## Opis choroby \*

## Definicja

A rare disorder of ornithine metabolism characterized by global developmental delay, alopecia, macrocephaly, and dysmorphic facial features (including high and broad forehead, hypertelorism, ptosis, blepharophimosis, downslanting palpebral fissures, deep-set eyes, large ears, and retrognathia or high arched palate). Additional reported manifestations are sensorineural hearing loss, spasticity, hypotonia, hypoplastic nails, cryptorchidism, and clinodactyly, among others. Brain imaging may show white matter abnormalities, periventricular cysts, enlarged lateral ventricles, or prominent perivascular spaces.

Dane

Klasyfikacja Synonimy

Choroba Bachmann-Bupp syndrome

Zespół Bachmanna i Buppa

Ornithine decarboxylase deficiency Ornithine decarboxylase deficiency

 Kod ORPHA
 Kod OMIM
 Kod ICD10

 544488
 619075
 E72.4

**Kod ICD11** 

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## \*Źródło

orphanet