

WG3

Testing for a congenital erythrocytosis

Consider screening those with

- True erythrocytosis
- No polycythaemia vera
- No identifiable secondary cause
- Young patients
- If positive family history

Low erythropoietin (EPO) level ----- consider lesions of EPO signalling pathway

Normal or raised EPO level ----- consider gene mutations in the oxygen sensing pathway

Sample

20 mls peripheral blood in EDTA

Where to send a sample

Belgium

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Genes screened

EpoR	Exons 7 and 8
VHL	All exons
PHD2	All exons
HIF2	Exon 12

Czech Republic

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Genes screened

EpoR	Exon 7 and exon 8
VHL	All exons
PHD2	All exons
HIF2	Exon 9 and 12
Others HBB, HBA1 and HBA2	All exons

Name: Drs Jiří Schwarz, Jana Marková

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Genes screened

EpoR	Exon 7 and exon 8
VHL	Exons 1-3
PHD2	Exons 2-4
HIF2	12

FRANCE

Name: Dr Anne-Paule Gimenez-Roqueplo

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Genes screened

EpoR	no
VHL	All exons
PHD2	All exons

HIF2	Exon 12

Name: Dr Brigitte Bressac de Paillerets

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Genes screened

EpoR	no
VHL	All exons
PHD2	No
HIF2	No

Name: Dr Valerie Ugo

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Genes screened

EpoR	Exons 7-8
VHL	No
PHD2	No

HIF2	No

VHL	Exons 1, 2 and 3
PHD2	P317, R371 and flanking regions
HIF2	M535, G537 and flanking regions

GERMANY

Name: Dr. Susanne Schnittger
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Genes screened

EpoR	Exon 8
VHL	Whole coding region (cDNA)
PHD2	Codons P317 and R371
HIF2	Exon 12

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Genes screened

EpoR	Exons 7-8
VHL	All exons
PHD2	All exons
HIF2	Exon 9-16

ITALY

Name: Dr. Silverio Perrotta

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Genes screened:

EpoR	Exons 7 and 8
VHL	Exons 1-3
PHD2	Exons 1-4
HIF2	Exons 12
Others	PHD1 (exons 1-5) PHD3 (exons 1-3) HIF1 (exons 9, 12, 150)
HIF1/CULLINS/PHD1/PHD3	

PORTUGAL

Name: Dr. Celeste Bento

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Genes screened

EpoR	Exon 7 and exon 8
VHL	Exons 1, 2, 3
PHD2	Exon 1-5
HIF2	Exon 12
Others HBB, HBA1 and HBA2, DPGM	All the exons

SPAIN

Name: Dr Beatriz Bellosillo

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Genes screened

EpoR	Exon 8
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SWITZERLAND

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Gene screened

EpoR	Exons 7+8
VHL	Exons 1-3
PHD2	No
HIF2	No

UNITED KINGDOM

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Genes screened

EpoR	Exons 7 and 8
VHL	Exons 1, 2 and 3
PHD2	Exons 1, 2, 3 and 4

HIF2	Exon 12