DISEASE:
Dihydropyrimidine dehydrogenase deficiency

NAME:	Dihydropyrimidine dehydrogenase deficiency
DESCRIPTION:	A rare disorder of pyrimidine metabolism characterized by a variable phenotype ranging from absence of symptoms to severe neurological involvement with developmental delay, intellectual disability, and seizures. Additional signs and symptoms may include hypotonia, microcephaly, ocular abnormalities (such as microphthalmia, nystagmus, and strabismus), and autistic behavior, among others. Analysis of urine typically shows high levels of uracil and thymine. Patients are at risk of suffering from severe toxicity after the administration of the anti-neoplastic agent 5-fluorouracil.
ORPHACODE:	1675
SYNONYMS:	Familial pyrimidinemia
XREF(S):	Orphanet ICD-10 OMIM MeSH MedDRA
ANALYTE(S):	DPYD
CREATED:	13 May 2019 - 01:02
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RELATED CONTENT

Related Genetic Tests

- Dihydropyrimidine dehydrogenase deficiency (5-fluorouracil (5-FU) toxicity) - Pharmacogenetics
- Dihydropyrimidine dehydrogenase deficiency/5-fluorouracil toxicity - Pharmacogenetics (4 variants: DPYD*2A, DPYD*13, c.2846A>T, HapB3)
- Dihydropyrimidine dehydrogenase deficiency; 5-fluorouracil toxicity - pharmacogenetics (4 variants: DPYD*2A, DPYD*13, c.2846A>T, HapB3) - Pharmacogenetics

Related Laboratories

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

Related Analytes

- dihydropyrimidine dehydrogenase