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
Choroba

Synonimy
Bachmann-Bupp syndrome
Zespół Bachmanna i Buppa
Ornithine decarboxylase deficiency
Ornithine decarboxylase deficiency

Kod ORPHA
544488

Kod OMIM
619075

Kod ICD10
E72.4

Kod ICD11
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*Źródło

orphanet