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FREEMAN-SHELDON (WHISTLING FACE) SYNDROME WITH HYPERPYREXIA IN THE NEWBORN: CASE REPORT

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Summary: *Freeman-Sheldon (whistling face) syndrome with hyperpyrexia in the newborn: case report:* Freeman-Sheldon syndrome (FSS) is a rare, multiple congenital contracture syndrome that is relatively well-known, since affected children have a striking appearance. This entity was historically referred to as the "whistling-face syndrome". Malignant hyperthermia and hyperpyrexia have been documented in FSS after general anesthesia related to the neuropathy. We report a male neonate with FSS and hyperpyrexia without anesthesia. To our knowledge, our patient is the first in the literature with hyperpyrexia in the newborn period without anesthesia.

Key-words: Freeman-Sheldon Syndrome – Whistling-Face Syndrome – Cranio-carpo-tarsal dysplasia – Distal Arthrogryposis Type 2A – hyperpyrexia.

INTRODUCTION

Freeman-Sheldon Syndrome (FSS) (Whistling-Face, or Cranio-carpo-tarsal dysplasia, or Distal Arthrogryposis Type 2A) is a rare syndrome that was first described in 1938 by Freeman and Sheldon (OMIM 277720). It is characterized by an atypical facial appearance (whistling face) and anomalies of the hand and foot. Generalized myopathy has been considered to be responsible for the facial and skeletal anomalies. Most cases of FSS are sporadic, although there has been evidence of autosomal-recessive, autosomal-dominant and X-linked-recessive transmission in FSS cases (1, 3, 7). Severe feeding difficulties, chronic respiratory tract infections and related chronic lung diseases, and the frequent need for surgical procedures for the deformities in these patients have a negative impact on the prognosis. Malignant hyperthermia and hyperpyrexia after general anesthesia have been documented in FSS related to neuropathy (6, 7).

CASE REPORT

A 22-days-old male patient presented to our clinic due to an abnormal facial appearance, hand deformities and feeding difficulties. He was born from the second pregnancy with cesarean section; the

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