DISEASE:
Fumaric aciduria

NAME:	Fumaric aciduria
DESCRIPTION:	Fumaric aciduria (FA), an autosomal recessive metabolic disorder, is most often characterized by early onset but non-specific clinical signs: hypotonia, severe psychomotor impairment, convulsions, respiratory distress, feeding difficulties and frequent cerebral malformations, along with a distinctive facies. Some patients present with only moderate intellectual impairment.
ORPHACODE:	24
SYNONYMS:	Fumarase deficiency
XREF(S):	Orphanet MeSH ICD-10 OMIM
ANALYTE(S):	FH
CREATED:	13 May 2019 - 01:02
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RELATED CONTENT

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- fumarate hydratase