

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	Synonimy
Choroba	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	Kod OMIM	Kod ICD10
752	264300	E29.1

Kod ICD11
LD2A.3

*Źródło

orphanet