

## **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	Synonimy
Zespół wad wrodzonych	MMI/CMZ embryofetopathy
	Embriopatia metimazolowa/karbamizolowa
	Embrio-fetopatia metimazolowa/karbamizolowa
	MMI/CMZ embryopathy
	Methimazole/carbimazole embryofetopathy
	Methimazole/carbimazole embryopathy

**Kod ORPHA** 1923      **Kod OMIM** -      **Kod ICD10** Q86.8

Kod ICD11  
LD2E.0Y

\* Źródło

orphanet