

Svenska barncancerregistret – VPH

Diagnosis – formulär

Version 210921

Report date:
2 0
Registered by:
Reporting clinical:
Forename:
Surname:
Date of Birth:
2 0 -
Gender:
☐ Female ☐ Male
City:
Country:

Diagnosis
Method of diagnosis (Several options)
☐ Clinical symptoms
☐ Phenotype Biological laboratory test
☐ Genotype Genetic testing
If Genotype Genetic testing, please fill in Genetic Diagnosis date and Gene:
Genetic Diagnosis date
2 0
Gene: _____
Date of diagnosis
2 0
ORPHA-Code: _____
ICD-10-code: _____
Genetic Diagnosis date
2 0
Undiagnosed case
☐ No ☐ Yes
Age at onset (One option)
☐ Antenatal
☐ At birth
☐ Undermined
☐ Date
If above option is date, please fill in date below:
Date
2 0

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Reason for diagnosis (One option)

- ☐ Antenatal diagnosis / screening
- ☐ Neonatal screening
- ☐ By the presence of affected related
- ☐ Clinical manifestations
- ☐ Abnormal laboratory values (without clinical manifestation)
- ☐ Other

If above option is Other, please specify below:

First symptoms (Several options)

- ☐ Acute anemia
- ☐ Acute Chest Syndrome
- ☐ Acute hemolytic crisis
- ☐ Acute infection
- ☐ Failure to thrive
- ☐ Fatigue
- ☐ Hand-Foot Syndrome
- ☐ Hemolytic anemia
- ☐ Hydrops fetalis
- ☐ Hypo or non-regenerative anemia
- ☐ Intra-uterine growth retardation
- ☐ Intra-uterine malformation
- ☐ Neonatal hemolytic crisis
- ☐ Neonatal jaundice
- ☐ Jaundice
- ☐ Splenic Sequestration
- ☐ Splenomegaly/splenic pain
- ☐ Vaso-occlusive crisis
- ☐ Other, Specify free text

If above option is Other, please specify below:

Diagnosis methods

PK activity assay (One option)

- ☐ Never performed
- ☐ At diagnosis
- ☐ After splenectomy
- ☐ After BMT

If PK activity assay are At diagnosis, after splenectomy or after BMT please fill in below

IU/L

☐ Yes ☐ No ☐ Data missing

If yes, please fill in IU/L number:

Reference range

☐ Yes ☐ No ☐ Data missing

If yes, please fill in IU/L number:

Date

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Other RBC enzyme activity measured at time of PK assay (One option)

- ☐ g6pd
- ☐ HK
- ☐ Not performed
- ☐ Unknown
- ☐ Other

If Other RBC enzyme activity measured at time of PK assay are g6pd or HK please fill in below

IU/L

☐ Yes ☐ No ☐ Data missing

If yes, please fill in IU/L number:

Reference range

☐ Yes ☐ No ☐ Data missing

If yes, please fill in IU/L number:

Date

| 2 | 0 | | | | | | |

If Other RBC enzyme activity measured at time of PK assay is Other, please fill in below:

Bone marrow aspiration (One option)

- ☐ Never performed
- ☐ Normal bone marrow
- ☐ Normal marrow cellularity with a paucity of erythroid precursors (DBA)
- ☐ Other bone marrow findings - specify

If Bone marrow aspiration is Other bone marrow findings, please specify below:

Chromosomal Breakage analysis

- ☐ Never performed
- ☐ Increased chromosomal breakage
- ☐ Normal findings
- ☐ Other

If above answer is other, please specify:

eADA

- ☐ Never performed
- ☐ Elevated eADA activity
- ☐ Normal value
- ☐ Other

If above answer is other, please specify:

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HbF

- ☐ Never performed
- ☐ Elevated value for age
- ☐ Normal value for age

EMA binding test / Band 3 protein expression

- ☐ Never performed
- ☐ Decreased EMA binding
- ☐ Normal value
- ☐ Increased EMA binding

OFT

- ☐ Never performed
- ☐ Increased Osmotic fragility
- ☐ Normal value
- ☐ Decreased Osmotic fragility

GLT/AGLT

- ☐ Never performed
- ☐ Increased hemolysis
- ☐ Normal value

Osmoscan/LORRCA

- ☐ Never performed
- ☐ Spherocytosis profile
- ☐ Stomatocytosis/Xerocytosis profile
- ☐ Other profile

If above answer is other profile, please specify:

SDS-PAGE

- ☐ Never performed
- ☐ Decreased EMA binding
- ☐ Normal value
- ☐ Increased EMA binding