TEXAS Health and Human Services | Texas Department of State Health Services

Newborn Screening ACT Sheet

Elevated C10:2

2, 4-Dienoyl-Coenzyme A Reductase Deficiency (DE RED)

Differential Diagnosis

2,4-Dienoyl-Coenzyme A Reductase Deficiency (DE RED).

Condition Description

The enzyme 2, 4 Dienoyl-CoA reductase helps break down unsaturated fatty acids. In 2, 4 Dienoyl-Co-A reductase deficiency (DE RED), the body may either not make enough or make non-working 2,4 dienoyl-CoA reductase. When this happens, the body cannot fully break down unsaturated fatty acids. This causes harmful substances to build up in the body. DE RED can cause weak muscle tone and can lead to the presence of too much acid in the blood. The effectiveness of treatment is currently unknown.

Medical Emergency: Take the Following IMMEDIATE Actions

- **Contact family to inform them of the newborn screening result and ascertain clinical status (poor feeding, vomiting, lethargy);**
- **Consult with pediatric metabolic specialist (See attached list);**
- **Evaluate the newborn (poor feeding, vomiting, hypotonia, irritability.);**
- **If signs are present or infant is ill, initiate emergency treatment with IV glucose. Transport to hospital for further treatment in consultation with metabolic specialist;**
- **If infant is asymptomatic initiate timely confirmatory/diagnostic testing, as recommended by specialist;**
- **Initial testing: plasma acylcarnitine profile, plasma quantitative amino acids, plasma carnitine levels, urine acylglycines, and urine organic acids;**
- **Repeat newborn screen if the second screen has not been done;**
- **Educate family about need for infant to avoid fasting. Even if mildly ill, immediate treatment with IV glucose is needed; and**
- **Report findings to newborn screening program.**

Diagnostic Evaluation

In a reported case of DE RED, high levels of lysine in the blood and the presence of 2-trans, 4-cis-decadienoylcarnitine in the blood and urine were associated with DE RED. Follow-up testing will probably also include skin biopsy for fibroblast studies or DNA testing.

Clinical Considerations

VLCAD Coordinate care closely with a metabolic geneticist.

Additional Information

Baby's First Test – 2,4 Dienoyl-CoA Reductase Deficiency

Revised: 5/23/2024