Opis choroby *

Definicja

A teratogenic embryofetopathy that results from maternal exposition to methimazole (MMI; or the parent compound carbimazole) in the first trimester of pregnancy. MMI is an antithyroid thionamide drug used for the treatment of Graves' disease. In the infant, MMI may result in choanal atresia, esophageal atresia, omphalocele, omphalomesenteric duct anomalies, congenital heart disease (such as ventricular septal defect), renal system malformations and aplasia cutis. Additional features that may be observed include facial dysmorphism (short upslanting palpebral fissures, a broad nasal bridge with a small nose and a broad forehead) and athelia/hypothelia.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
MMI/CMZ embryofetopathy
Embriopatia metimazolowa/karbamizolowa
Embriofetopatia metimazolowa/karbamizolowa
MMI/CMZ embryopathy
Methimazole/carbimazole embryofetopathy
Methimazole/carbimazole embryopathy

Kod ORPHA
1923

Kod OMIM
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Kod ICD10
Q86.8

Kod ICD11
LD2F.0Y

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*Źródło

orphanet