DISEASE:
Autosomal recessive primary microcephaly

NAME:	Autosomal recessive primary microcephaly
DESCRIPTION:	Autosomal recessive primary microcephaly (MCPH) is a rare genetically heterogeneous disorder of neurogenic brain development characterized by reduced head circumference at birth with no gross anomalies of brain architecture and variable degrees of intellectual impairment.
ORPHACODE:	2512
SYNONYMS:	MCPH Microcephalia vera Microcephaly vera True microcephaly

XREF(S):	Orphanet OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM ICD-10 OMIM

ANALYTE(S):	WARS1 SARS1 CIT NUP37 NCAPD3 CENPE COPB2 TRAPPC10 MCM7 METTL5 TRAPPC14 KIF14 TAF13 PYCR2 CDK5RAP2 CENPJ ASPM MCPH1 STIL WDR62 CEP152 CEP63 CEP135 KNL1 PHC1 CDK6 SASS6 MFSD2A ANKLE2
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Brain malformations (gene panel)
- Short stature/ Growth retardation/ (gene panel)

Related Laboratories

- Centre de Génétique Humaine - Erasme ULB
- Centrum Medische Genetica - UZ Antwerpen

Related Analytes

- ankyrin repeat and LEM domain containing 2
- assembly factor for spindle microtubules
- CDK5 regulatory subunit associated protein 2
- cyclin dependent kinase 6
- centromere protein E
- centromere protein J
- centrosomal protein 135
- centrosomal protein 152
- centrosomal protein 63
- citron rho-interacting serine/threonine kinase
- COPI coat complex subunit beta 2
- kinesin family member 14
- kinetochore scaffold 1

- minichromosome maintenance complex component 7
- microcephalin 1
- methyltransferase 5, N6-adenosine
- MFSD2 lysolipid transporter A, lysophospholipid
- non-SMC condensin II complex subunit D3
- nucleoporin 37
- polyhomeotic homolog 1
- pyrroline-5-carboxylate reductase 2
- seryl-tRNA synthetase 1
- SAS-6 centriolar assembly protein
- STIL centriolar assembly protein
- TATA-box binding protein associated factor 13
- trafficking protein particle complex subunit 10
- trafficking protein particle complex subunit 14
- tryptophanyl-tRNA synthetase 1
- WD repeat domain 62

Related Gene Panels

- Brain malformations (34 genes) - ULB
- Growth retardation/short stature (genepanel) - UZA
