

61st ANNUAL DIAGNOSTIC SLIDE SESSION 2020.

CASE 2020 #7

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Clinical History:

Our patient was a three month old male born at 35 weeks with hypotonia, hypothermia, and respiratory distress subsequently presenting at three months of age with exacerbated respiratory distress and worsened hypotonia. Physical exam revealed a lethargic infant with global hypotonia, leftward gaze deviation, symmetric reflexes, tachypnea, left undescended testis, and hypospadias. Laboratory testing showed lactic acidosis. Imaging revealed abnormal diffusion restriction throughout the white matter tracts of the brain and spine. The patient's respiratory status declined rapidly resulting in death at three months of age.

Autopsy findings:

- 1) Widespread leukoencephalopathy with spongiform changes
- 2) Microvesicular steatosis of liver

Material submitted: Diagnostic magnetic resonance imaging

Points for discussion:

1. Diagnosis
2. Pathogenesis