Reporting newborn sickle cell screening results

[Note: 'Thalassaemia' = β^0, β^+, δβ, γδβ and Hb Lepore as appropriate]

Analytical results (Hb written in order of %)	Diagnostic possibilities	Report format	Status code for child health reporting
Sickle cell disease			
FS (Fetal and sickle haemoglobin)	Sickle cell anaemia (81%) Sickle cell - β thalassaemia (17%) Sickle cell - HPFH (2%) The figures for the birth prevalence of sickle cell disease were obtained from 146 children identified by newborn screening in London and confirmed by parental results.	Results consistent with sickle cell disease Clinical referral required Actual genotype will require further investigation	08
FSA (Fetal haemoglobin, sickle haemoglobin and a small amount of haemoglobin A)	Sickle cell - β thalassaemia Transfusion Sickle cell carrier	Results consistent with sickle cell disease Result valid only if not transfused Clinical referral required Actual genotype will require further investigation	08
FSC (Fetal haemoglobin, sickle haemoglobin and haemoglobin C)	Haemoglobin SC disease	Results consistent with Hb SC disease Clinical referral required	08
FSD (with the characteristics of D^{Punjab}) Fetal haemoglobin, sickle haemoglobin and haemoglobin D	Haemoglobin SD disease	Results consistent with Hb SD disease Clinical referral required	08
FSE (Fetal haemoglobin, sickle haemoglobin and haemoglobin E)	Haemoglobin SE disease	Results consistent with Hb SE disease Clinical referral required	08
FSO^{Arab} (Fetal haemoglobin, sickle haemoglobin and haemoglobin O^{Arab})	Haemoglobin SO^{Arab} disease	Results consistent with Hb SO^{Arab} disease Clinical referral required	08
FSV (Fetal haemoglobin, sickle haemoglobin and unidentified haemoglobin variant)	Clinical assessment required, unless “V” can be identified and is known to be clinically benign in combination with Hb S.	Possible Sickle Cell Disease Referral required for clinical assessment to confirm diagnosis unless “V” has been confirmed and is known to be clinically benign in combination with Hb S.	08

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Other potentially clinically significant conditions			
F only (Fetal haemoglobin [or with haemoglobin A outside designated action values])	Possible β thalassaemia major Prematurity Homozygous HPFH HPFH with β thalassaemia	Only fetal haemoglobin detected Clinical referral required	07
FE (Fetal haemoglobin and haemoglobin E [with haemoglobin A outside designated action values])	Haemoglobin E/β thalassaemia; a form of haemolytic anaemia which may cause transfusion dependence Haemoglobin E disease; a mild form of haemolytic anaemia Haemoglobin E with HPFH	Possible homozygous HbE or Hb E/β thalassaemia Clinical referral required Valid if not transfused	07
FEA (Fetal haemoglobin, haemoglobin E and haemoglobin A)	Compound heterozygote for HbE and β+ thalassaemia; which may cause transfusion dependence	Possible compound heterozygous HbE and β+ thalassaemia. Clinical referral required	07

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Analytical results (Hb written in order of %)	Diagnostic possibilities	Report format	Status code for child health reporting
Benign conditions and carriers			
FC or FD or FO^{Arab} (Fetal haemoglobin and haemoglobin C or haemoglobin D or haemoglobin O^{Arab})	Homozygous HbC or D or O^{Arab} Compound heterozygote for HbC or D or O^{Arab} and β thalassaemia or HPFH	Possible homozygous HbC or D or O^{Arab} Possible compound heterozygous HbC or D or O^{Arab} and β thalassaemia or HPFH	07
FCA or FDA or $FO^{Arab}A$ (Fetal haemoglobin and haemoglobin C or D or O^{Arab} and haemoglobin A)	Compound heterozygote for HbC or D or O^{Arab} and β^+ thalassaemia	Possible compound heterozygous HbC or D or O^{Arab} and β^+ thalassaemia Valid if not transfused	07

FAS (Fetal haemoglobin, haemoglobin A and haemoglobin S)	Sickle cell carrier; usually clinically benign but genetically significant	Results consistent with sickle cell carrier Valid if not transfused	05
FAC or FAD or FAE or FAO^{Arab} (Fetal haemoglobin, haemoglobin A and haemoglobin C or D or E or O^{Arab})	HbC carrier or HbD carrier or HbE carrier or HbO^{Arab} carrier; clinically benign but genetically significant	Results consistent with HbC carrier or HbD carrier or HbE carrier or HbO^{Arab} carrier Valid if not transfused	0601 FAC 0602 FAD 0603 FAE 0604 FAO^{Arab}
FV and FVA (Fetal haemoglobin, with or without haemoglobin A and a haemoglobin variant [not S, C, D, E or O^{Arab}])	Clinical assessment required, unless confirmed that there is no coexisting beta thalassaemia disease. If confirmed beta chain variant, then referral is not required, unless known to be clinically significant. Note that alpha and/or gamma chain variants may mask beta thalassaemia disease in this scenario.	Beta thalassaemia disease cannot be excluded. Referral required for clinical assessment to confirm diagnosis unless beta thalassaemia disease has been excluded.	07
FAV (Fetal haemoglobin, with haemoglobin A and a haemoglobin variant [not S, C, D, E or O^{Arab}])	Most likely clinically insignificant haemoglobin but may be genetically significant.	Hbs S, C, D, E and O^{Arab} not detected. Note – Carriers of beta thalassaemia and Hb Lepore cannot be excluded at this age. Valid if not transfused	04
FA (Fetal and adult haemoglobin)	No haemoglobin variant detected	Hbs S, C, D, E and O^{Arab} not detected. Note – Carriers of beta thalassaemia and Hb Lepore cannot be excluded at this age. Valid if not transfused	04