Belgian Genetic Tests database

DISEASE:
Rett syndrome

NAME:	Rett syndrome
DESCRIPTION:	A rare severe, X-linked, neurodevelopmental disorder characterized by rapid developmental regression in infancy, partial or complete loss of purposeful hand movements, loss of speech, gait abnormalities, and stereotypic hand movements, commonly associated with deceleration of head growth, severe intellectual disability, seizures, and breathing abnormalities. The disorder has a progressive clinical course and may associate various comorbidities including gastrointestinal diseases, scoliosis, and behavioral disorders.
ORPHACODE:	778
XREF(S):	Orphanet MeSH MedDRA OMIM ICD-10
ANALYTE(S):	MECP2
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RELATED CONTENT

Related Genetic Tests

- Epilepsy (gene panel)
- Rett syndrome
- Rett syndrome / MECP2 Duplication Syndrome

Related Laboratories

- Centre de Génétique Humaine - CHU Sart-Tilman
- Centrum Medische Genetica - UZ Antwerpen
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- methyl-CpG binding protein 2

Related Gene Panels

- Rare epilepsy with developmental delay (> 240 genes) - UZA