

# Niedobór UPS18

**Kod Orpha: 481665 Kod OMIM: 617397**

## Opis choroby \*

### Definicja

A rare genetic neurological disorder characterized by severe pseudo-TORCH syndrome with signs of brain damage and occasionally systemic manifestations resembling the sequelae of congenital infection, but in the absence of an infectious agent. Characteristic features include microcephaly, white matter disease, cerebral atrophy, cerebral hemorrhage, and calcifications, among others. Affected individuals typically have seizures and respiratory insufficiency and die in infancy.

### Dane

### Klasyfikacja

Choroba

### Kod ORPHA

481665

### Kod OMIM

617397

### Kod ICD10

Q04.8

### Kod ICD11

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### [\\*Źródło](#)

orphanet

## Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.