28 October 2008 Definition consultation feedback: Specialised Haemoglobinopathy Services (all ages) - Definition No. 38

Briefing for 30th October meeting

- This feedback document was produced on 28th October 2008 by Rob Couch of the NSC Team. The NSC Team has yet to review this feedback.
- Please can the 30th October meeting contributors review each of the comments below and provide short notes on whether they agree/disagree and add their proposed changes, where necessary, in the right hand column.
- These notes will be reviewed with the original feedback by the NSC Team to produce a further definition draft for review at the 11th November DH Clinical Services meeting.
- The final definition will be submitted to the NSCG meeting on 16th December.

Feedback contributors

Professor Irene Roberts, Imperial College London Email of 02nd Oct. 2008

Dr Nicola Bienz, Heatherwood and Wexham Park Hospitals Email of 03rd Oct. 2008

Professor Barbara Bain, Imperial College London Email of 07th Oct. 2008

Dr Jenny Welch, Sheffield Children's Hospital Email of 15th Oct. 2008

Professor Simon Dyson, Unit for the Social Study of Thalassaemia and Sickle Cell, DeMontford University Email of 16th Oct. 2008

Dr Salah Tueger, Countess of Chester Hospital Email of 20th Oct. 2008

Dr Marie Donohue, Nottingham University Hospital Email of 22nd Oct. 2008

Beryl Juma, Leeds Sickle Cell & Thalassaemia Service, Chapeltown Health Centre Email of 23rd Oct. 2008

Dr Allison Streetly, NHS Sickle Cell & Thalassaemia Screening Programme Email of 24th Oct. 2008 via Roma Haigh

Dr Shah, Whittington Hospital Email of 27th Oct. 2008 sent via Roma Haigh

Dr Claire Hemmaway, Barking, Havering and Redbridge Hospitals Email of 20th Oct. 2008 via Brenda Allen of EoE SCG

Dr Trevor Baglin, Addenbrookes Hospital Email of 14th Oct. 2008 via Brenda Allen of EoE SCG

Dr Maggie Harding (personal comments), London SCG Email of 17th Oct. 2008

Sue Assar, Acting Chief Exec., Luton PCT Email of 27th Oct. 2008 via Brenda Allen

South Central SCG Email of 21st Oct. 2008 from Sally Nelson

South East Coast SCG & local providers Email of 22nd Oct. 2008 via Nick Haslem

General comments on Definition No. 38 Specialised Haemoglobinopathy Services (all ages)		
Contributor & source	**Comment**	**Action required & by whom**
Professor Irene Roberts, Imperial College London Email of 02nd October 2008	Dear Roma, I read this a few days ago for the Royal College of paedaitrics and Child Health and fed back my comments in note form to their representative to add to other paediatric views. But I have now made these comments a bit more logical and easy to follow (I hope!) and attach this new version in response to the consultation. As you'll see, my may concern is that by generalising too much in some areas, in particular 'down playing' the complexity of the diseases, there is a risk of people thinking it is very straightforward to look after these patients and it can mostly be done locally. Sadly, at the moment, this has a strong chance of leading to deaths and mismanagement. So local care is great BUT it isn't straightforward and I think more attention needs to be paid, at this stage of services in the UK, yo safe practice. Once good practice becomes more widespread, more local care will be possible- but not yet! My personal view of course- but I'm sure it's what our CNS and the rest of our team would say as well!	
Dr Nicola Bienz, Heatherwood and Wexham Park Hospitals Email of 03rd October 2008	Personally I would take issue with some of the statements about what will consitute "specialist centre" activity and what will be "local hospital" activity, but ultimately I guess that it will be for the service providers in each unit to determine and agree appropriate agreed pathways for their local population of patients.	
Professor Barbara Bain, Imperial College London Email of 07th October 2008	Comment by NSC Team - In addition to her detailed comments, Professor Bain has very kindly made many small corrections to the text. Only those that alter the meaning of the text and should be reviewed by the 30th October meeting have been included in this consultation feedback, the others will be completed by the NSC Team.	
Dr Salah Tueger, Countess of Chester Hospital Email of 20th October 2008	Thank you for your email regarding the new SSNDS document. I work in a trust with very low prevalence of SCD. I think generally the document is very useful and informative, we normally tend to manage those patient locally and refer if needs specialist care. I would suggest if you include contact details of high prevalence trusts who deals with SCD and thalassemia as it may be useful to contact them for advice on management of complicated cases.	

General comments on Definition No. 38 Specialised Haemoglobinopathy Services (all ages)		
Contributor & source	**Comment**	**Action required & by whom**
Dr Maggie Harding (personal comments) London SCG Email of 17th Oct. 2008	This is a very detailed description but does not consistently spell out what is recommended as a specialised rather than local or community service – both the latter will be commissioned by PCTs. From a high prevalence, London perspective much of what appears to be recommended as specialised would be better commissioned at sector level through clinical networks and care pathways (cf my comments on liver failure which may be less common than sickle cell & variants thereof in London). However the consistent failure of Health Authorities & PCTs to actively commission an adequate service for these patients (cf the damning NCEPOD report which must relate in large part to care within London given the prevalence) probably justifies a specialised commissioning focus. However NCEPOD weaknesses relate to care in local hospitals commissioned by PCTs so inclusion in SSNDS will only address this if robust shared care arrangements, with SSNDS commissioned monitoring of local hospitals by specialist centres can be enacted as clinical networks have no remit for clinical governance across the pathway of care. (**Dianne** please comment on how this might be achieved through your knowledge of haemophilia shared care/quality assurance by centres of units).	
Dr Trevor Baglin, Addenbrookes Hospital Email of 14th Oct. 2008 via Brenda Allen of EoE SCG	No real comments on this other than to say I agree with it all. It would seem reasonable to include in specialised services. Our local problem is lack of expertise and consultant time for adult patients but the new appointment to Papworth/Addenbrookes includes specialist care for this group of patients so we do have a plan in place to meet requirements.	
South East Coast SCG & local providers Email of 22nd Oct. 2008 via Nick Haslem	It was felt that the definition set clearly describes the major haemoglobinopathies and the level of services required for managing them. It was also noted that this approach is advocated by the SCT programme centre.	
Sue Assar, Acting Chief Exec., Luton PCT Email of 27th Oct. 2008 via Brenda Allen	Generally the definition sounds fine. Would need to ensure links between annual review (specialist) and routine (PCT commissioning). Not sure where specialist service sits. Luton will have the highest levels of these conditions in the SHA.	

General comments on Definition No. 38 Specialised Haemoglobinopathy Services (all ages)		
Contributor & source	**Comment**	**Action required & by whom**
Dr Allison Streetly, NHS Sickle Cell & Thalassaemia Screening Programme Email of 24th Oct. 2008 via Roma Haigh	**Summary:** The NHS Sickle Cell & Thalassaemia Screening programme welcomes the development of this definition which has been in need of attention and development for some while. We have some points to make relating both to the specific definition and also to ensuring coherence "across the piece". We strongly recommend that all aspects of screening and care relating to haemoglobinopathies are fully "joined up". We also want to ensure consistency with the approach being taken to metabolic conditions and the Newborn Bloodspot Screening Programme. We draw your attention to the current draft of the medical genetics definition. This gives the following quote "the haemoglobin disorders are serious blood disorders requiring specialist care and treatment affecting 1:4000 – 1:5000 [now shown to be 1:2000] live births. Currently there is no clarity about the commissioning arrangements for these services, which are funded through multiple sources. They do not form a separate definition and they are included here to highlight the need for appropriate attention". Please also refer to Appendix 1. **Specialist service definition:** The main points we wish to highlight relate to the need to ensure that commissioning is effectively co-ordinated to cover all aspects of the needs for care for these conditions. It is important that new arrangements do not detach the specialist aspects of care from the need for a managed clinical network with a strong emphasis on support in the community even though community services may not be within the specific remit of specialist commissioning. **Need for consistency across definitions:** We highlight here the need for some joined up work and cross linkage to other relevant definitions. We note that there is currently no definition for the specialist commissioning role in relation to screening although a role has been agreed in outline. Without such a definition it is difficult to make cross links. There needs to be clarity about what will be included in medical genetics and what will be included in any future screening definition. This is rather unclear if using genetics methods – PIGD will be linked with the medical genetics definition but PND will not. We note the concerns raised recently about the high costs of PND for haemoglobinopathies and consider that geneticists involvement in advising	

	that samples be sent overseas (eg to Holland etc) is unhelpful but such advice serves to highlight the need for a commissioning "grip" on this issue. There is also the need to ensure a consistent approach to newborn bloodspot screening which is currently proposed to be removed from the medical genetics definition. Will this be listed as part of the new screening definition or will it be incorporated within the proposed metabolic definition? We as a Screening Programme do not have any strong view on which the correct place is but we do think that there should be a consistent approach across haemoglobinopathies and metabolic conditions. In particular there is a need to mention the sickle cell aspect of newborn screening *including the second line testing which should only be provided by centres with expertise in haemaglobinopathies* – and with reference to the clinical networks (as mentioned above). **Conclusion:** We welcome the development of this definition. We wish to see it go forward but also wish to ensure that all the relevant cross-links are made.	

General comments on Definition No. 38 Specialised Haemoglobinopathy Services (all ages)		
Contributor & source	**Comment**	**Action required & by whom**
Professor Simon Dyson, DeMontford University Email of 16th October 2008	Dear Colleague, You have invited comments on the Draft Specialist Haemoglobinopathy Services Definitions, the purpose of which you state is to "identify activity that should be regarded as specialized and therefore within the remit of PCT collaborative commissioning". My comments are as follows: There is little in the definitions that would address primary prevention for sickle cell disorder (SCD) in the community. This is because the definitions do not consider where primary prevention could take place. The primary care-givers in the community (primary care-givers who could recognise and act upon early signs of strokes and crises) are often the school teacher and school nurse, rather than the parent, given that children may spend up to 30,000 hours over a school career in school. This is important because: 1) There is currently no obligation on a Director of Public Health to notify schools in the local area of the numbers of children being born in that area for each school intake cohort, figures that should be available from universal neo-natal screening in England. Whilst most local authorities agree that children with SCD should be given an individual health care plan (IHCP), only 14/107 could state how many children under their jurisdiction had an IHCP and only 15/107 could even state how many children with SCD they had in their schools. Only 2/107 had any written policy on children with SCD. Thus at this level SCD as a potential issue affecting the health and safety of pupils is relatively invisible in education policy terms*. 2) Around 10% of parents do not tell the school their child has SCD for fear of the way their child will be treated. Opportunities to prevent sickle cell complications (keeping warm; being allowed to drink water in class in order to remain well-hydrated; being permitted to go to the toilet, as children with SCD cannot concentrate urine as easily; avoiding strenuous exercise, such as cross-country runs in the rain; avoiding stress) are not currently highlighted to schools in DCFS Guidance. Children with SCD report not being cared for adequately at school. For example a majority of children say that they have been prevented from going to the toilet when needed; about half report that they are not permitted to drink water; about a third that they are made to take unsuitable exercise. All of these could trigger a severe sickle cell painful crisis. Furthermore, 30% say that they have	

	been called lazy when in fact they are tired from their anaemia. These percentages represent hundreds of children with SCD reporting these experiences in school.** 3) A key resource in prevention and liaison is systematically lacking: the school nurse, for whom PCTs are generally responsible. There are only around 2,500 full-time equivalent school nurse posts for all schools in UK. Most school nurses cover 5-10 schools. A school nurse who is in school for less than half a day a week is not going to be able to help school teachers identify and respond adequately to SCD in the community, nor help teachers with basic preventive measures that could dramatically reduce the impact of SCD complications such as crises and strokes on health services. * 2007 Survey of all local authorities in England on their response to sickle cell. Dyson, SM; Abuateya, H; Atkin, K; Culley, LA; Dyson, SE; and Rowley, DT (2008) Local authorities and the education of young people with sickle cell disorders (SCD) in England *International Studies in Sociology of Education* 18 (1) 47-60. ** 2008 National Survey of 569 children living with SCD in England (unpublished data from Economic and Social Research Council Project, Grant Number ESRC RE000-23-1486)	

Preface		
Contributor & source	**Comment**	**Action required & by whom**
South Central SCG Email of 21st Oct. 2008 from Sally Nelson	We are concerned by the statement that "inclusion of a treatment or intervention in a definition should not be taken to mean that there is established evidence of clinical or cost-effectiveness". Our view is that no intervention should be recommended for commissioning as a specialised service unless such evidence exists. To do so undermines the credibility of the definition set.	Note – NSC Team have recently raised this issue with Sally Nelson, no further feedback necessary.

1. Introduction and general description		
Contributor & source	**Comment**	**Action required & by whom**
Dr Nicola Bienz, Heatherwood and Wexham Park Hospitals Email of 03rd October 2008	page 2, 2nd para whilst thalassaemia affects mostly Asian andMediterranean peoples (.....also prevalent in Chinese and other pan-Pacific origins) page 2, 4th para SCD includes the genotypes HbSS, HbSC, HbS/beta thalassaemia, chronic fatigue, stunting of growth due to the effects of SCD patients are intermittently sick....change "sick" to "unwell"	
Professor Barbara Bain, Imperial College London Email of 07th October 2008	This definition focuses on people with thalassaemia major, sickle cell disease (SCD) and other rare inherited anaemias. There are about 800 patients with thalassaemia and 15,000 with SCD in England. A large number are under 19 years **of age (Comment - instead of old)**. SCD is one of the commonest inherited conditions in England; around 300 babies are born in England each year with SCD. In contrast around 20-30 babies are born in England each year with thalassaemia. SCD predominantly affects black people, whilst thalassaemia affects Asian and Mediterranean peoples. The prevalence varies according to geographical area, being highest in urban immigrant populations, **(Comment - This is not altogether correct. Many are born in the UK and might rightly be offended by being described as 'immigrant')** particularly London, where about two-thirds of SCD patients reside. **Sickle Cell Disease** SCD includes the genotypes HbSS, HbSC, HbS beta thalassaemia, and several rarer combinations. Homozygous SCD (HbSS) accounts for about 70% of patients and is clinically the most severe **(Comment - There are some more severe, e.g. S/S-Antilles)** of the common genotypes. SCD is a complex condition characterised by bouts of severe and occasionally life-threatening acute illness (crises), increased susceptibility to specific severe infections, chronic fatigue, stunting due to the effects of severe anaemia **(Comment - I don't think there is usually any stunting, just delayed growth and then a catch up. There are not 'effects of severe anaemia' as this is a low affinity haemoglobin and oxygen delivery is good)** and progressive tissue and organ damage. SCD patients are intermittently sick, and severely affected patients miss large amounts of school and find it difficult to sustain employment. Management includes prophylaxis against infection; treatment of pain; expert management of acute, life-threatening complications; monitoring and treatment of chronic complications; and education and psychosocial support for patients and carers. About 10-20% of children and a smaller percentage of adults require regular blood transfusions, together with iron chelation therapy.	

1. Introduction and general description		
Contributor & source	**Comment**	**Action required & by whom**
Professor Barbara Bain, Imperial College London Email of 07th October 2008	**Thalassaemia Major** Thalassaemia major causes severe anaemia, bone expansion and failure to thrive in infancy and these patients require regular blood transfusions, usually starting between 6 months and 2 years of age, every 3 to 4 weeks for life. Iron accumulates as a result of regular blood transfusions, and if not **(detoxified and - deleted)** removed (chelated), will cause damage to endocrine glands, liver and heart, with death from heart complications in the second or third decade. Good organization and monitoring of transfusions, together with safe and effective iron chelation therapy are the medical goals of a thalassaemia service combined with education and psychosocial support for patients and carers to minimize disability and optimize quality of life.	
Beryl Juma Chapeltown Health Centre Email of 23rd Oct. 2008	I have spent sometime looking through Definition for Sickle Cell and Thalassaemia SSNDS number 38, and would like to commend the clinicians involved in compiling this draft. Having been a Specialsit Sickle Cell and Thalassaemia Nurse Counsellor of nearly twenty years standing, I feel they have done a good job. I however have a small observation I would like to make that is that in the second paragraph in the Introduction/description: which deals with the people that sickle cell and thalassaemia affect. In low prevalence areas where clinicians do not see many cases of Sickle Cell and thalassaemia. I feel that it should be more explicit that although sickle cell predominantly affects black people, it is not exclusively a black persons disease and the same goes for thalassaemia. In my experience, I have come accross health professionals who have have not believed that people other than black or asian people have this condition when thay have been faced with these people in their clinics or surgeries.	
South East Coast SCG & local providers Email of 22nd Oct. 2008 via Nick Haslem	It was noted that page 2 states Thalassaemia can be identified on the newborn bloodspot, which was different to local understanding in that only Thalassaemia major can be seen and that the test is not designed to detect thalassaemia generally - it is a by product of screening and only identified as these babies hav no adult haemoglobin.	

2. Rationale for the Service being included in the Specialised Services Definitions Set		
Contributor & source	**Comment**	**Action required & by whom**
Professor Barbara Bain, Imperial College London Email of 07th October 2008	Critical aspects of management need specific facilities and equipment as well as staff possessing knowledge and experience. This is the case not only for management of acute complications, but also the supervision and monitoring of long-term therapy to avoid chronic complications. Continuing advances in treatment, complex treatment decisions, new drugs and investigations are best supervised by a specialist working in a specialist centre attending to a large number of **patients** (cases – deleted) each year.	
South Central SCG Email of 21st Oct. 2008 from Sally Nelson	• We agree with the rationale for inclusion as a specialised service.	

3. Links to Other Services in the Specialised Services Definitions Set		
Contributor & source	**Comment**	**Action required & by whom**
Professor Barbara Bain, Imperial College London Email of 07th October 2008	No.21 Specialised learning disability services (adult) Comment - Why not children since cognitive impairment can start early?	NSC Team response – noted, to be passed to project lead for this definition which is about to be reviewed.

4. Detailed Description of Specialised Activity		
Contributor & source	**Comment**	**Action required & by whom**
Professor Barbara Bain, Imperial College London Email of 07th October 2008	**Levels of Care for SCD and Thalassaemia** ….Consequently in high-prevalence areas the local hospital is likely to provide much of the day-to-day care and look to the specialist centre to provide certain outreach services and joint clinics, whereas in low-prevalence areas the local hospital is more likely to refer patients to the specialist centre. However ***all*** local hospitals will need to provide prompt, effective and safe management of (**deleted** – simple) acute painful SCD episodes and follow appropriate protocols for pain management in emergency departments and in-patient environments.	
Dr Maggie Harding (personal comments) London SCG Email of 17th Oct. 2008	**Comments regarding sections 4.1, 4.2 and 4.3 plus proposed new section – 'specific therapy'** I agree that supervision of transfusion, iron chelation, prevention of neurological complications and **specific therapy** (hydroxyurea/other – **which is not specified but needs to be)** should be defined as specialised, through robust shared care arrangements if necessary at least for 5 years in London, but not in perpetuity given the high prevalence which should result I a limited range of institutions delivering specialised & local hospital level care, commissioned by sectors in due course.	

4. Detailed Description of Specialised Activity		
Contributor & source	**Comment**	**Action required & by whom**
Professor Irene Roberts, Imperial College London Email of 02nd October 2008	**4.1Supervision of blood transfusion management of SCD and Thalassaemia and exchange transfusions for SCD** Acute exchange transfusions are required for certain severe complications of SCD and are best done in a specialist centre, ideally using erythrocytapheresis apparatus, although manual exchanges are acceptable. If the patient is too unwell to travel or needs the transfusion urgently, the exchange can be done in the local hospital after liaison with the specialist centre. **Comment: For young children usually manual exchanges are simpler and far more feasible so it is important not to make manual exchanges appear generally inferior. I would suggest rewording this to:** Either manual exchange or automated exchange (erythrocytapheresis) may be used; strict adherence to protocol is essential.	
Professor Irene Roberts, Imperial College London Email of 02nd October 2008	**4.1 Supervision of blood transfusion management of SCD and Thalassaemia and exchange transfusions for SCD** Chronic transfusion therapy is ***not*** a specialised service and should be done in the ***local hospital*** close to the patient's home or work **provided appropriately qualified staff are available.** It entails regular hospital attendances every 3-4 weeks and each episode **lasts at least 4-8 hours**. The majority is straightforward top-up transfusions, but some patients with SCD will have exchange transfusion, which is technically more demanding. **Comment: While local involement is highly desirable, it is more important that there is a good quality of service from the holistic point of view and this may not always be possible locally. Episodes freqently last more than 4 hours! I have suggested amended wording.**	
South Central SCG Email of 21st Oct. 2008 from Sally Nelson	4.1 **Transfusion:** We agree that acute exchange transfusion is a specialised service, and that chronic transfusion therapy is not.	

4. Detailed Description of Specialised Activity		
Contributor & source	**Comment**	**Action required & by whom**
Professor Barbara Bain, Imperial College London Email of 07th October 2008	**4.2Supervision of iron chelation management in SCD and Thalassaemia, prescribing iron-chelating drugs, monitoring and adverse event management and optimization of compliance** There are now three licensed iron chelating drugs: deferoxamine (**Comment** - **Surely the correct up-to-date name should be used - not desferrioxamine**)(given by subcutaneous infusion), deferiprone (orally administered) and deferasirox (orally administered). All of these drugs are expensive and have significant side effects. The overall supervision of the patient resides with the specialist centre and will cover: choice of drug, monitoring for efficacy and side effects, dose adjustments and changes to the chelation regime although shared care arrangements may be applied for ongoing monitoring.	
South Central SCG Email of 21st Oct. 2008 from Sally Nelson	**4.2 Iron chelation:** We agree that overall supervision of patients on home based chelation therapy is a specialised service.	
Dr Allison Streetly, NHS Sickle Cell & Thalassaemia Screening Programme Email of 24th Oct. 2008 via Roma Haigh	**4.2 – Iron chelating drugs** ***Expensive drug costs*** – we understand that it is proposed to include two iron chelators on the DH list of high cost drugs excluded from tariff (deferasirox and deferiprone) . To be consistent , we suggest that the third iron chelating drug - desferrioxamine. - mentioned in the definition should also be included on the exclusion list. We also urge the DH to confirm this proposal as soon as possible to reduce any uncertainties between providers and PCT commissioners. ***Hydroxurea*** is a drug currently prescribed off-licence (and therefore cheaply) for sickle cell disease in some more severe cases and this drug is likely to be licensed shortly specifically for use in children with sickle cell disease. This should be flagged up in the definition alongside the iron chelators as it will shortly be costing the NHS considerably more.	Note – these comments are repeated in Section 5 – identifying and costing activity.

4. Detailed Description of Specialised Activity		
Contributor & source	**Comment**	**Action required & by whom**
Professor Irene Roberts, Imperial College London Email of 02nd October 2008	**4.3 Prevention and management of neurological complications of SCD including Transcranial Doppler screening in childhood, specialised neuroradiology, neurology and neuropsychology services** Clinically apparent (overt) ischaemic stroke is seen in about 10% of children. Transcranial Doppler (TCD) screening is a non-invasive method of identifying children at risk. All children with HbSS will require annual TCD scanning from the age of 2 and abnormal and borderline (seen in about 25%) will need to be repeated frequently and investigated with MRI/MRA scans. TCD scans are done by an experienced sonographer (ie. experienced nurse, doctor or radiographer) under the direct supervision of the specialist centre and subject to a quality assurance system. **Comment: MRA is also important (this is acknowledged elsewhere but I think it is important to include it here.** Neurological symptoms and abnormal or inadequate TCD scans are investigated by MRI and magnetic resonance angiogram (MRA) of the brain. These scans reveal a complex spectrum of ischaemic and cerebrovascular damage, whose evaluation and treatment requires specialist experience. **Comment: It is potentially misleading to imply this is only for inadequate scans. Suggested amendment is shown.**	
South Central SCG Email of 21st Oct. 2008 from Sally Nelson	**Neurological complications of SCD:** We agree the prevention and management of stroke and other neurological complications is part of the specialised service.	
Dr Shah, Whittington Hospital Email of 27th Oct. 2008 sent via Roma Haigh	**Paragraph of section 4.3:** Clinically apparent (overt) ischaemic stroke is seen in about 10% of children. Transcranial Doppler (TCD) screening is a non-invasive method of identifying children at risk. All children with **HbSS** (**Comment** : ? S beta 0 thal as well should be here or is HbSS being used to mean sickle children rather then genotype. Please clarify this point.) will require annual TCD scanning from the age of 2 and abnormal and borderline (seen in about 25%) will need to be repeated frequently and investigated with MRI scans. TCD scans are done by an experienced sonographer (ie. experienced nurse, doctor or radiographer) under the direct supervision of the specialist centre and subject to a quality assurance system.	

4. Detailed Description of Specialised Activity		
Contributor & source	**Comment**	**Action required & by whom**
Professor Irene Roberts, Imperial College London Email of 02nd October 2008	**4.4Management of severe and life-threatening acute complications of SCD and Thalassaemia** **Acute complications in SCD** The most severe episodes with life threatening acute complications will be treated by the specialist centre, these include: • fulminant sepsis • acute sickle lung syndrome • acute splenic or hepatic sequestration • ischaemic and haemorrhagic stroke • subarachnoid haemorrhage, acute renal failure • multi-organ failure • biliary obstruction • fulminant priapism • post-transfusion hyperhaemolysis • sickle retinopathy/central retinal artery occlusion • osteonecrosis of major joints (eg hip, shoulder). **Comment: Suggested important additions to this list.** In these cases, management is extremely challenging since there are features of these acute events which resemble general medical emergencies, yet the management may be very different. Exchange transfusion, intravenous analgesia, inhaled oxygen, mechanical ventilation and/or CPAP, incentive spirometry, careful attention to fluid balance and appropriate choice of antibiotics all form part of the management. **Comment: Suggested important additions**	
Dr Maggie Harding (personal comments) London SCG Email of 17th Oct. 2008	4.4. Management of severe complications should be centralised & specialised: it is unlikely (hopefully) that these will ever be sufficiently common to justify 5 London centres and so default to sector commissioning, but we need better data.	

4. Detailed Description of Specialised Activity		
Contributor & source	**Comment**	**Action required & by whom**
Professor Irene Roberts, Imperial College London Email of 02nd October 2008	**4.4Management of severe and life-threatening acute complications of SCD and Thalassaemia** **Acute complications in Thalassaemia Major** include:- • heart failure and cardiac arrhythmias • post-splenectomy sepsis • iron chelator therapy-associated sepsis • acute endocrine disturbances (eg hypocalcaemic tetany) • acute hepatic decompensation. Acute complications are uncommon in children with thalassaemia, but need to be recognized and treated urgently usually within the specialist setting or in close liaison with the centre. Adult patients, are more likely to develop acute complications and management of pregnancy is particularly challenging and requires expert care. These patients are iron loaded, may have impaired immunity and impaired cardiac and endocrine function, potentially leading to decompensation during intercurrent illness and a rapidly progressive and fatal outcome. The treatment of heart failure in thalassaemia is unique to this condition. General physicians will be unfamiliar with the spectrum of acute infections emphasizing the need for specialist involvement. **Comment: I would suggest this is misleadingly reassuring unless qualified- suggestions shown.** **Uncomplicated SCD crises** ***All local hospitals*** will manage the more severe SCD crises requiring expert hospital management with strong opiate analgesia, good hydration and occasionally antibiotics and transfusion (this does NOT reflect our experience in children and could be seriously misleading if widely practiced by inexperienced clinicians in local hospitals with little experience of sickle cell disease). It also implies that antibiotics are used as infrequently as transfusion which is clearly inappropriate. Adverse effects of opiates are common and patients need to be monitored closely to ensure sustained effective analgesia and minimization of side effects until the pain settles and the opiate analgesia can be withdrawn. Prompt, effective and safe management of simple acute painful episodes aims to avoid or minimise hospital admissions. Such treatment will be provided in every local hospital. Specialist centres may support local hospitals in developing protocols for pain management in emergency departments and in-patient environments, as well as providing training for local hospital staff. **Comment: Antibiotics- Of course it is sensible not to treat every single admission with antibiotics. But**	

	many children have infection as the trigger or coinciding pathology; infection causes many deaths in sickle cell disease. I feel extremely strongly that this is not a safe emphasis and could be seriously misleading if widely practiced by inexperienced clinicians in local hospitals with little experience of sickle cell disease) - paradoxically this will be safer in larger centres who will already be aware of the issues. I THINK THIS SHOULD BE AMENDED TO WITH ANTIBIOTICS AS INDICATED AND OCCASIONALLY TRANSFUSION. The commonest acute complication is the painful SCD crisis which is generally ***managed at home*** with support from family and other regular carers and community services input as appropriate. These crises result in time lost from school and work and may have severe psychosocial effects on the whole household. **Comment: Such effects are not inevitable and may would be more appropriate here.**	

4. Detailed Description of Specialised Activity		
Contributor & source	**Comment**	**Action required & by whom**
Dr Jenny Welch, Sheffield Children's Hospital Email of 15th October 2008	**4.4Management of severe and life-threatening acute complications of SCD and Thalassaemia** Comments have been requested on the data sent out with this document. I feel it is extremely difficult to draw any conclusions from this as it is so 'broad' and so many more than the set 55 organisations may list a haemoglobinopathy as a diagnosis where actually it was not relevant to that episode (witness the many traits, who could only possibly be legitimately counted if attending to be given a result or for antenatal reasons). The data does not adequately separate out the sickle cell with crisis data - some crises can be managed at home, some need ICU which should count as a specialised service in SCD. Similarly 'exchange transfusion' covers a multitude of clinical situations. I would strongly advocate that emergency exchange transfusion in paediatric sickle cell disease should be a specialised service.	Note – this comment is repeated in Section 5 feedback.
Dr Shah, Whittington Hospital Email of 27th Oct. 2008 sent via Roma Haigh	**Acute complications in Thalassaemia Major** include:- • heart failure and cardiac arrhythmias • post-splenectomy sepsis • iron chelator therapy-associated sepsis • acute endocrine disturbances (eg hypocalcaemic tetany) (**Comment** - Diabetes associated complications such as hyperosmolar or ketoacidosis should be added here) • acute hepatic decompensation.	

4. Detailed Description of Specialised Activity		
Contributor & source	**Comment**	**Action required & by whom**
Professor Irene Roberts, Imperial College London Email of 02nd October 2008	**4.5Management of chronic complications of SCD and Thalassaemia** Regular monitoring will detect early evidence of complications which can occur during childhood, adolescence and adulthood, and tend to accumulate with age, resulting in significant disability and reduced life expectancy. There is a weak evidence base for the management of chronic complications. Prevention and treatment requires specialist knowledge and experience, close collaboration with other specialties (usually through regular joint clinics) and monitoring protocols agreed between the specialist centre and the local hospitals which can be implemented throughout the network. **Comment: Why is this stated? (I cannot see how it can possibly be in the patient's best interest to state this as it is an invitation to reduce services!) What about stroke? Evidence may not be strong but that is often because people don't look!** Most of these complications are due to chronic iron overload and can be reduced by sustained effective chelation, although bone disease has a complex and poorly understood aetiology. **Comment: Suggest this is amended to more accurately reflect the real outcome.**	
Dr Maggie Harding (personal comments) London SCG Email of 17th Oct. 2008	4.5. is unspecified: I do not consider this activity specialised, though the activity is unstated & as noted evidence of benefit scanty.	
Dr Shah, Whittington Hospital Email of 27th Oct. 2008 sent via Roma Haigh	**Chronic complications in Thalassaemia Major** include:- • endocrine dysfunction (growth hormone deficiency, hypogonadotrophic hypogonadism, hypothyroidism, hypoparathyroidism, Type 2 diabetes) (**Comment** - insulin dependant diabetes type 2 is misleading and makes the diabetes seem less problematic. The commonest form of diabetes in thalassaemia patients is insulin dependant diabetes.) • cardiac dysfunction • chronic liver disease (cirrhosis, portal hypertension, hepatic failure, hepatocellular carcinoma, often associated with transfusion-transmitted hepatitis B or C) • bone problems (avascular necrosis, osteoporotic fractures of the hips and spine, disc disease).	

4. Detailed Description of Specialised Activity		
Contributor & source	**Comment**	**Action required & by whom**
Dr Maggie Harding (personal comments) London SCG Email of 17th Oct. 2008	**4.6 Surgical management of SCD and Thalassaemia** 4.6. I agree surgical management – or more accurately anaesthetic and post –operative care is specialised: the surgery described is routine & non-specialised.	
Dr Maggie Harding (personal comments) London SCG Email of 17th Oct. 2008	**4.7 Management of pregnancy in SCD and thalassaemia** 4.7. Very little ante natal & intrapartum care is included within the SSNDS and so inclusion for maternal haemoglobin disorder needs to be carefully considered. The risk to mother & infant is probably equally high for other categories of chronic disease and this of itself does not (so far as I am aware) mandate inclusion for severe GUCH; CF etc. Though not specialised, emphasis could usefully be put on local hospital services for pre-conceptual counselling, early risk assessment of an affected fetus & informed choice on whether to continue the pregnancy. This is far more relevant to most people with a haemoglobin disorder than recommending PIGD to generate a saviour sibling. The latter, though employing some specialised services, is and is likely to remain an individual PCT commissioning decision.	
South Central SCG Email of 21st Oct. 2008 from Sally Nelson	**Pregnancy:** We have concerns regarding the inclusion of pre-implantation genetic diagnosis and selection of an HLA matched 'saviour sibling' within this definition. This is a highly contentious area, and we are not persuaded that the case has adequately been demonstrated, nor the issues properly considered either ethically or in terms of cost-effectiveness.	
Dr Shah, Whittington Hospital Email of 27th Oct. 2008 sent via Roma Haigh	**4.7 Management of pregnancy in SCD and thalassaemia** Each year in the UK 40-50 women with SCD will become pregnant and 4-5 women with thalassaemia major. These are high risk pregnancies which ideally will be managed in a joint high-risk antenatal clinic by the specialist in SCD and thalassaemia together with an obstetrician who has particular expertise in this area. Decisions about treatment during pregnancy and mode of delivery are usually made on an individual basis. There is significant maternal mortality in both conditions, with one maternal death in the UK every one or two years. (**Comment -** really that frequent! Reference? Sickle I can understand but with the Thals I feel that that is not the case, please see CEMACH Reports 200-2002 and 2003-2005 neither uclh or whittington have seen one since mri cardiac iron assessment became more widespread.)	

4. Detailed Description of Specialised Activity		
Contributor & source	**Comment**	**Action required & by whom**
Dr Shah, Whittington Hospital Email of 27th Oct. 2008 sent via Roma Haigh	**4.7 Management of pregnancy in SCD and thalassaemia** Thalassaemic women are often (**Comment** - Page: 23 May be hypogonadal, younger cohort tend not to be, older cohort mostly are.) hypogonadic and require fertility treatment in order to conceive. The risks for the pregnancy depend largely on the state of iron overload. Transfusion and chelation treatment will need to be modified during the pregnancy to avoid complications for the mother and the fetus. Fetal outcomes for women with thalassaemia major are usually good.	

4. Detailed Description of Specialised Activity		
Contributor & source	**Comment**	**Action required & by whom**
South Central SCG Email of 21st Oct. 2008 from Sally Nelson	**4.8 Stem cell transplantation for SCD and thalassaemia** **Stem cell transplantation:** This is a specialised service in its own right.	
Dr Maggie Harding (personal comments) London SCG Email of 17th Oct. 2008	4.8. Stem cell transplantation is included within the SSNDS.	

4. Detailed Description of Specialised Activity		
Contributor & source	**Comment**	**Action required & by whom**
Professor Irene Roberts, Imperial College London Email of 02nd October 2008	**4.9 Out-patient review of SCD and Thalassaemia** **Routine monitoring.** Routine outpatient monitoring is required every 3 months for thalassaemia and every 3-6 months for SCD depending on age and complications. In the case of thalassaemia, this is for the monitoring of haemoglobin (Hb) level, adjusting the transfusion regime and optimising iron chelation therapy through assessment of adherence, efficacy and adverse effects. In the case of SCD this is to monitor symptoms, the frequency of crises and adherence to medication and for parental education and advice on life style and crisis avoidance and monitoring of chronic disease (eg chronic hypoxaemia, hypersplenism/splenomegaly, abnormal TCD studies, difficulties at school). These 3-6 monthly monitoring visits are ***not*** considered a specialised service. However, if the SCD patient is being treated with hydroxyurea or transfusion, this requires monitoring at least every 4 weeks and adjustment as necessary and should be managed with more frequent specialist centre visits (eg alternating with local hospital). Thalassaemia intermedia should be managed mainly in specialist centres. **Comment: The wording here was misleading and incomplete. I hae suggested amendments which reflect what I believe should be done in a good centre to minimise chronic problems and pass children on to the adult service able to cope better with their disease and make better progress after leaving school.**	
Professor Barbara Bain, Imperial College London Email of 07th October 2008	**4.9 Out-patient review of SCD and Thalassaemia** **Routine monitoring.** Routine outpatient monitoring is required every 3 months for thalassaemia and every 3-6 months for SCD depending on age and complications. In the case of thalassaemia, this is for the monitoring of haemoglobin (Hb) level, adjusting the transfusion regime and optimising iron chelation therapy through assessment of adherence, efficacy and adverse effects. In the case of SCD this is to monitor symptoms, the frequency of crises and adherence to medication and for parental education and advice on life style and crisis avoidance. If the SCD patient is being treated with **hydroxycarbamide (Comment - The International Recommended Non-Proprietary name should surely be used – see BNF)** or transfusion, this also requires monitoring and adjustment as necessary. These 3-6 monthly monitoring visits are ***not*** considered a specialised service.	
Dr Maggie Harding (personal comments) London SCG Email of 17th Oct. 2008	**4.9 Out-patient review of SCD and Thalassaemia** 4.9. details the activity missing in 4.5 so is to some extent duplicative: as before it is not specialised activity: if depending on caseload there needs to be oversight from specialised service this (& the case load threshold at which this might operate) should be specified.	

4. Detailed Description of Specialised Activity		
Contributor & source	**Comment**	**Action required & by whom**
South Central SCG Email of 21st Oct. 2008 from Sally Nelson	**4.9 Out-patient review of SCD and Thalassaemia** **Out-patient review:** We do not support the inclusion of out-patient activity within specialised service definitions, because it cannot be validated, may include general haematology and non-specialised activity, and would not normally incur additional costs compared with non-specialised out-patient activity.	
Dr Shah, Whittington Hospital Email of 27th Oct. 2008 sent via Roma Haigh	The specific information needed to inform the annual review will depend on the patient concerned and may include reports from other specialties, including neurology (SCD), cardio-respiratory (SCD), musculoskeletal (SCD and thalassaemia) and gastrointestinal (SCD and **thalassaemia**). (**Comment** - ? Endocrine review for diabetes , osteoporosis, other endocrinopathies for thalassaemia and not mentioned annual cardiac assessment or cardiology review for the TMs also)	

5. Identifying and Costing Specialised Activity		
Contributor & source	**Comment**	**Action required & by whom**
Dr Jenny Welch, Sheffield Children's Hospital Email of 15th October 2008	Comments have been requested on the data sent out with this document. I feel it is extremely difficult to draw any conclusions from this as it is so 'broad' and so many more than the set 55 organisations may list a haemoglobinopathy as a diagnosis where actually it was not relevant to that episode (witness the many traits, who could only possibly be legitimately counted if attending to be given a result or for antenatal reasons). The data does not adequately separate out the sickle cell with crisis data - some crises can be managed at home, some need ICU which should count as a specialised service in SCD. Similarly 'exchange transfusion' covers a multitude of clinical situations. I would strongly advocate that emergency exchange transfusion in paediatric sickle cell disease should be a specialised service.	
South East Coast SCG & local providers Email of 22nd Oct. 2008 via Nick Haslem	It is suggested that determining what is specialised and what is secondary care on the basis of 50 organisations providing that care is problematic for haemoglobinopathies, because their prevalence is low. Therefore the 'cut off' number for the organisations needs to be much lower. One approach would be to describe the services of a centre and the critical mass for centres to be viable. Decide on the number of centres and deem that service to be specialised.	
Dr Marie Donohue, Nottingham University Hospital Email of 22nd Oct. 2008	Thank you for the opportunity to respond to this proposal which will be welcomed by haemoglobinopathy clinics . There are a few points I will try and respond to 1) in my clinic haematology attendance tariff would not cover haemoglobinopathy visit as these appointments are 30 mins each , providining , I hope, a more holistic service 2) opcs CODE - I think that erythrocytopheresis is specialised and high cost drugs of thiose you gave us but there are ommissions- TCD, surgery MRA etc 3) in terms of ICD codes they are a mixed bag but traits should not be specialised - I think most of the others come into may be specialised depending on the problem at the time. IN general looking after patients with major haemoglobinopathy should be specialised with some aspects of care devolved	
South Central SCG Email of 21st Oct. 2008 from Sally Nelson	**Consultation question:** We believe that haemoglobinopathy outpatient activity can appropriately be both coded within, and commissioned as general haematology outpatients. We can see no added value in commissioning it separately as a specialised service.	

5. Identifying and Costing Specialised Activity		
Contributor & source	**Comment**	**Action required & by whom**
Dr Allison Streetly, NHS Sickle Cell & Thalassaemia Screening Programme Email of 24th Oct. 2008 via Roma Haigh	***Expensive drug costs*** – we understand that it is proposed to include two iron chelators on the DH list of high cost drugs excluded from tariff (deferasirox and deferiprone) . To be consistent , we suggest that the third iron chelating drug - desferrioxamine. - mentioned in the definition should also be included on the exclusion list. We also urge the DH to confirm this proposal as soon as possible to reduce any uncertainties between providers and PCT commissioners. ***Hydroxurea*** is a drug currently prescribed off-licence (and therefore cheaply) for sickle cell disease in some more severe cases and this drug is likely to be licensed shortly specifically for use in children with sickle cell disease. This should be flagged up in the definition alongside the iron chelators as it will shortly be costing the NHS considerably more.	Note – this section repeated from Section 4.1 as feedback relevant to both sections.
Dr Claire Hemmaway, Barking, Havering and Redbridge Hospitals Email of 20th Oct. 2008 via Brenda Allen of EoE SCG	I have looked at these codes and below are my thoughts: Thalassaemia needs to be divided into_ Beta thalassaemia major – life long transfusion dependence 4/52ly/ need for iron chelation/ 3/12ly reviews in outpatients/ cardiology/ endocrine etc input Beta thalassaemia trait which has no clinical significance unless pregnant and the partner has the same condition/ potential for Beta thal major in the child Haemoglobin H disease (alpha thalassaemia) – review in outpatients only usually Alpha thalassaemia trait – no clinical significance unless pregnant and partner also has alpha 0 thalassaemia trait, potential for child to have Bart's hydrops fetalis Delta-beta thalassaemia – possible transfusion dependence/ regular outpatient follow up required Delta-beta thalassemia trait – no clinical significance unless pregnant – see above Thalassaemia, unspecified Without a specific code for beta thalassaemia major we will be missing the blood transfusions that these adults/children have every month, their very expensive iron chelation therapy and outpatient appointments. The rest have much less/ nil activity	

6. National Standards, Guidelines and Protocols		
Contributor & source	**Comment**	**Action required & by whom**
	No specific comments received on this section – but see comments in other sections.	

Appendix 1		
Contributor & source	**Comment**	**Action required & by whom**
Professor Irene Roberts, Imperial College London Email of 02nd October 2008	**Appendix 1: Levels of care for Sickle Cell Disease and Thalassaemia** Some of these functions may be undertaken by specialist haemoglobinopathy nurses. **Comment: Sugested amendment as all of these functions cannot be carried out by specialist nurses.**	
Professor Barbara Bain, Imperial College London Email of 07th October 2008	**Local hospital care includes:** • Management of acute, uncomplicated crises (SCD) • Routine monthly day case transfusions (thalassaemia major and transfusion-requiring SCD) • Routine out-patient monitoring (SCD and thalassaemia) • Agreed shared care arrangements for specific therapies (SCD and thalassaemia) (including support with adherence to iron chelation regimes, monitoring of **hydroxycarbamide, care following** stem cell transplantation).	
Dr Maggie Harding (personal comments) London SCG Email of 17th Oct. 2008	Appendix 1: the definition of specialist centre is just that; I do not consider all the activity specified is specialised. Specifically I would exclude as non-specialised:- • Management of chronic complications (SCD and thalassaemia) • Annual out-patient review (SCD and thalassaemia) • Outreach clinics in local hospitals (SCD and thalassaemia). And management of pregnancy (SCD and thalassaemia) needs more careful consideration in the light of risk in other non-SSNDS conditions Although entirely non-specialised, I would expect the role of community haemoglobin disorder services to include explicitly under self management: prevention, early recognition and home management of painful crises, including appropriate triggers for seeking hospital care.	

Other outstanding issues regarding Definition 38.		
Contributor & source	**Comment**	**Action required & by whom**
Dr Allison Streetly, NHS Sickle Cell & Thalassaemia Screening Programme Email of 24th Oct. 2008 via Roma Haigh	***Prenatal Diagnostic testing (and potentially in future Free foetal DNA)***. There are problems with this test as it lies between the molecular genetics definition, a screening definition (not yet developed) and mainstream tariff costs. Leadership from specialist commissioning is needed urgently as we are now facing medical geneticists advising that tests be sent outside the UK due to their high cost in this country. If tests are done outside the UK, it will mean that the screening programme looses a "whole picture understanding" of what is happening to PND testing and the ability to ensure quality. We remain concerned that the approach currently adopted of dealing with PND testing as part of the tariff is not allowing the market to work – prices are too high which results in continuing "noise" in the system. We will continue to raise our concerns on this issue until an appropriate resolution is reached. We also suggest that cross referencing is included in this definition to the need for these services and that they should be covered in a future screening definition or within the medical genetics definition (especially relevant with the advent of FFDNA testing if/when that becomes available). ***Pre-implantation Genetic Diagnosis*** – cross reference to this is needed. Currently it appears to be a problem with access to this service, especially compared to cystic fibrosis, with most referrals coming through infertility treatment routes rather than a specific request for PIGD. With continuing establishment of our Screening Programme we expect more couples to be aware of their risk status and request PIGD initially after the birth of an affected child but increasingly prospectively too. Linkage of commissioning oversight between "specialist care" and "specialist aspects of screening" is essential. Services should be commissioned in a consistent manner – i.e. should not proceed without taking into account where the specialist aspects of screening are provided and where the screening expertise is to ensure that diagnostic and screening expertise is concentrated or well linked together. This requires a joined up process with the commissioning of the specialist aspects of care as both should be concentrated in the same places/ or with some oversight and linkage. This approach should be covered in the definition. There is no mention anywhere in the definition of quality assessment or, by implication, designation. We recommend that there must be something in the definition on quality assurance	

	and assessment. As a minimum there should be reference to the published standards listed and the need for some form of process – such as the proposed peer review process - to be developed. Without this the achievement of the definition and the assurance of quality will be very difficult to assess. We also recommend that there should be a mention of the need for cross linkage with the development of Quality Assurance processes for screening which is currently underway. Visits to sites would benefit from some co-ordination with the assessment of the specialist centres and their networks so that the issues identified are seen in a more co-ordinated way. Particularly relevant to this definition is the work by the Screening Programme on the development of standards and quality assurance processes for the introduction of TCD scanning for children with sickle cell disease . Please refer to the NHS Sickle Cell & Thalassaemia Screening Programme Standards for the Linked Antenatal and Newborn Screening Programme : http://www.sct.screening.nhs.uk/AimsObject.htm#ProgStandards - The draft standards for TCD scanning for children with sickle cell can be found at : http://www.sct.screening.nhs.uk/Documents/TCDStandards.pdf *The final draft of this will be available in the spring of 2009.* We note the recent document on paediatric co-dependencies produced by Dr Ted Baker and ask that the relevance of this to specialist aspects of haemoglobinopathies be registered especially in relation to complex procedures such as Bone Marow Transplants and complex neurological problems. The NCEPOD report and concerns identified by this report should also be referenced in the documents relevant to commissioners. The HTA Exjade document should also be referenced – the current draft is on our website at http://sct.screening.nhs.uk/publications.htm#exjade	

End of Haemoglobinopathy feedback