DISEASE:
Pfeiffer syndrome type 1

NAME:	Pfeiffer syndrome type 1
DESCRIPTION:	Pfeiffer syndrome type 1 (PS1) is a mild to moderately severe type of Pfeiffer syndrome (PS; see this term), characterized by bicoronal craniosynostosis, variable finger and toe malformations, and in most cases, normal intellectual development.
ORPHACODE:	93258
SYNONYMS:	Classic Pfeiffer syndrome
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	FGFR1 FGFR2
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Brain malformations (gene panel)
- Craniosynostosis (gene panel)
- Craniosynostosis syndrome (hot spot mutation - p.Pro252Arg)

Related Laboratories

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

Related Analytes

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- fibroblast growth factor receptor 2

Related Gene Panels

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