

Opis choroby *

Definicja

17-beta-hydroxysteroid dehydrogenase isozyme 3 (17betaHSD III) deficiency is a rare disorder leading to male pseudohermaphroditism (MPH), a condition characterized by incomplete differentiation of the male genitalia in 46X,Y males.

Dane

Klasyfikacja

Choroba

Synonimy

17-beta-hydroxysteroid dehydrogenase 3 deficiency
Niedobór 17-ketoreduktazy
Niedobór 17-ketosteroidoreduktazy
Niedobór dehydrogenazy 17-beta-hydroksysteroidowej typu 3
Pseudohermafrodytyzm męski spowodowany niedoborem dehydrogenazy 17-beta-hydroksysteroidowej typu 3
17-ketoreductase deficiency
17-ketosteroidreductase deficiency
46,XY disorder of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency

Kod ORPHA

752

Kod OMIM

264300

Kod ICD10

E29.1

Kod ICD11

LD2A.3

*Źródło

orphanet