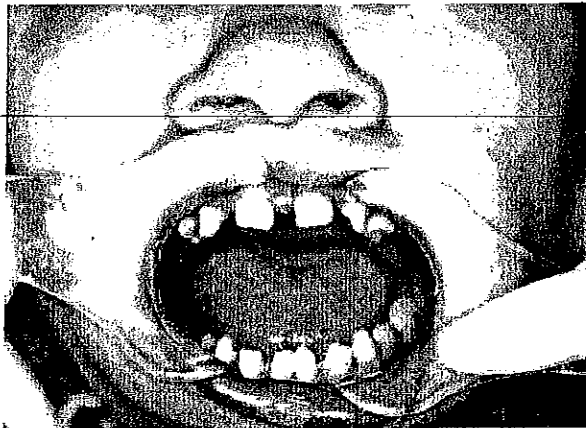


Fig. 2. Patient 1. Note widely spaced teeth, absent maxillary lateral incisors, and reduced width of incisal edges compared to the cervixes of lower incisors.

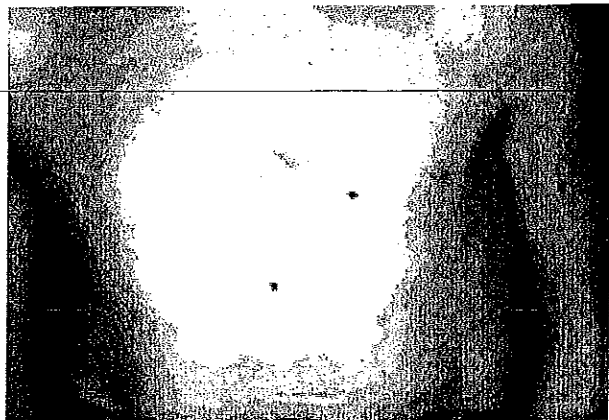


Fig. 3. Patient 1. Note pectus excavatum, widely spaced hypoplastic nipples and areolae, nevi.

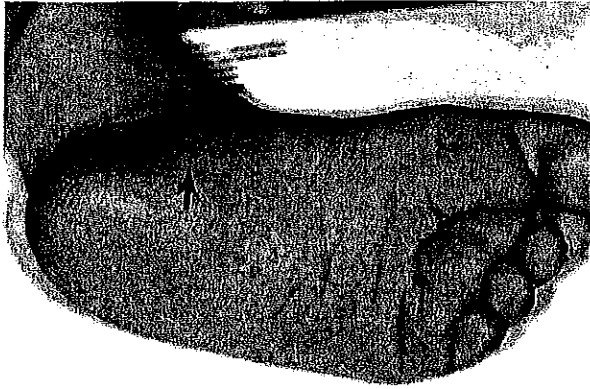


Fig. 4. Patient 1. Plantar surface showing grooves, pillowing of inter-groove soft tissues and fat pad anteromedial to heel (arrow).

eruption. He has not had any seizures. Vision and hearing appear normal. A renal ultrasound is normal.

At 30 months, head circumference is below the 3rd centile. Height is at the 10th centile and weight at the 50th centile. There is a high forehead and hypoplastic midface (Fig. 6). The nose has a "square" shape and the nostrils are anteverted. Upslanting palpebral fissures are present. There is a central palatal ridge. Bony swellings on the palatal surface of his gingiva are seen. The teeth are widely spaced. The pinnae are enlarged and appear low-set with fleshy lobes. There is an increased internipple distance.

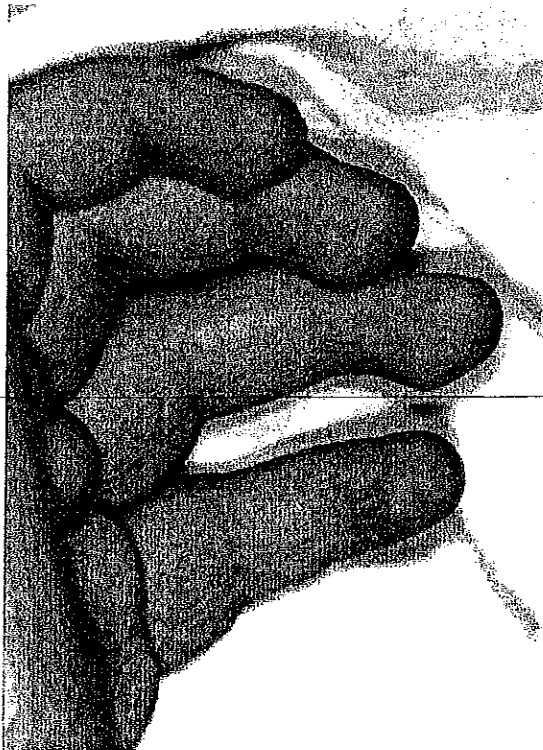


Fig. 5. Patient 1. Note fetal finger pads.



Fig. 6. Patient 2. Note high forehead, small square nose, upslanting palpebral fissures, and large pinnae.

Fetal finger and toe pads are evident (Fig. 7). Bilateral congenital plantar fat pads are present anteromedial to the heels (Fig. 8). The palmar grooves are deeper than normal with "pillowing" of the areas between the grooves. The toes are overlapping with the right second toe over the third and the left fourth toe over the third. The penis is small.

DISCUSSION

Congenital fatty heel pads are rare. They have been described as an isolated clinical finding [Livingstone and Burd, 1995]. Fatty heel pads may be painless or produce mild discomfort on walking. These pads can be bulky and should be distinguished from piezogenic pedal papules which represent small papular herniations of subcutaneous fat into the skin of the medial aspect of the heel [Schlappner et al., 1972; Shelley and Rawnsley, 1968].

Our patients have virtually identical large fatty pads anteromedial to the heels. In patient 1, these heel pads were first observed at age 4 months and they have remained the same in character up to the present age of 9 1/2 years. Other limb findings include "pillowing" between plantar and palmar grooves and fetal finger and toe fat pads. These may be related to a similar

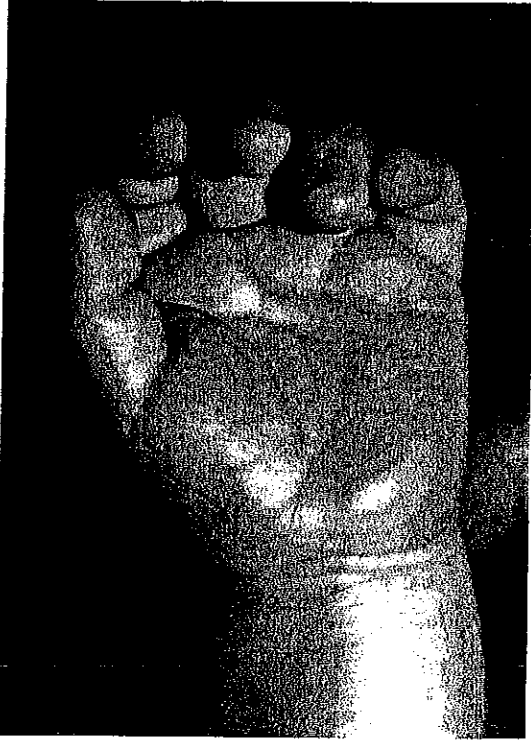


Fig. 7. Patient 2. Left hand showing fetal finger pads, deep grooves, and mild degree of pillowing.

process which produced the fatty heel pads. The facial appearance of our two patients is also very similar, with photographs of each at roughly the same age revealing their resemblance.

The natural history of this condition is evident in the

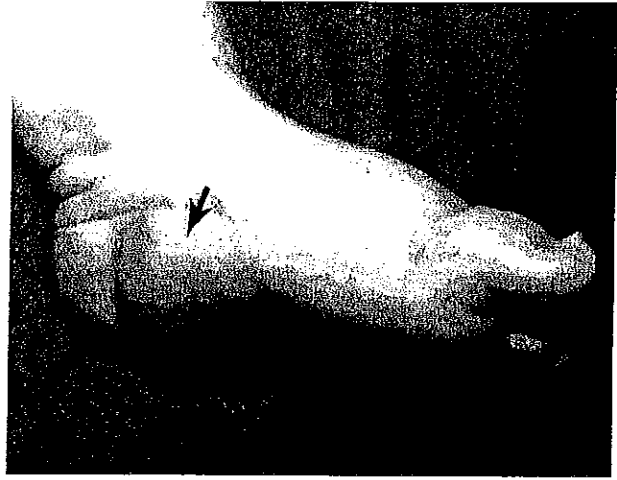


Fig. 8. Patient 2. Medial side of foot showing fat pad anteromedial to heel (arrow).

course of patient 1, who has no speech and severe mental retardation. He also has a seizure disorder, the onset of which occurred at an older age than the present age of patient 2. So far, the early course and findings in both patients are similar. The findings of these two patients likely represent a syndrome with recognizable physical traits and developmental course.

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