



PEDIATRIC IMAGES

Restrictive Dermopathy

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A girl neonate was admitted to a neonatal intensive care unit with tight skin all over the body, multiple joint contractures, and breathing difficulty. She was third born to



Figure 1 Neonate with Restrictive Dermopathy showing tight and denuded skin with multiple joint contractures.

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third degree consanguineous parentage and delivered at 33 weeks of gestation by caesarean section. The weight at birth was 1.02 kg, and the occipitofrontal circumference was 30.5 cm. She had a wide anterior fontanelle, small palpebral fissures, microretrognathia, tightening of skin, areas of denuded skin, and contracture of almost all joints of the extremities (Figure 1). Her mother previously had one stillbirth and another early neonatal death of unknown etiology. Her thyroid stimulating hormone and complete blood count were within the normal limits. In view of poor prognosis, her parents decided against continuing medical care and the baby was discharged. Due to the presence of the typical features of restrictive dermopathy, genetic confirmation was carried out at the Division of Nutrition and Metabolic Diseases, Center for Human Nutrition, Department of Internal Medicine, University of Texas Southwestern Medical Center, Dallas, TX, USA. The results revealed a homozygous c.1085insT (p.F361fs*380) mutation in *ZMPSTE24* gene, thereby confirming the diagnosis.

Conflicts of interest

The authors have no conflicts of interest relevant to this article.

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