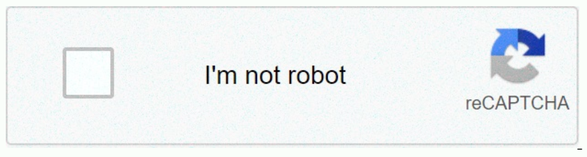

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Antenatal thalassaemia screening guidelines

Author:The Hong Kong College of Obstetricians and Gynaecologists, Hong Kong College Foundation Academy of Medicine Region:International Topics: Genetic testing and screening Document type:Year Guidelines:October 2003 URL: SummarySince July 2000, antenatal thalassaemia screening has been offered to all pregnant women in every public hospital and Maternal and Child Health Center in Hong Kong. The document provides guidelines for screening Hong Kong's local population (mostly southern Chinese ethnicities) for talasemia. A basic technical introduction of molecular and the prevalence of talasemia is provided, followed by a discussion of screening times and the educational and counseling needs of patients to obtain proper approval. Contact details We are pleased to announce the publication of an updated sickle cell and thalassaemia (SCT) examination handbook GOV.UK. Thank you to everyone who took part in a consultation earlier this year on a handbook that provides guidance and support to health professionals involved in SCT screening pathways. Your valuable feedback has helped ensure the handbook meets the needs of our users. Documents in HTML format are easily accessible. You can simply navigate chapters online although you can still print individual chapters and attachments if you need to. The SCT program screens pregnant women to see if they are at risk of having a baby with sickle cell disease or majoring in thalassaemia. It also screens babies for sickle cell disease as part of a newborn blood point screening program. Legacy diagrams of haemoglobin disorders included in the Haemoglobinopathies Understanding chapter of the program handbook Various updated guides and resources Handbook includes information about: antenatal screening pathways with specific guidelines on how to care for 'at-risk' couples known for antenatal screening processes and how to follow up on family origin questionnaire results and important issues for settlement in special high and low circumstances to look out for including adoption, blood transfusions and paternal screening of bone marrow transplants and tips for improving participation and referral counseling for prenatal diagnosis testing in subsequent pregnancy screening pathways of newborns, what results are expected and how to deal with those sickle cell diseases, major thalassaemia and other haemoglobinopathy disorders and carrier countries failsafe patient support organizations, quality assurance and data collection resources and training courses to support screening programs If you want a copy of the consultation report , please contact the filtering helpdesk. Phe Phe Screening blog provides the latest news from all NHS screening programmes. You can sign up to receive updates directly to your inbox, so there's no need to keep checking new blogs. Antenatal screening identifies women with haemoglobinopathy and provides approved biological father screening. When both parents are carriers of significant haemoglobinopathy, there is 1 in 4 (25%) chance, in each pregnancy, that their baby can inherit a condition that requires treatment. The most important conditions are sickle cell disease and majoring in thalassaemia. Sickle cell disease and serious major thalassaemia, inherited blood disorders. See the e-learning program for more information. They affect haemoglobin and its oxygen transport capacity. Individuals who have one of these conditions require lifelong care and treatment. People who are carriers are healthy and unaware of their status unless they have a specific blood test. Carrier women and 'at-risk' couples having babies with major haemoglobin disorders need information, advice, and counseling to make choices for pregnancy. This includes the decision to have a prenatal diagnosis, and to take further action if they choose to. This means that screening should occur early in pregnancy, by 10 weeks gestation. This allows time for the next action as needed. Early screening usually results in greater absorption of prenatal diagnosis (PND), which should ideally be done by a gestational age of 12 weeks + 6 days. All components of the antenatal filtration program path must be enforced for effective filtering. Stages in the line work most efficiently with the coordination of a multidisciplinary team of professionals. These include: midwives, screening coordinators and maternity services counselling teams of voluntary sector primary care teams To ensure quality services are delivered, there must be named individuals who have primary responsibility for each stage of the pathway. These stages are: the identification of eligible populations provides information before screening and completion of family origin questionnaires (FOQ), along with obtaining blood samples that process blood samples and reporting the results offering testing to all biological fathers of babies where the mother has been identified with haemoglobinopathy communicating blood test results to the baby's mother and father (if relevant) performing actions, such as PND, based on the diagnosis of parental decisions (if requested) of babies at risk of inheriting major haemoglobin disorder referrals from affected individuals for treatment and maintenance 1. Prevalence There are 2 approaches to delivery of screening programmes based on the geographical prevalence of haemoglobinopathy conditions in high-risk populations living in the UK. A list of high and low prevalence trusts is available for more information In the belief of low prevalence, where less than 1% of the booking blood is received by the is a positive screen: the red blood cell index will screen all women (regardless of family origin) for FOQ thalassaemia used as an early screening tool to identify women, or the baby's biological father, at high risk of becoming carriers for sickle cells, and other haemoglobin variants in which both parents fall into high-risk groups, Blood screening tests for haemoglobin variants should be offered to women In high prevalence trust, where 2% or greater of the blood order received by the laboratory is a positive screen: all women should be offered blood screening tests for sickle cells, talasemia and other haemoglobin variants, regardless of the origin of the Trust family that falls between the cut-off for high prevalence and low prevalence (between 1% should continue to use their current antenatal screening algorithm and monitor their position each year. In high and low prevalence beliefs: where a woman is diagnosed with haemoglobinopathy, the baby's biological father (regardless of family origin) should be offered screening for sickle cells, talasemia and other haemoglobin variants, it is important to note that not all haemoglobinopathies will be diagnosed and where there are inconclusive results, systems should be in place to follow up on women/couples where relevant complete paper or electronic FOQ should accompany all blood samples to laboratory examinations should be in place to ensure all women have been offered screening, and the results have been followed up appropriately There are detailed algorithms for processing antenatal samples in high and low prevalence areas, outlined in the laboratory handbook. If there is an amalgamation between high prevalence and low prevalence trust, a high prevalence screening policy should be adopted on both sites. 2. Reservations for antenatal care 2.1 Choice and consent All women must receive information about antenatal screening tests in early pregnancy, before they are required to make screening decisions. This should include information about when results will be available after filtering absorption. There should be an opportunity to discuss screening options with a professional who is informed of the condition. Health professionals who offer screening tests should make sure women understand the test, have given their consent for antenatal screening and are aware of the options that will follow if the test is positive. When offering screening for sickle cells and thalassaemia, health professionals should: provide oral and written information about screening tests, use the Screening test booklet for you and your baby offers women the opportunity to discuss screening tests and its decisions offer resources to address specific needs that women have such as literacy, visual impairment, language to be aware of, and sensitive to, values and women and support women to make informed decisions for the approval of their records or non-consenting screening in the woman's maternity records communicates non-consent for screening to the appropriate professional, including laboratory staff establishing whether the couple is aware of their screening status or not. Couples known to be 'at risk' should be fast-tracked for PND women on time and the baby's biological father (if relevant) should be offered screening for sickle cells and talasemia in each pregnancy, regardless of their previous results. However, it is only necessary to offer screening once in the same pregnancy. If a woman is screened in an area of low prevalence, but chooses to give birth in a maternity unit in a high prevalence area, the current results are sufficient, and do not need re-screening. If a woman changes an NHS provider during pregnancy, there is no need to repeat the blood test if the results are available. In both cases, previous results must be from laboratories accredited by the UK Accreditation Service (UKAS) and consistently, unequivocally, well documented and interpreted and reported in the testing algorithms in the laboratory handbook. During reservations for antenatal care, it is important for health professionals to collect information relevant to sickle cells and thalassaemia screening programs. This includes questions about: adoption or possible lack of awareness of family ancestry on the part of parental fertility treatment: donor egg donor sperm either donor eggs or sperm donors currently regular blood transfusions or a history of blood transfusions (why, when, where and frequency) bone marrow history or stem cell transplantation (why, where and when) a history of haemoglobin disorders or other inherited conditions (for the elderly or in one of their families) If women consent , is a best practice for filtering samples to be taken on the first booking appointment. All women need to be informed that regular analysis of blood can reveal possible carrier status of thalassaemia. If women refuse screening for sickle cells and talasemia, the laboratory team should be aware of this information before processing a full blood count sample (FBC). If the red blood cell index reveals the possibility of talasemia, further research to confirm carrier status should not occur if the woman has not approved the screening. In low prevalence areas, women who are at low risk for haemoglobin variants can opt for screening if they wish, as there may be family history information they have not revealed at the time of booking. 3. Family origin questionnaires (FOQ) Although people of any population can have this condition, individuals from some geographic regions of the world are more likely to be genetic carriers based on their ancestry. The purpose of FOQ is to identify population groups with the highest risk of sickle cells, talasemia and other haemoglobin variants. The completion of FOQ information is the responsibility of women's health professionals for antenatal antenatal Details required: for the birth mother and father of the baby in areas of high and low prevalence to be completed in each pregnancy and sent with a blood sample to the laboratory, or accessible by laboratory teams if using electronic systems for all ancestors, to the extent that individuals can remember (at least 2 generations, but more if possible – this is especially important for individuals with mixed/dual ethnic backgrounds) In low prevalence areas FOQ information is used as an early screening tool that asks about the family origins of both parents, to assess a woman's eligibility for screening haemoglobin variants. If the woman falls into a high-risk group she should be offered screening for haemoglobin variants. If the woman falls into a low-risk group, but the baby's biological father falls into a high-risk group, then the woman should be offered screening for a variant of haemoglobin (regardless of her family's origin). In areas of high prevalence and low FOQ: should accompany all blood samples to the laboratory, or relevant information should be accessible to laboratory teams if using electronic requests can avoid unnecessary father testing and unnecessary anxiety for parents when completed accurately relevant in the interpretation of red blood cell index, especially when high-risk screening groups of alpha zero thalassaemia aid accurate DNA analysis of prenatal diagnosis samples, ensuring that relevant genotypes are included in tests If women refuse screening, there must be a system to notify the laboratory team of this information before processing fbc samples. Nhs Sickle Cell and Thalassaemia screening programmes produce paper FOQ forms as templates. Integration of FOQ categories into local antenatal filtering forms or incorporated into electronic request driven systems. The flexibility of the national template should be reflected locally, and the categories are constantly updated if any changes are made. Current FOQ forms can be ordered from national print providers. 4. The condition and state of the operator to be detected There are more than 1,000 variants of haemoglobin and thalassaemia mutations, but not all are clinically relevant. National screening programmes in the UK have determined significant haemoglobinopathies that must be detected by antenatal filtration. The reason for choosing the country and condition of this operator is based on a high-risk population living in the UK. Significant maternal haemoglobin condition (this is important for maternal care) This is: Sickle cell disease: Hb SS (also called sickle cell anemia) Hb SC Hb SDPunjab Hb SE Hb SOArab Hb S/Lepore Hb S/β(0; +) thalassaemia Hb S/δβ thalassaemia β thalassaemia major/intermedia Hb Lepore/β thalassaemia Hb E/β thalassaemia Hb H Disease (- -/-α) Operator states in mothers This is: sickle cell carrier (HB AS) carrier haemoglobin C (Hb AC) D carrier (Hb ADPunjab) haemoglobin E carrier (Hb AE) haemoglobin OArab carrier (Hb AOArab) haemoglobin Lepore carrier (Hb A/Lepore) β thalassaemia carrier Aβthalassaemia delta beta (δβ) thalassaemia carrier alpha zero (α0) thalassaemia carrier (- -/αα) hereditary persistence of fetal haemoglobin carriers (HPFH) The state of other heterogous compounds includes one or more of the above carrier states. Another homozygous state of the above operator's condition. 5. Screening for haemoglobin variants In low prevalence areas information about women and biological fathers of infants in FOQ, along with its approval, determines which women should be screened for haemoglobin variants. In the area of high prevalence all approved women are screened for haemoglobin variants regardless of their family origins. 6. Screening for beta thalassaemia All women in areas of high and low prevalence should be offered screening for thalassaemia. The home screen for talasemia risk involves a review of the full blood count: haemoglobin (Hb) - the normal value in pregnancy equals, or above =>110g/l. A low value can indicate the average cell volume of anemia (MCV) - the normal range is 77-95 fl. Low values can indicate the production of deficient haemoglobin such as iron deficiency anemia or thalassaemia meaning haemoglobin cells (MCH) - the normal range is 27-32 pg. Low values seen in thalassaemia or iron deficiency anemia If MCH is lower than 27 pg, Hb A2 should be measured. The A2 range between 3.5% and 8% is common for beta thalassaemia operators. Screening for beta talasemia can sometimes be complex and may require further investigation or DNA for a definitive diagnosis. 7. Screening for alpha zero thalassaemia There are no direct tests in antenatal screening laboratories to diagnose carriers of alpha talasemia, and DNA is required for definitive diagnosis. Approved laboratories for DNA testing are listed in the lab handbook and at the end of this chapter. Alpha+ (alpha plus) talasemia is not considered clinically significant, and suspected carriers will not require further investigation. Alpha0 (alpha zero) talasemia is clinically significant and most commonly found in people with ancestry from: Eastern Mediterranean (Cyprus, Greece, Sardinia or Turkey) Southeast Asia (China, Hong Kong, Thailand, Taiwan, Cambodia, Laos, Vietnam, Myanmar (formerly Burma), Singapore, Indonesia or the Philippines) Screening policy in the UK aims to identify couples where both parents are alpha zero thalassaemia carriers (alpha0) and their babies are at risk of inheriting alpha thalassaemia majors (Hb Bart's Hydro If the women's preliminary screening results indicate that she may be a carrier of alpha zero thalassaemia, but only one parent comes from a high-risk group and the other parents do not, then no further investigation is required. If the results are early women indicated that she may be the operator of alpha zero thalassaemia, thalassaemia, both biological parents come from one of the high-risk groups (see list above), so the baby's father should be offered a screening test. If both parents' test results indicate the possible carrier status of alpha zero thalassaemia, then blood samples from each parent should be sent for DNA analysis to confirm if they are alpha zero thalassaemia operators. If both parents are carriers, then PND should be offered. Only a small number of major alpha thalassaemia cases occur in the UK each year. 8. Reference of antenatal samples to dna laboratory for analysis of haemoglobinopathy mutation Most operators are diagnosed in antenatal screening laboratories. However, it may sometimes be necessary to refer samples for DNA analysis. See the list of laboratories at the end of this chapter. Local processes should be put in place to advise and notify maternity or counselling teams whose samples need to be referred for molecular studies. A special consent form for DNA analysis must be completed by the baby's biological woman and/or father if this investigation is required, and additional blood samples collected as needed. 9. Problems that may arise during regular antenatal filtering during filtering some operators may be missed and it is possible for false positive and false negative results to be reported. Assuming foq has been resolved accurately, carrier countries can be missed because: some operators β-thalassaemia may have: silent or almost silent genotypes, associated with hb A2 limits their operator status levels obscured by severe iron deficiency anemia; medical conditions (B12 or folate deficiency; liver disease); or treatment (such as HIV therapy); or haemoglobinopathy alpha0 other thalassaemia occurs outside the origin of prescribed high-risk families or in women with anemia, haemoglobin is significantly masked by blood transfusions or bone marrow transplants that are not reported haemoglobinopathy present in egg donors or sperm donors where unan declared or untested variants of second haemoglobin can be sustained by haemoglobin A or other haemoglobinopathy. In low prevalence areas, other than the above, operators state that occurring in individuals who are outside the specified high-risk family origin, or in individuals who have not accurately disclosed their family origin, may be missed. If a woman is booked for antenatal care for subsequent pregnancy and receives screening, health care professionals should: offer women screening for haemoglobinopathy, regardless of previous screening history of completing FOQ, or have a system to make family origin information accessible to the laboratory team if using an electronic test that asks to take a blood sample and send it to the laboratory If the carrier or affected woman is identified, the father infants should be offered a screening test, regardless of previous screening history. If it is not possible to test the biological father of the baby in pregnancy and previous results are being considered for use, please check that this is the same father. Previous results should be from laboratories accredited by UKAS and consistent, firm, well documented and interpreted and reported in testing algorithms in laboratory handbooks. This information should be noted in the woman's record for the current pregnancy. If a written copy of the result is available, it must also be included in the woman's record. Guidance on collecting biological father information has been provided by the Fatherhood Institute. 11. Screening results must be reported within 3 business days after receipt of blood samples in the laboratory. Each maternity unit must have a robust process to inform women of the results. If further investigation is required, an interim report will be provided by the laboratory until a final report is available. Midwives are expected to act on this interim report and begin screening the baby's biological father, if he is available. If nothing unusual is detected in the father's results, then the risk of the baby inheriting major haemoglobin disorders can be excluded. If the baby's biological father is not available for screening, confirmed maternal results are required before PND can be offered. PND should not be done based on the initial screening results. All women should be informed of their screening results (normal, carrier, inconclusive, haemoglobin disorder) and local protocols and pathways should be in place to support this. Women who have inconclusive results, operators, or are affected should be offered the opportunity to receive the results in a face-to-face counseling session, combined with the provision of written notice of the results. Information leaflets about the status of a particular operator must be provided and information about the operator's results must be provided in writing. Screening results should be accessible to all health professionals involved in the screening program, and details should be recorded in the electronic maternity records of primary care records Laboratory will report one of the following results: No abnormalities detected, Hb AA Approximately 97% of women screened will have these results. No biological father testing is required. Non-significant carriers Are not clinically significant and there is no risk of babies inheriting major haemoglobin disorders. No biological father testing is required. Significant carrier It is clinically significant and the baby may be at risk of inheriting major haemoglobin disorder if both parents are carriers. About 2.5% or 1 in 40 pregnant women will be identified as carriers. Biological father testing is required. Benign haemoglobin disorder It is, for example, Hb CC, Hb DD or Hb EE. Women should be referred for haematology consultations but often there is no special treatment during pregnancy A biological father examination is required, and the baby may be at higher risk (50% chance) of inheriting haemoglobin disorder if the father is a significant carrier of haemoglobinopathy. Clinically significant disorder It embraces sickle cell disease, e.g. HB SC, or the condition thalassaemia. Most of these women are aware of their condition but sometimes this can be identified for the first time during antenatal screening. Urgent referrals to obstetrics teams and obstetric consultants are required. Joint medical and midwifery care and close monitoring throughout pregnancy is required, and women should be booked for hospital delivery. Testing of the baby's biological father is required. There is a higher risk (50% chance) of babies inheriting haemoglobin disorder if the biological father is a significant carrier of haemoglobinopathy. Inconclusive results Further testing of women may be required depending on the suspected variant. For this couple, the results should be explained to the woman and testing should be offered to the baby's biological father. If the baby's biological father does not have haemoglobinopathy there is no risk of the baby inheriting major haemoglobin disorder. There may be no further maternal testing required (this is determined locally). However, if there is no further testing of the woman she will remain unaware of her specific carrier status and risk to future pregnancies if she changes partners. If the baby's biological father does have haemoglobinopathy, then further maternal testing may be required for an accurate assessment of the risk of the fetus inheriting a significant haemoglobin condition, and the partner needs to be followed up appropriately. See Appendix 5 for examples of usable counseling forms. 12. Follow-up screening for clinically significant results (operators, affected, inconclusive, benign haemoglobin disorders) Results should be immediately communicated to women. Mothers need time to arrange screening for the baby's biological father and consider the implications for pregnancy and her unborn child. Receiving positive screening results can be emotionally traumatic for women, as this may not be anticipated. A trained professional should be available to explain all significant results. Time is of the moment in making decisions for further investigation. Women should be given written confirmation of their results along with explanatory leaflets. The woman should be invited for counselling (template letter available) and be aware: implications for her being a carrier or have implications for this haemoglobin condition for this and future pregnancies the fact that the baby's biological father needs to be tested to assess the risk to the baby of choice available for pregnancy the fact that other members of her family can also be carriers and that they can request testing by their GP in specialist centers, especially if they are plan to have 13 babies. Beta thalassaemia carrier Beta thalassaemia carrier has inherited the unusual globin beta gene from one parent and a normal gene from another. Where a person is a carrier of beta thalassaemia, it is important for them to be aware of that: an unusual gene can be passed on to their children even if their child has inherited a gene that cannot be diagnosed at birth with a routine infant's blood spot examination if the parent chooses, the baby can be tested when they are over 9 months old to confirm their carrier status When arranging and encouraging screening of the baby's father , health care professionals should be sensitive to possible paternity issues, and clarify to women the importance of screening the baby's biological father. The baby's biological father should be invited for counseling and blood tests as soon as possible in all cases where the woman is identified with haemoglobinopathy or if there are inconclusive results (regardless of her family's origin). Leaflets, Sickle cells and thalassaemia screening: information for the father, and letters should be given to the father before screening. Fathers should be offered screening in every pregnancy as for mothers. If possible couples should have counseling sessions together to discuss women's results and implications for pregnancy, and for fathers to be tested. Sessions should be professionally trained in providing haemoglobinopathy information. If a joint appointment is not possible, then the father should be offered his own appointment to discuss the results of the screening and to conduct a blood test. The health professional responsible for screening the baby's biological father should provide the laboratory with information about the woman when the father is screened so that the results can be attributed. The fathers' test results should be recorded in the mother's antenatal hand-held notes and on counseling records. The Fatherhood Institute has provided guidance on collecting biological father information. 15. Maximizing the absorption of father testing It may be difficult for biological fathers to be present for screening, or they may be reluctant to be screened. Possible barriers to accessing screening include: the assumption by the man that he has been tested and has negative results, based on the fact that he may have had unrelated blood tests in the past a lack of understanding of the test, the significance of being a carrier, how the condition is inherited and the risk to their baby of the stigma that may be attached to screening men who think that if they are good they cannot be carriers , how the condition is inherited and the risk to their baby the stigma that may be attached to screening men who think that if they are good they cannot be pregnancy carriers and blood tests are seen as part of the female world, compounded by antenatal clinic system difficulties with taking time off work to attend appointments for blood tests fear needles Some may have been screened before, either in the UK or abroad, and do not recognise the need for re-screening. Health should explain that: we need to see a copy of the laboratory report with previous screening results that the man's previous screening may not include all variants tested in the screening test results of the English screening program should be from an accredited laboratory the previous results need to be confirmed when having a PND Health professional who reviewed the results of the previous father screening should document this in the women's records and, if possible, keep a copy of the results. Points to consider include: time - can screening be offered at a convenient time for the father, for example outside his normal business hours? location - can filtering be offered in a more convenient or neutral location? socio-cultural barriers to absorption (listen to what the mother says) the possible benefits of direct contact between health professionals and fathers to support the need to conduct blood tests, and to highlight the importance of screening If the baby's biological father is unavailable, unknown or refuses testing then health care professionals should discuss with the woman: does she stay with the father/contact during this pregnancy? have biological fathers been tested in the past and have any confirmed results? Is he willing (or able) to deliver letters and leaflets to fathers? if he doesn't stay with dad and is no longer in touch, can he provide his details so that information about the test can be sent to him directly? The responsible health care professional should try to make direct contact with the baby's biological father, with the woman's consent, if the woman is unwilling or unable to make contact, to offer information and blood screening tests. 16. Follow-up after paternal examination the results should be reported to the designated health professional within 3 business days from the time of receipt of blood samples in the laboratory. All results of the father's examination should be reviewed and attributed to maternal outcomes. Checks should be carried out to ensure the father's results have been received and acted upon. Fathers should be informed of the results, whether this is clinically significant or not. The operator must receive the information in writing, along with the appropriate carrier handout if relevant. 17. Father carrier results (the baby is at risk of inheriting benign haemoglobin disorder) If the man is identified as a carrier of haemoglobinopathy then the spouse should be invited to a follow-up counseling session and the results are explained face-to-face. If a partner is at risk of having a baby with benign haemoglobin disorder that does not require long-term care (e.g. conditions such as Hb CC; Hb DD; Hb C/Beta thalassaemia) then this should be explained and the pair reassuring. Confirmation in writing of the status of the carrier and the appropriate carrier flyer should be given to the father. Prenatal diagnosis is not required one of these conditions. 18. Father carrier (babies at risk of inheriting major haemoglobin disorder) Women and couples at risk of having an affected baby should be offered a prenatal diagnosis (PND) as soon as possible, ideally with a gestational age of 12 weeks + 0 days. They should: be offered urgent counseling appointments with appropriately trained professionals (e.g. a professional trained in approved courses such as genetic risk assessment and counseling courses) to explain the risks to their babies, details about the conditions their baby can inherit, and options for pregnancy. Explanatory leaflets[footnote 1] [footnote 2] should be given to couples to support counselling sessions immediately referred if they decide to continue with the NHS SCT Screening Programme PND presenting some challenges for practitioners. Defining family origins, inheritance and ancestry Identifying family origins, inheritance or ancestry is integral to screening for sickle cells and thalassaemia. It is important this is not equated with nationality. FOQ identified the groups with the highest risk of sickle cells, talasemia and other haemoglobin variants. This includes other Mediterranean and European populations which, under normal circumstances, may be 'missed' for screening. Practitioners need to help parents complete FOQ for screening in low prevalence areas to ensure the laboratory has the correct information for screening and analysis of results. Cultural influence during screening Perceptions of what carrier status means can affect family attitudes to screening. In some groups with the highest risk of haemoglobinopathies, there may be religious or cultural beliefs that influence decisions about prenatal diagnosis and termination of pregnancy. Research confirms this and practitioners need to be aware of relevant issues. The genetic properties of sickle cell disease and primary thalassaemia mean it is important to link information from parental outcomes with infant screening results. Local systems should be put in place to facilitate this. 21. Reference Reference