## Opis choroby *

Definicja

A rare genetic disease characterized by pre- and postnatal growth restriction, developmental delay, adrenal hypoplasia, genital abnormalities (such as microphallus, hypospadias, or cryptorchidism), thrombocytopenia and/or anemia, recurrent severe invasive infections, and enteropathy with chronic diarrhea. Myelodysplastic syndrome and dysmorphic features (including downslanting palpebral fissures, low-set and posteriorly rotated ears, anteverted nares, camptodactyly, and arachnodactyly, among others) may also be observed.

Dane

**Klasyfikacja**
Choroba

Synonimy
Myelodysplasia-infection-restriction of growth-adrenal hypoplasia-genital anomalies-enteropathy syndrome
Zespół mielodysplazji, infekcji, zahamowania wzrastania, hipoplazji nadnerczy, wad narządów płciowych zewnętrznych i enteropatii
Myelodysplasia-infection-restriction of growth-adrenal hypoplasia-genital phenotypes-enteropathy syndrome

**Kod ORPHA**
494433

**Kod OMIM**
617053

**Kod ICD10**
D46.7

**Kod ICD11**
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*Źródło

orphanet