

Symptoms

-Lack of head control

- -Central hypotonia
- -Peripheral hypertonia
- -Intellectual disability
- -Inability to gain and maintain weight
- -Increased heart rate
- -Motor delay
- -Low bone density

Diagnosis

MCT8 Deficiency may be diagnosed by a pediatric neurologist, a pediatric endocrinologist or a geneticist.

Detection

The earliest abnormalities include floppy body and the inability to support the head. Typical thyroid tests may indicate the syndrome (high serum T3 and borderline low T4) Confirmation requires genetic testing.

