

CASE 1991-5

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CLINICAL ABSTRACT:

A small gestational age neonatal male was born at 37 weeks gestation to a 17 year old Gravida I, Para I mother by C-section. The APGARs were 3 and 4 and prenatal care was limited. At delivery the child was severely hypotonic. He was transferred to our institution following resuscitation in the delivery room. There was no cry or suck reflex and the infant required ventilator support because of absence of spontaneous respirations. Evaluation included urinary amino acids which were essentially normal. Chromosomal studies revealed a normal male karyotype. Echocardiogram demonstrated a large atrial septal defect, a moderate muscular ventricular septal defect, biventricular enlargement and a small patent ductus arteriosus. A muscle biopsy was performed at ten days. The child subsequently died at the age of 27 days and an autopsy was performed.

MATERIAL SUBMITTED: 1. H & E stained slide of muscle biopsy.
2. H & E stained section of spinal cord (could be from different levels)

POINTS FOR DISCUSSION: 1. What is your diagnosis on the muscle biopsy?
2. Is there more than one diagnosis for this type of a picture in the muscle biopsy?