Birmingham Children's Hospital

NHS Foundation Trust

Paediatric Laboratory Medicine
Clinical Chemistry Department

WEST MIDLANDS
NEWBORN SCREENING CENTRE
ANNUAL REPORT
2009-2010

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The Newborn Screening Laboratory is a CPA Accredited Laboratory
as part of the Clinical Chemistry Department

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1 Introduction

The West Midlands Newborn Screening Centre is part of the Paediatric Clinical Chemistry Department at the Birmingham Children's Hospital NHS Trust (BCH). It was formed from the merger of two separate West Midlands screening services, Birmingham and Hollymoor, in 1993 and provides a service for newborn screening for the West Midlands Strategic Health Authority. Babies are tested for phenylketonuria (PKU), congenital hypothyroidism (CHT), sickle cell disorders (SCD), cystic fibrosis (CF) and medium chain acyl Co-A dehydrogenase deficiency (MCADD). A diagnostic and follow up service for patients with PKU and MCADD is provided by the clinical inherited metabolic disorders services (IMD) at BCH.

The screening team comprises clinical scientists, biomedical scientists, administrative and clerical staff and clinical nurse specialists with input from the West Midlands Child Health Screening Coordinator. The team members work with other health professionals as part of multidisciplinary working across the whole programme.

History of newborn screening in the West Midlands

1969: Universal newborn screening for PKU using heelprick blood began in the West Midlands and is now performed using automated tandem mass spectrometry.

1981: Universal newborn screening for CHT began and is now performed using a fully automated, fluorimetric immunoassay, autoDELFIA.

1978: Universal newborn screening for sickle cell disease began in Birmingham using cellulose acetate electrophoresis.

1996: Newborn screening for sickle cell disease extended to Coventry using isoelectric focussing.

2003/4: Newborn screening for sickle cell disease rolled out across the whole of the West Midlands with improved technology and two tier testing. The current technology is high pressure liquid chromatography (HPLC) as the first line screening test and isoelectric focussing as the second line test.

2004: From 1st March newborn screening for MCADD (tandem mass spectrometry) began as a 2 year pilot programme as part of a six centre UK collaborative programme. MCADD screening was made universal in England from April 2009.

2006: From 1st November universal newborn screening for cystic fibrosis commenced.

This report summarises the activity of the newborn screening service from 1 April 2009 to 31 March 2010.

2 Executive summary

Outcomes

- Total number babies screened — **71443 total**
  71026 (West Midlands residents)
- Total number repeat collections requested — **3071** (4.3%)
- Total diagnoses 2009-10 (2008-9)
  - Phenylketonuria (PKU) — **4** (6)
  - Congenital Hypothyroidism (CHT) — **43**(60)
  - Sickle Cell Disorders (SCD) S, SC,S-other disorders — **27**(17)
  - Medium Chain Acyl CoA Dehydrogenase Deficiency (MCADD) — **4**(3)
  - Cystic fibrosis (CF) — **22**(16)

Performance against national standards (new standards published in August 2008)

- **Standard 1:** Completeness of offer. Coverage data is provided by Child Health. The total number of declines notified to the laboratory is provided.
- **Standard 2:** The laboratory did not routinely record the presence of the NHS number, therefore a snapshot estimate from 3 days at the beginning, middle and end of the financial year has been used.
- **Standard 3**: 15 of the 21 Child Health districts in the West Midlands met the core Standard for timely sample collection.
- **Standard 4**: Samples do not reach the laboratory in a timely manner for some PCTs.
- **Standard 5:** Quality of blood spot sample – The avoidable repeat rate is 3.5%, target ≤2.0%.
- **Standard 6**: Timely receipt of a repeat/second blood spot sample - unable to record for this year's report.
- **Standard 7:** Timely processing of positives for PKU – 100% compliance with core standard
  Timely processing of positives for CHT – 100 % of cases met the core standard
  Timely processing of positives for CF – 78 % of cases met the core standard
  Timely processing of positives for MCADD – 100 % of cases met the core standard
- **Standard 7:** Clinical referral for PKU – 100 % compliance with core standard
  Clinical referral for CHT –79 % compliance with core standard
  Clinical referral for CF – 89 % compliance for babies with 2 mutations
  Clinical referral for MCADD – 100 % compliance with core standard
- **Standard 8**: This data is to be provided by the Child Health Record Departments
- **Standard 9**: This data is to be provided by the Child Health Record Departments

Teaching and Education
Regular teaching sessions for West Midland's health professionals in practical aspects of newborn screening have continued.

Future Plans

- Development of pterin measurement in dried blood spots by HPLC
- To continue developing the research project evaluating methodology to screen for tyrosinaemia type 1.
- The department is actively involved in national discussions about extending the screening repertoire using tandem mass spectrometry.

3 Staffing

Clinical Scientists

Paul Griffiths, BSc, DipCB, FRCPath, MBA
Director of Newborn Screening and Consultant Clinical Biochemist
Kate Hall, BSc, MSc FRCPath
Head of Newborn Screening Section and Principal Biochemist
Pippa Goddard, BSc, PhD, FRCPath
Principal Clinical Biochemist
Sarah Dowden BSc, MSc
Senior Clinical Biochemist

Head MLSO/Laboratory Manager

Glynis Kane

Biomedical Scientists

Russell Denmeade	Chief BMS
Claire Murray, BSc, AIBMS	Senior BMS
Deb Finnerty, BSc, AIBMS	Senior BMS
Mohammed Asman*	
Karen Sandhu*	
Sandra Zurawan-Valley*	
Anna Lygas*	
Angela Atkinson*	
Chris Johnston*	

Medical Laboratory Assistants

Sharon Jackson
Deb Johnson
Rosy Panesar

Clinical Nurse Specialists**

Rachel Gould, RN-Child (commenced Feb 2010)
Louise Simmons, RN-Child
Elaine Salmons, RGN, RSCN, BSc Hons Advanced Nursing Practice

* Rotational posts
** Shared posts with Inherited Metabolic Disease and Paediatric Clinical Chemistry section

Secretarial/Clerical

Sheila Preston (retired December 2009)
Bernadette Cunningham
Bernadette Vincent

Establishment in the section (excluding shared posts) was as follows:-

- Principal Biochemists (Clinical Scientists) 1.4 wte
- Senior Biochemist 0.33 wte
- Chief Biomedical Scientist 0.5 wte
- Senior Biomedical Scientists 2.0 wte
- Basic Grade/Trainee Biomedical Scientists 1.0 wte (Rotational post)
- Medical Laboratory Assistants 1.5 wte
- Secretary 0.81 wte
- Clerical officers 1.08 wte
- Clinical Nurse Specialists 1.8 wte

4 Services Provided

A. Newborn screening for phenylketonuria (PKU), congenital hypothyroidism (CHT), sickle cell disorders (SCD), cystic fibrosis (CF) and MCADD on all babies resident in the West Midlands.

B. A national screening service for biopterin metabolism defects for the diagnosis of non-classical PKU variants.

C. Quantitation of bloodspot phenylalanine by tandem mass spectrometry for confirmation of the diagnosis of PKU and monitoring of dietary therapy.

D. Diagnostic follow up for other metabolic disorders, detected by screening (e.g. galactosaemia, tyrosinaemia). There is a seamless link between the newborn screening and the diagnostic follow up metabolic services.

E. Consultant/Advisory service for the investigation, diagnosis, treatment and biochemical management of patients detected by the screening programme.

F. Clinical Nurse Specialist support. New cases diagnosed from the screening programme and requiring recall to see a consultant metabolic paediatrician are supported by clinical nurse specialists. PKU patients on treatment and older females of child bearing age receive on-going support.

G. Teaching and Education. The laboratory provides teaching sessions and education material in the theory and practice of newborn screening for health professionals.

H. Performance Management and Audit. The quality and effectiveness of the service is monitored by the provision of detailed performance statistical data as part of this annual report and via clinical audit multidisciplinary groups.

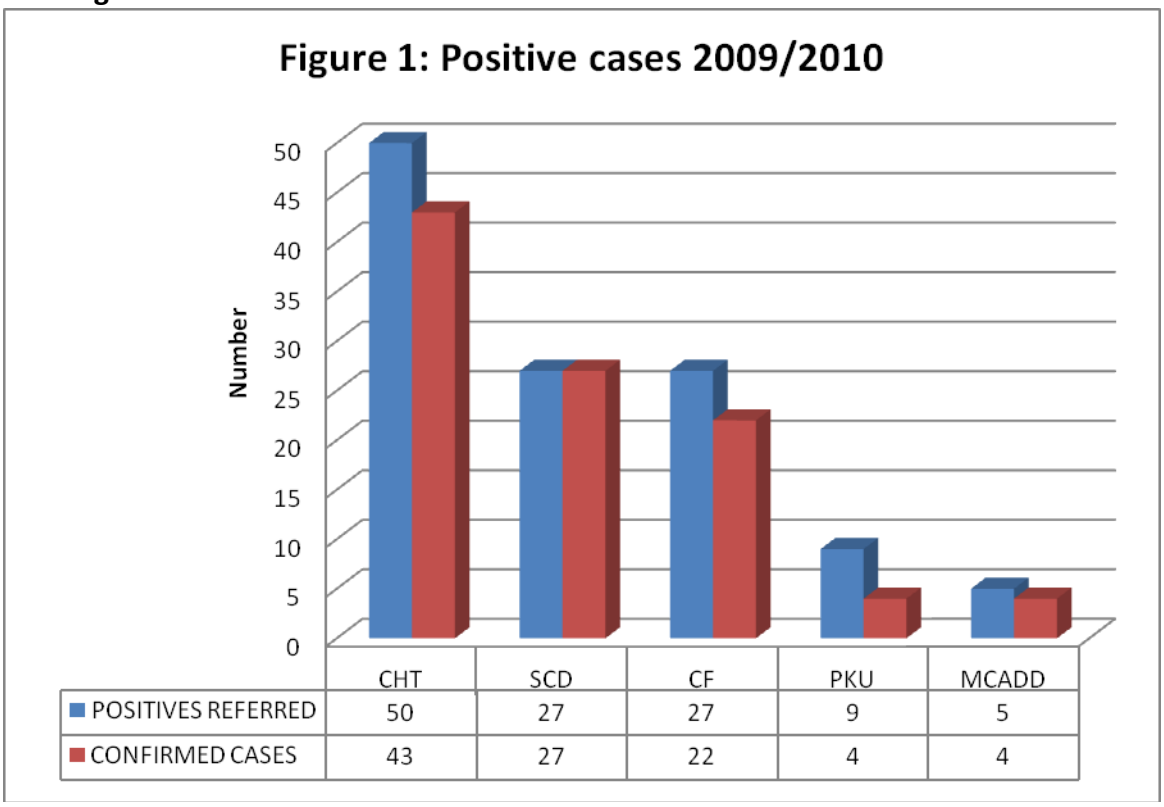
I. Research and Development infrastructure for new programmes and projects.

5 Activity

5.1 Laboratory Workload

The laboratory's workload is very similar to last year's. See Table A on page 12.

5.2 Diagnoses



In addition to the 27 cases of sickle cell disorders (FS, FSC, FS-other), **13** other haemoglobinopathies were detected by newborn screening, therefore the total referred from sickle screening = 40.

Cumulative birth prevalence
Calculated birth prevalence figures PKU, CHT, SCD, MCADD and CF are shown in Table F, p 14.

Comments

CHT The number of cases of CHT has decreased compared to last year's record number, whilst remaining significantly higher than all other preceding years. The reason for this increase is not known. The cases that were not confirmed were transient conditions.

SCD The overall birth prevalence for 6 years of screening the whole of the West Midlands is 1 in 3787 babies, less than seen in the London area.

MCADD 1 of the 5 presumptive positive cases referred was a false positive. The baby was unwell (substantial weight loss/not breast feeding appropriately) at the time the blood sample was taken.

CF The overall birth prevalence is 1 in 3644 babies, remaining less than an expected ~1 in 2,500. It may take several more years of screening before a meaningful prevalence can be calculated.

5.3 Other Diagnoses

Table A: Other disorders detected by PKU screen

Child Health District Code	PCT	Disorder
N	Birmingham East and North	Tyrosinaemia type 1
W	Sandwell	Galactosaemia
G	North Staffordshire	Raised tyrosine due to liver problems not due to galactosaemia or tyrosinaeimia
E	Shropshire	Initial raised tyrosine which decreased on repeat – no further follow-up required

6 Enquiries

The laboratory has specific arrangements with some Child Health Records departments to follow up testing for new immigrants who have not been screened.

6.1 Immigrants

Immigrant children to Birmingham without newborn screening test results are identified by Birmingham Child Health checking mechanisms and notified to the Screening Administrative Office. Parents of children under 12 months of age were written to with a parent information leaflet enclosed, offering them newborn screening and an appointment at the hospital for a blood spot specimen to be collected by the Clinical Nurse Specialist. Of the **107** we were notified of, only **24** took up the offer of screening for their baby.

Outcome	Number
TOTAL	107
Did not attend appointment for blood collection	73
Had already been tested elsewhere	4
Over 1 year old before testing could be arranged	6
Took up neonatal screening	24

6.2 Adoptions

In connection with adoption medicals, we provided newborn screening test results to the following enquiries:-

Enquirer	Number of enquiries
Coventry	29
South Birmingham	5

7 Clinical Nurse Specialists

The Clinical Nurse Specialists provide liaison for patients across the West Midlands with other health professionals, families and many agencies. Their roles involve teaching and training, clinic support, home visits (see Table F, p16) and visits to schools and other hospitals. The work undertaken includes:-

- Screening programme-including immigrants/refusals
- PKU treatment/management all aspects
- PKU pregnancy/counselling
- CHT all aspects
- CF
- SCD
- MCADD
- Other metabolic disorders arising from the programme including pre-natal diagnosis counselling and support (see IMD report)

8 Other Investigations

Total biopterin and DHPR

These tests are required for the follow up of positive PKU screening tests to exclude disorders of biopterin metabolism and are provided for the UK and Republic of Ireland. This workload is recorded in the IMD Annual Report.

Plasma and dried blood spot phenylalanine

The annual workload for quantitative phenylalanines for diagnostic follow up and treatment is recorded in the IMD Annual Report.

9 Other Activities

The laboratory distributes screening laboratory address labels; 'screening tests for your baby' booklets are available on request but should normally be provided as pre-screening leaflets ordered from Prolog. These leaflets are available in other languages via the UKNSPC website (http://newbornbloodspot.screening.nhs.uk/languages).

10 Denominator Populations

The following groupings have been used for the data analysis based on the residence address of babies screened.

PCT	Code	Old Child Health District	Associated main Maternity Hospital	Babies tested
Worcestershire	A	North Worcestershire	Alexandra	1928
	C	Kidderminster	Kidderminster (community midwifery, no deliveries)	1110
	D	Worcestershire	Worcester Royal	3127
Herefordshire	B	Hereford	Hereford County	1793
Telford and Wrekin **Shropshire County**	E	Shropshire	Shrewsbury	5089
South Staffordshire	F	Mid.Staffordshire	Stafford	3208
	H	S.E. Staffordshire	Queen's Burton	3396
North Staffordshire & Stoke on Trent	G	North Staffs	North Staffs	5783
Warwickshire	J	N.E. Warwickshire (Rugby)	Walsgrave	1070
	K	North Warwickshire	George Eliot Nuneaton	2124
	L	South Warwickshire	Warwick	2677
Birmingham East and North	N	East Birmingham	Heartlands	4278
	P	North Birmingham	Good Hope	2177
South Birmingham	R	South Birmingham	Birmingham Women's	6797
Heart of Birmingham Teaching	S	West Birmingham	City	3937
Coventry Teaching	T	Coventry	Walsgrave	4806
Dudley	U	Dudley	Russells Hall	3725
Sandwell	W	Sandwell	Sandwell	4581
Solihull	X	Solihull	Solihull	2129
Walsall Teaching	Y	Walsall	Walsall Manor	4013
Wolverhampton City	Z	Wolverhampton	New Cross	3357
Outside West Midlands	O	N/A		337

11 Workload Tables

11.1 Table B: Annual workload (2008-2009 in brackets)

NUMBER OF BABIES SCREENED	71443	(70336)
TOTAL NUMBER OF SPECIMENS	77451	(75335)
∴ NUMBER OF REPEATS *RECEIVED*	6008	(4999)
REPEATS AS % OF BABIES TESTED	8.4%	(7.1)

11.2 Table C: Overall reasons for repeats *requested*

Affecting all tests (e.g. insufficient, taken within 72hrs blood transfusion, too young, unsuitable) (excluded pre-transfused samples)	2692	(2913)
Affecting CHT Test only Pre-term Borderline result	1997 87	(gestation repeats were not organised by the laboratory at this time) (89)
Affecting SCD Test only Transfusion Too premature	274 1st specimen, 664 were repeat requests 5	(174)
Affecting CF Test only Inconclusive	41	(40)
Affecting PKU Test only Raised Tyrosine Borderline result	99 7	(NA) (NA)
TOTAL	5866	(3219)
REPEATS AS % OF BABIES TESTED	8.2%	(4.6%) (this figure does not include gestation repeats)

Note The repeat blood collections requested during the financial year do not match the number of repeats received as the time frames differ slightly, unrequested repeat (i.e. duplicate) specimens are received and due to computing limitations we are unable to identify some repeats taken early for SCD prior to a transfusion or gestation <36 weeks repeat

Table D: Reasons for repeats requested (avoidable)

PCT	Code	Insuff blood	within 72hrs of blood transfusion	*Too young	Unsuitable	Total repeats	Total number of cards	Avoidable repeat rate (%)
Worcestershire	A	24	2	15	16	57	2070	2.8
	C	5	2	2	11	20	1165	1.7
	D	31	3	7	55	96	3395	2.8
Herefordshire	B	17	2	12	32	63	1929	3.3
Telford and Wrekin Shropshire County	E	35	6	6	117	164	5453	3.0
South Staffordshire	F	78	1	12	71	162	3486	4.6
	H	49	5	10	46	110	3670	3.0
North Staffordshire & Stoke on Trent	G	52	13	7	148	220	6268	3.5
Warwickshire	J	5	3	5	20	33	1134	2.9
	K	49	5	13	35	102	2322	4.4
	L	22	4	10	37	73	2832	2.6
Birmingham East and North	N	84	9	14	125	232	4757	4.9
	P	36	8	10	38	92	2365	3.9
South Birmingham	R	87	10	21	102	220	7334	3.0
Heart of Birmingham (T)	S	72	7	22	37	138	4249	3.2
Coventry Teaching	T	84	15	26	102	227	5226	4.3
Dudley	U	80	8	15	47	150	4098	3.7
Sandwell	W	74	13	17	97	201	5034	4.0
Solihull	X	24	0	7	65	96	2295	4.2
Walsall Teaching	Y	21	9	20	45	95	4314	2.2
Wolverhampton City	Z	12	8	9	69	98	3638	2.7
Outside WM	O	6	19	4	14	43	417	10.3
Grand Total		**947**	**152**	**264**	**1329**	**2692**	**77451**	

- **Please note: Specimens collected < 4 days were excluded from the 'too young' repeats on the ground that they are most likely to be pre-transfusion collections.**

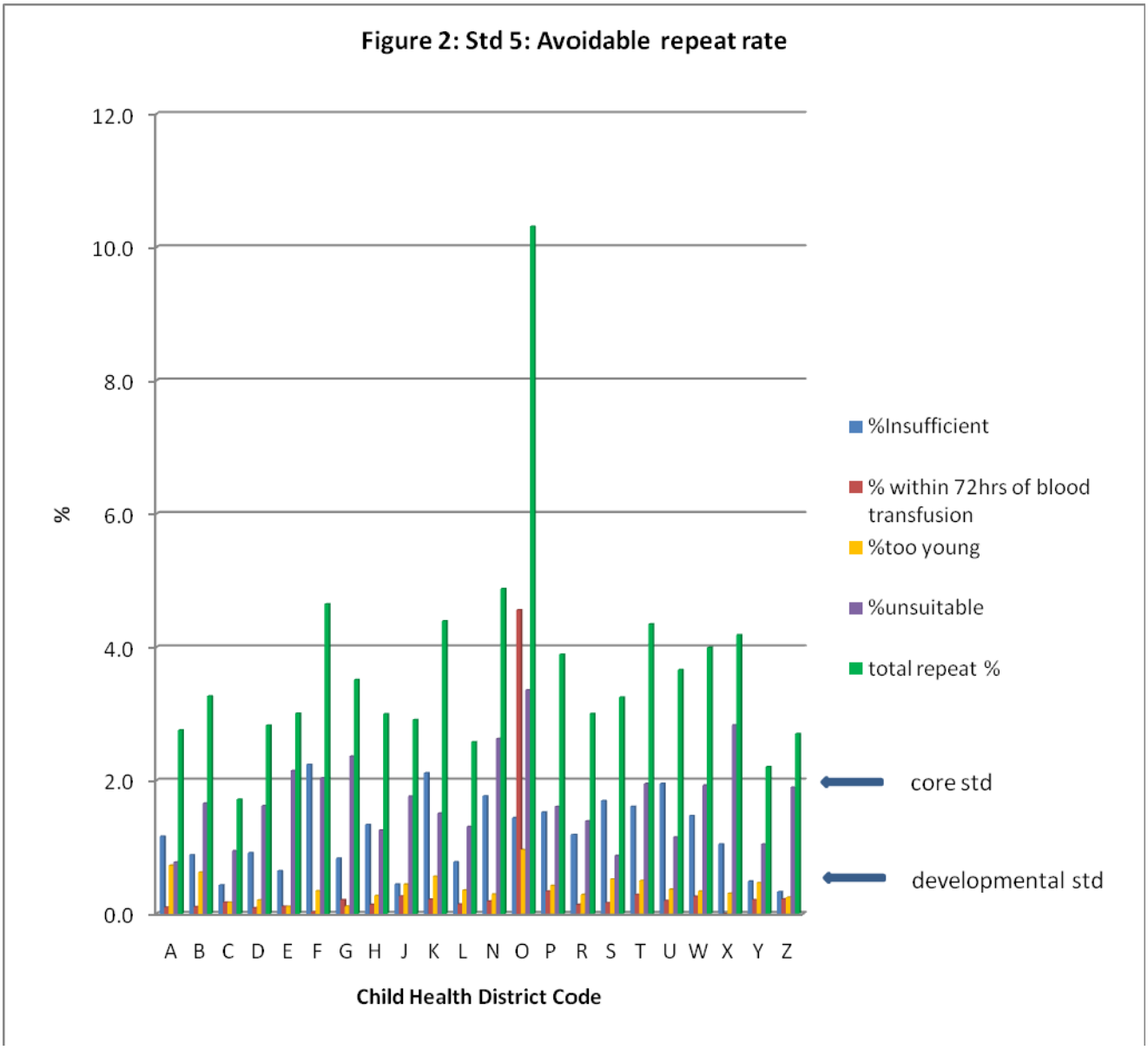


Figure 2: Std 5: Avoidable repeat rate
12.0
10.0
8.0
6.0
4.0
2.0
0.0
%
A B C D E F G H J K L N O P R S T U W X Y Z
Child Health District Code
%Insufficient
% within 72hrs of blood transfusion
%too young
%unsuitable
total repeat %
core std
developmental std

Table E: Reasons for repeats requested (unavoidable)

PCT	Code	PKU equivocal	(PKU) raised Tyr	CHT equivocal	CHT pre-term	CF inconclusive
Worcestershire	A	0	3	1	63	0
	C	0	3	0	27	1
	D	1	6	4	113	1
Herefordshire	B	0	1	3	46	2
Telford and Wrekin Shropshire County	E	0	8	7	136	3
South Staffordshire	F	0	3	2	65	1
	H	0	4	3	103	0
North Staffordshire & Stoke on Trent	G	2	8	4	158	1
Warwickshire	J	0	1	0	17	0
	K	0	3	5	29	3
	L	0	7	2	41	1
Birmingham East and North	N	0	5	14	124	2
	P	0	3	1	72	0
South Birmingham	R	1	8	10	161	8
Heart of Birmingham (T)	S	0	6	5	98	3
Coventry Teaching	T	0	6	2	118	4
Dudley	U	0	3	3	120	2
Sandwell	W	1	6	5	130	1
Solihull	X	0	1	2	41	0
Walsall Teaching	Y	2	5	9	154	3
Wolverhampton City	Z	0	7	4	101	3
Outside WM	O	0	2	1	80	2
Grand Total		**7**	**99**	**87**	**1997**	**41**

11.3 Cumulative Incidence

Table F: Cumulative Incidence

Disorder	Number tested	Diagnoses	Cumulative Incidence
PKU*	1811756	136	1 in 13322
CHT*	1811756	750	1 in 2416
SCD**	412821	109	1 in 3787
MCADD**	412821	39	1 in 10585
CF***	240521	67	1 in 3590

* West Midlands figures from January 1983 onwards

** West Midlands data from 1 April 2004

*** West Midlands data from 1 November 2006

11.4 Table G: Home Visits by Clinical Nurse Specialists

PCT	Code	PKU	CHT	SCD	MCADD	CF	Other
Worcestershire	A					1	
	C						
	D						
Herefordshire	B	1			1	1	
Telford and Wrekin Shropshire County	E				1	3	
South Staffordshire	F					1	
	H	1	1			2	1
North Staffordshire & Stoke on Trent	G				2	3	2
Warwickshire	J						
	K						
	L						
Birmingham East and North	N		8	6		1	
	P			2			
South Birmingham	R		4	2		1	
Heart of Birmingham (T)	S		1	11		1	2
Coventry Teaching	T	1		1		1	
Dudley	U						
Sandwell	W	2					
Solihull	X			1			
Walsall Teaching	Y	1		1			
Wolverhampton City	Z			5	1	6	
Total	**80**	**6**	**14**	**29**	**5**	**21**	**5**

1st visits to patients from newborn screening programme by CNSs '09-10'

12 Performance Measures

The measures used are the 9 key process standards and clinical referral standards developed by the UKNSPC (ref 1). The core standards set out the expected level of performance required to deliver an acceptable level of quality. The developmental standards are a level of performance which delivers enhanced quality. Where possible we have used the core standard to measure against.

Due to the limitations of our current laboratory information management system system (SIMS) there are some restrictions in how we can analyse data – see explanatory notes below.

Explanatory notes to the data

Data have been analysed according to the **residence of each baby**.

i) This annual report describes **specimens <u>received</u>** in the laboratory between 01/04/09 and 31/03/10. We are currently unable to sort work by date of birth.

ii) Sample transit time to the laboratory refers to **all samples** not just first samples (i.e. includes repeats).

iii) Days to referral for positive results is in **working days** which have been manually calculated.

12.1 STANDARD 1 - Completeness of offer

Total Declines Notified To Laboratory

PCT	Code	Associated main Maternity Hospital	1st Decline (number babies)	Babies remaining untested
Worcestershire	A	Alexandra	1	1
	C	Kidderminster (community midwifery, no deliveries)	0	0
	D	Worcester Royal	4	3
Herefordshire	B	Hereford County	3	2
Telford and Wrekin Shropshire County	E	Shrewsbury	1	1
South Staffordshire	F	Stafford	0	0
	H	Queen's Burton	1	1
North Staffordshire & Stoke on Trent	G	North Staffs	3	3
Warwickshire	J	Walsgrave	0	0
	K	George Eliot Nuneaton	0	0
	L	Warwick	0	0
Birmingham East and North	N	Heartlands	8	6
	P	Good Hope	0	0
South Birmingham	R	Birmingham Womens'	6	6
Heart of Birmingham Teaching	S	City	2	2
Coventry Teaching	T	Walsgrave	2	2
Dudley	U	Russells Hall	2	2
Sandwell	W	Sandwell	1	1
Solihull	X	Solihull	0	0
Walsall Teaching	Y	Walsall Manor	2	2
Wolverhampton City	Z	New Cross	1	1
		Total	**37**	**33**

12.2 STANDARD 2 – Enhanced tracking abilities

Core Standard

100% of blood spot cards received by a laboratory include the babies' NHS Number.

Below is a **snapshot estimate** from 3 days during the financial year.

PCT	Code	Associated main Maternity Hospital	Cards received over 3 days	*% with NHS number recorded	% with NHS number label
Worcestershire	A	Alexandra	25	100	60
	C	Kidderminster (community midwifery, no deliveries)	23	100	91
	D	Worcester Royal	54	91	81
Herefordshire	B	Hereford County	57	100	72
Telford and Wrekin Shropshire County	E	Shrewsbury	98	98	81
South Staffordshire	F	Stafford	34	97	53
	H	Queen's Burton	49	82	35
North Staffordshire & Stoke on Trent	G	North Staffs	104	93	0
Warwickshire	J	Walsgrave	26	96	<1
	K	George Eliot Nuneaton	63	90	0
	L	Warwick	56	95	54
Birmingham East and North	N	Heartlands	82	89	<1
	P	Good Hope	50	98	<1
South Birmingham	R	Birmingham Womens'	99	96	47
Heart of Birmingham Teaching	S	City	41	90	59
Coventry Teaching	T	Walsgrave	116	97	0
Dudley	U	Russells Hall	42	100	52
Sandwell	W	Sandwell	87	92	77
Solihull	X	Solihull	43	91	<1
Walsall Teaching	Y	Walsall Manor	61	100	77
Wolverhampton City	Z	New Cross	52	94	83

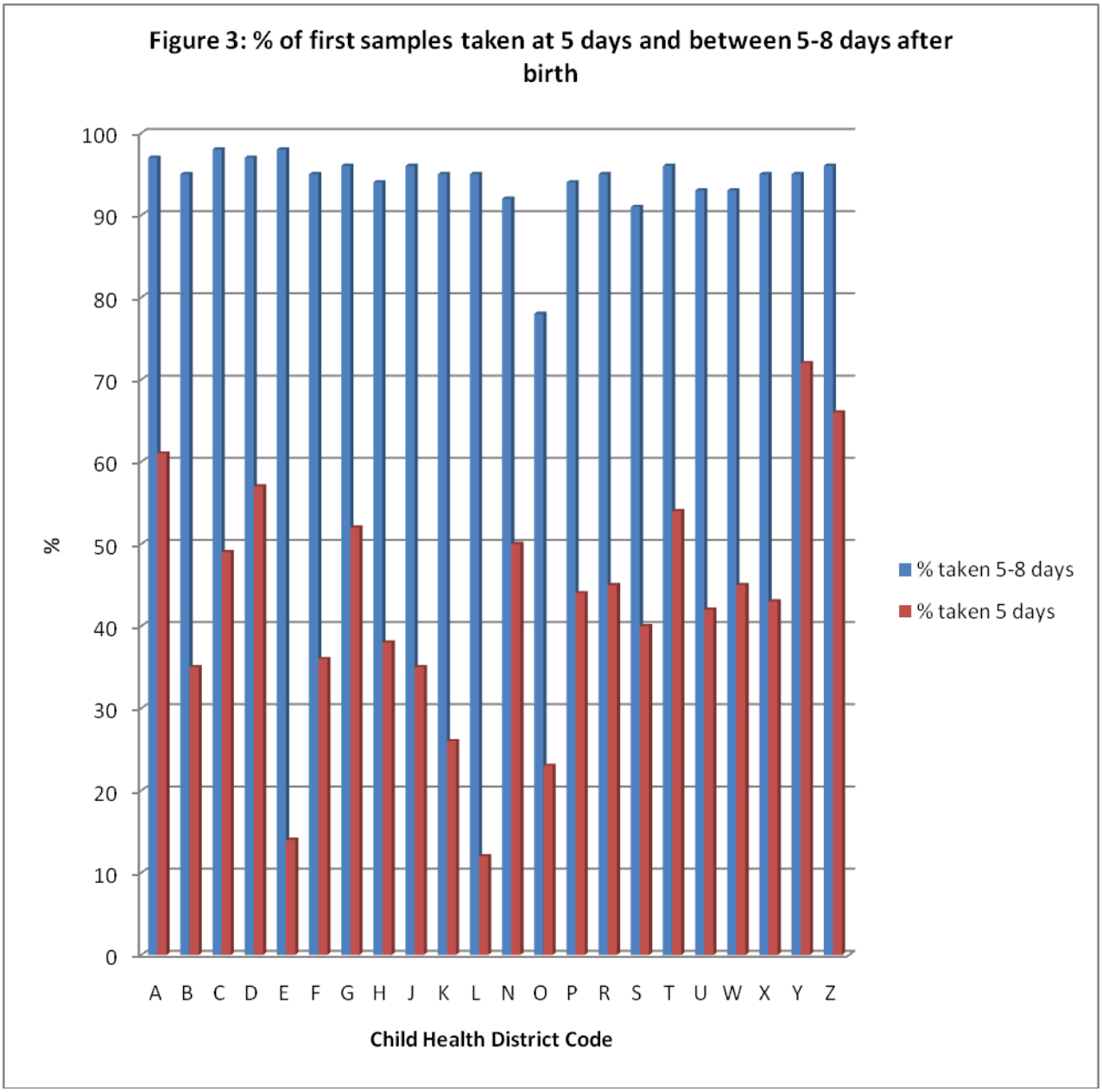
As of March 2010, the number of NHS numbers and labels has been recorded on a daily basis, therefore the report for the next financial year will have data for that complete year.

12.3 STANDARD 3 – Timely sample collection

Core standard: 95 % first samples taken 5-8 days after birth i.e. days of age

Developmental standard: 100% first samples taken 5-8 days after birth

PCT	Code	Associated main Maternity Hospital	% taken 5-8 d of age	% taken 5 days of age
Worcestershire	A	Alexandra	97	61
	C	Kidderminster (community midwifery, no deliveries)	98	49
	D	Worcester Royal	97	57
Herefordshire	B	Hereford County	95	35
Telford and Wrekin Shropshire County	E	Shrewsbury	98	14
South Staffordshire	F	Stafford	95	36
	H	Queen's Burton	94	38
North Staffordshire & Stoke on Trent	G	North Staffs	96	52
Warwickshire	J	Walsgrave	96	35
	K	George Eliot Nuneaton	95	26
	L	Warwick	95	12
Birmingham East and North	N	Heartlands	92	50
	P	Good Hope	94	44
South Birmingham	R	Birmingham Womens'	95	45
Heart of Birmingham Teaching	S	City	91	40
Coventry Teaching	T	Walsgrave	96	54
Dudley	U	Russells Hall	93	42
Sandwell	W	Sandwell	93	45
Solihull	X	Solihull	95	43
Walsall Teaching	Y	Walsall Manor	95	72
Wolverhampton City	Z	New Cross	96	66
Outside West Midlands	O		78	23



Figures for health authority code 'O' (outside region) includes any resident out of the region who may have been transferred in for treatment or may have been tested elsewhere.

The PCT's that fall outside the core standard (95 % first samples taken 5-8 days after birth) are:-

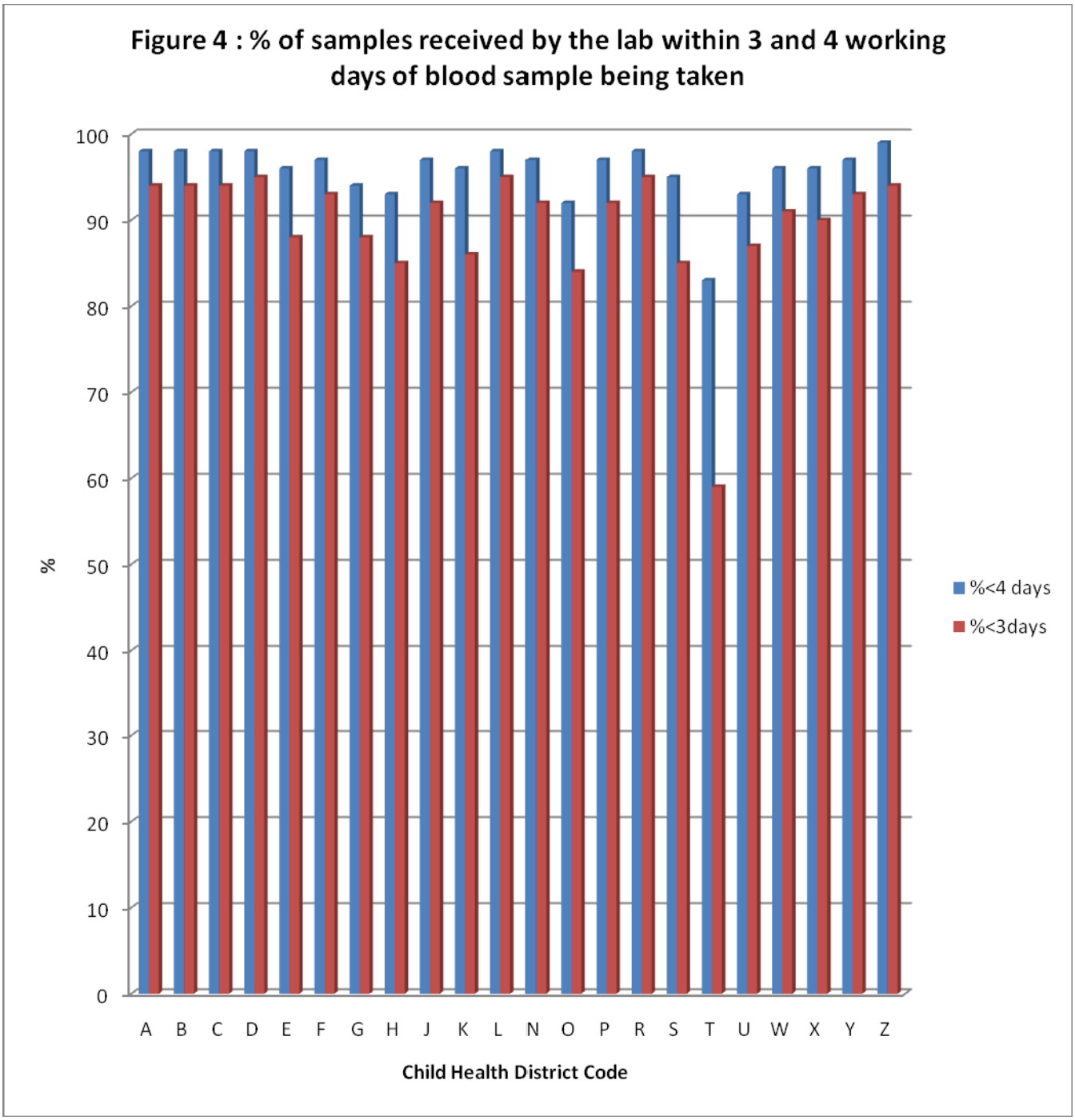
South Staffordshire (H) (94%)
Birmingham East and North (N and P) (92%, 94%)
Heart of Birmingham teaching (S)(91%)
Dudley (U)(93%)
Sandwell (W) (93%)

12.4 STANDARD 4 – Timely sample despatch

Core Standard: 100% of samples received by laboratory within 4 working days of sample collection

Developmental standard: 100% of samples received within 3 working days

PCT	Code	Associated main Maternity Hospital	% received within 4 working days	% received within 3 working days
Worcestershire	A	Alexandra	98	94
	C	Kidderminster (community midwifery, no deliveries)	98	94
	D	Worcester Royal	98	95
Herefordshire	B	Hereford County	98	94
Telford and Wrekin Shropshire County	E	Shrewsbury	96	88
South Staffordshire	F	Stafford	97	93
	H	Queen's Burton	93	85
North Staffordshire & Stoke on Trent	G	North Staffs	94	88
Warwickshire	J	Walsgrave	97	92
	K	George Eliot Nuneaton	96	86
	L	Warwick	98	95
Birmingham East and North South Birmingham Heart of Birmingham Teaching	N P R S	Heartlands Good Hope Birmingham Womens' City	97 97 98 95	92 92 95 85
Coventry Teaching	T	Walsgrave	83	59
Dudley	U	Russells Hall	93	87
Sandwell	W	Sandwell	96	91
Solihull	X	Solihull	96	90
Walsall Teaching	Y	Walsall Manor	97	93
Wolverhampton City	Z	New Cross	99	94
Outside West Midlands	O		92	84



There are no PCTs that meet the core standard (100% of samples received by laboratory within 4 working days of sample collection). The PCT's that fall below 95% are:
South Staffordshire (H) (93%)
North Staffs (G) (94%)
Coventry (T) (83%)
Dudley (U) (93%)

The batching of cards for postage within the Coventry region has been addressed and we anticipate an improvement in the figures for 2010-2011.

12.5 STANDARD 5 – Quality of blood spot sample

A good quality blood spot sample is essential in order to prevent the delay in the identification and treatment of screen positive babies. Poor quality samples that require a repeat to be requested cause anxiety to parents, distress to babies and wastes health care resources.
A good quality blood spot sample is one that is taken at the right time, contains sufficient blood to perform all tests, is despatched to the laboratory in a timely manner and has not been contaminated (UK NSPC Standards and Guidelines for Newborn Blood Spot screening in the UK, August 2008).

A number of repeats are requested for clinical reasons and these are unavoidable repeats (see Table E), however, many of the repeats are avoidable and these include insufficient specimen (e.g. 4 blood spots that have not soaked through to the back of the card), too soon after transfusion (less than 72 hours after transfusion for PKU, CHT, MCADD and CF and less than 4 months for SCD), too young (on or before day 4), unsuitable specimen (e.g. on a blood spot card that has gone past the expiry date, a contaminated sample, transit time in excess of 14 days, anti-coagulated sample).

Core standard
The avoidable repeat rate is less than or equal to 2%.

Developmental Standard
The avoidable repeat rate is less than or equal to 0.5%.

Avoidable repeat rate

The overall avoidable repeat rate affecting all tests is 3.5%. The reasons for repeats requested are shown in Table C pg 14. The number of samples requested for a CF repeat due to unsatisfactory results, due to contamination, for example, = 11 (<1%).

12.6 STANDARD 6 – Timely receipt of a repeat/second blood spot sample

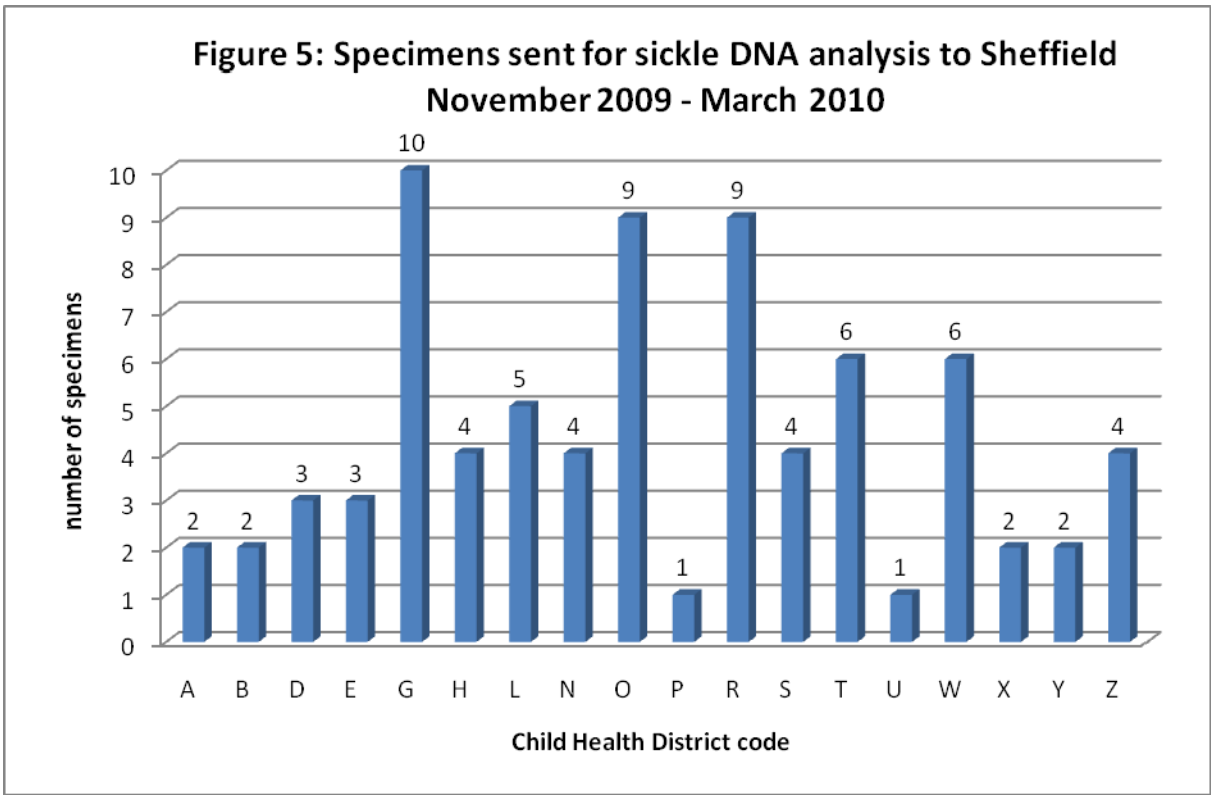
Core standard
95 % of repeat samples should be taken within 72 hours of the laboratory request or specified date of sample.

For CHT this is within 72 hours after the baby reaches the equivalent of 36 weeks gestation (35 weeks + 7 days).

For SCD this is within 72 hours. Pre transfusion specimens should avoid the need for repeats but in the rare cases where this is not possible a sample is sent for sickle DNA testing. The latter service commenced November 2009. Figure 5 shows the number of specimens sent for DNA analysis from November 2009 to March 2010.

For CF the second sample must be taken between day 21 and day 28.

Developmental standard
95 % of repeat samples should be taken within 48 hours of the laboratory request or specified date of sample.
For CHT this is within 48 hours after the baby reaches the equivalent of 36 weeks gestation
The laboratory is currently unable to collect this information.



We are unable to provide the number of specimens sent for DNA analysis which are unavoidable due to intrauterine or immediate post delivery transfusion, although the percentage is very small. The data shown in figure 5 represents the specimens sent for sickle DNA which have largely resulted from the omission of pre-transfusion blood spot collection.

12.7 STANDARD 7 - Timely processing of positive screening samples PKU

PCT	Code	Associated main Maternity Hospital	No with positive result	Age card (s) collected	Working days to referral	Age at referral	Age start of treatment (days)
Worcestershire	A	Alexandra	1	5	1	9	10
	C	Kidderminster					
	D	Worcester Royal	1	5,28	3	35	HPA
Herefordshire	B	Hereford County	1	6	1	11	12
Telford and Wrekin Shropshire County	E	Shrewsbury					
South Staffordshire	F	Stafford					
	H	Queen's Burton	1	3, 7	Early testing as known sib	Early testing as known sib	4
North Staffordshire & Stoke on Trent	G	North Staffs	1	7,8,10	2	16	transient
Warwickshire	J	Walsgrave					
	K	George Eliot Nuneaton					
	L	Warwick					
Birmingham East and North	N	Heartlands					
	P	Good Hope					
South Birmingham	R	Birmingham Womens'					
Heart of Birmingham Teaching	S	City					
Coventry Teaching	T	Walsgrave	1	5	3	11	11
Dudley	U	Russells Hall					
Sandwell	W	Sandwell	1	7,79,108	3	118	HPA
Solihull	X	Solihull					
Walsall Teaching	Y	Walsall Manor	2	5,23	3	29	HPA
				5,24	2	29	transient
Wolverhampton City	Z	New Cross					

HPA:- Hyperphenylalaninaemia

9 presumptive positives from PKU screen

4 cases were confirmed PKU, 1 of these was a sib of a known PKU patient who therefore had early testing done at 3 days of age
3 cases were persistent hyperphenylalaninaemia which were not treated
2 cases were transiently raised phenylalanines that normalised on subsequent specimens

Core standard:	100% positive screening results available **and clinical referral initiated** within 4 working days of sample receipt
Developmental standard:	100% positive screening results available **and clinical referral initiated** within 3 working days of sample receipt

100% compliance with core standard

100% compliance with developmental standard

Clinical referral guidelines

98% babies should have a clinical referral initiated by 14 days of age

Out of the 4 confirmed cases, 4 had a clinical referral initiated by 14 days of age.

100% compliance with core standard

Dietary treatment should be commenced by:

Core standard	21 days of age
Developmental standard	18 days of age

Out of the 4 babies treated, 4 were treated by 18 days of age

100% compliance with core standard

100% compliance with developmental standard

12.8 STANDARD 7 – Timely processing of positive screening samples CHT

PCT	Code	Associated main Maternity Hospital	No with positive result	Age card (s) collected	Working days to referral	Age at referral	Age start of treatment (days)
Worcestershire	A	Alexandra					
	C	Kidderminster	1	8	1	15	15
	D	Worcester Royal	3	5	2	9	10
				5	2	8	9
				5	2	9	10
Herefordshire	B	Hereford County	3	6	2	17	17
				7	2	14	16
				6,16	2	22	transient
Telford and Wrekin Shropshire County	E	Shrewsbury	2	7	2	15	19
				6,9,25	2	36	38
South Staffordshire	F	Stafford					
	H	Queen's Burton	4	11,18	2	23	transient
				10	3	16	Referred to Sheffield
				6	2	8	10
				6	2	9	10
North Staffordshire & Stoke on Trent	G	North Staffs	2	8	2	14	15
				6	1	12	13
Warwickshire	J	Walsgrave					
	K	George Eliot Nuneaton	2	6,30,43	2	48	transient
				6	3	13	14
	L	Warwick	2	11,19,20	2	26	42
				10	1	16	17

12.8 STANDARD 7 – Timely processing of positive screening samples CHT *(cont.)*

PCT	Code	Associated main Maternity Hospital	No with positive result	Age card (s) collected	Working days to referral	Age at referral	Age start of treatment (days)
Birmingham East and North	N	Heartlands	6	5,23	2	27	32
				5	2	11	14
				5	2	9	14
				6	2	14	14
				8	2	14	15
				5	2	12	13
	P	Good Hope					
South Birmingham	R	Birmingham Womens'	5	8	3	14	14
				5	2	12	13
				7	2	15	17
				11	2	15	19
				6	2	11	16
Heart of Birmingham Teaching	S	City	2	5,9,16	2	22	22
				6	3	13	14
Coventry Teaching	T	Walsgrave	6	5	2	10	10
				21	*NA	*NA	28
				6	2	13	13
				5	2	11	11
				2,9,18	2	22	34
				9	2	15	16
Dudley	U	Russells Hall	2	7	2	10	10
				5	3	12	13
Sandwell	W	Sandwell	5	6	2	14	15
				5	2	12	15
				12,42	4	50	Downs Syndrome, transient
				6,22	3	29	transient
				6	2	10	transient

12.8 STANDARD 7 – Timely processing of positive screening samples CHT *(cont.)*

PCT	Code	Associated main Maternity Hospital	No with positive result	Age card (s) collected	Working days to referral	Age at referral	Age start of treatment (days)
Solihull	X	Solihull					
Walsall Teaching	Y	Walsall Manor	3	Not written on card	2	11	12
				6,21	3	28	31
				5	2	11	11
Wolverhampton City	Z	New Cross	2	5	2	11	12
				5	2	10	13

*Initial newborn screening specimen analysed in Sheffield.

Core standard: 100% positive screening results available **and clinical referral initiated** within 4 working days of sample receipt

Developmental standard 100% positive screening results available **and clinical referral initiated** within 3 working days of sample receipt

100 % compliance with core standard
100 % compliance with developmental standard

Clinical referral guidelines

98% babies should have a clinical referral initiated by 14 days of age

For those babies that had a presumptive positive result based on the first TSH result, i.e.no repeat required, 30/38 (79%) were referred by 14 days of age.

79 % compliance with core standard

Thyroxine should be commenced in 100% cases by:

Core standard 21 days of age
Developmental standard 18 days of age

(Accepted that this standard may not be achievable in babies with borderline results or when positive only on repeat screening)

43 babies were treated for CHT. Treatment commenced by 21 days of age in 36 of the 43 patients (84%). The developmental standard of 18 days of age was met for 34 babies (79%).

84% compliance with core standard

79% compliance with developmental standard

Details of babies treated after 21 days of age

District code	Age at referral (days)	Age at start of treatment (days)	Background or reason
T	NA	28	Initial newborn screening specimen analysed in Sheffield. Patient was subsequently followed up in the West Midlands and treated.
S	22	22	In-patient, first specimen too soon after transfusion. Second sample was unsuitable due to multispotting.
Y	28	31	Borderline TSH, subsequently treated.
E	36	38	Borderline TSH, subsequently treated.
T	22	34	First 2 specimens taken too soon after transfusion. Third specimen taken at 18 days and showed marginally increased TSH. Subsequently treated.
L	26	42	Borderline TSH, subsequently treated at 42 days.
N	27	32	Initial borderline TSH, repeat not taken until 23 days, subsequently treated.

6 out of the 7 babies treated >21 days of age had only marginally increased TSH and therefore could be classed as non severe cases of congenital hypothyroidism. The one baby that did have an initial TSH >100 mU/L was treated at 22 days of age due the initial sample being taken too soon after transfusion, the repeat sample was unsuitable due to the sample being multispotted. The third sample was not taken until 16 days of age and took 4 days to reach the laboratory.

12.9 STANDARD 7 – Timely processing of positive screening samples CF

Standards for CF: Time to referral: Within 1 day of DNA result availability
Clinic by 28 days old when a repeat card was not required, others by 35 days

PCT	Code	Associated main Maternity Hospital	No with positive result	Age card (s) collected	Working days to referral	Age (start of treatment) clinic appointment	Mutation Analysis
Worcestershire	A	Alexandra	0				
	C	Kidderminster	0				
	D	Worcester Royal	1	8,29	2	37	None detected (false positive)
Herefordshire	B	Hereford County	2	7	0	19	P.Phe508del hom
				6	1	22	P.Phe508del hom
Telford and Wrekin Shropshire County	E	Shrewsbury	2	6	0	21	P.Phe508del hom
				5	0	22	P.Phe508del hom
South Staffordshire	F	Stafford	2	6	0	18	P.Phe508del hom
				5,34	1	40	P.Phe508del/unknown
	H	Queen's and Lichfield	2	10	3	44	None detected (false positive)
				5	1	27	P.Phe508del hom
North Staffordshire & Stoke on Trent	G	North Staffs	3	6	0	19	P.Phe508del hom
				7	0	Prenatal diagnosis	P.Phe508del hom
				6	0	23	P.Phe508del hom
Warwickshire	J	Walsgrave	1	6	0	30	P.Phe508del hom
	K	George Eliot Nuneaton	0				
	L	Warwick	0				
Birmingham East and North	N	Heartlands	2	8	0	26	P.Phe508del hom
				27	0	Clinical diagnosis	P.Phe508del hom
	P	Good Hope	0				
South Birmingham	R	Birmingham Womens'	2	6	0	15	P.Phe508del hom
				5,29	3	39	c.292C>T/c.292C>T
Heart of Birmingham (T)	S	City	0				
Coventry Teaching	T	Walsgrave	3	6	2	26	P.Phe508del/p.Gly542X
				5	0	0	P.Phe508del hom
				91	0	echogenic bowel*	P.Phe508del/p.Gln493X
Dudley	U	Russells Hall	0				

12.9 STANDARD 7 – Timely processing of positive screening samples CF *(cont.)*

PCT	Code	Associated main Maternity Hospital	No with positive result	Age card (s) collected	Working days to referral	Age (start of treatment) clinic appointment	Mutation Analysis
Sandwell	W	Sandwell	0				
Solihull	X	Solihull	0				
Walsall Teaching	Y	Walsall Manor	3	5	0	20	P.Phe508del hom
				5	0	33	P.Phe508del hom
				6	1	25	P.Phe508del hom
Wolverhampton City	Z	New Cross	4	6,29,37	2	42	None detected
				5	0	21	P.Phe508del hom
				6	1	26	P.Phe508del hom
				6	4	42	P.Phe508del/unknown
**Out of area	O	Sheffield	1	7	1	meconium ileus	P.Phe508del hom
		Bristol	1	7	0	17	P.Phe508del hom

*No blood spot was collected in the newborn period for this baby (in-patient at Leicester). Clinical diagnosis of CF was made before blood spot received in our lab at 91 days of age.

**External results

Core standard: 100% clinical referral should be initiated for positive screening results within 1 day of DNA result availability

21/27 babies (78%) were referred within 1 day.

78% compliance with standard for time to clinical referral

Clinical referral guidelines

100% to clinic by 28 days old, others by 35 days when a repeat card was required

22 of the 27 referred cases were confirmed positives. Out of those 22 cases, 3 were already known cases of CF.
Out of the 19 cases, a repeat card was not required in 18 cases, 16 were seen by 28 days (89%). 1 case where a repeat was required was seen in clinic by 39 days of age.

89% compliance with standard for babies with 2 mutations

CF Carriers

16 carriers were detected, 15 were carriers for the P.Phe508del, 1 was a carrier for the G551D mutation. All cases were referred to a specialised CF counsellor.

12.10 STANDARD 7–Timely processing of positive screening samples MCADD

5 presumptive positive cases for MCADD have been detected in this financial year.
Out of the 5 presumptive positive cases:-
1 case is homozygous for the common mutation (985A>G)
3 cases are compound heterozygotes

*1 case is not MCADD. This baby was unwell at the time the blood was taken and had substantial weight loss.
No missed cases of MCADD have been notified to the laboratory

PCT	Code	No with positive result	Age card (s) collected	Working days to referral	Age (start of treatment) clinic appointment	Diagnosis
Worcestershire	A					
	C					
	D					
Herefordshire	B	1	6	2	12	*NOT MCADD
Telford and Wrekin Shropshire County	E	1	6	1	12	985A>G/985A>G
South Staffordshire	F					
	H					
North Staffordshire & Stoke on Trent	G	2	5	1	11	985A/G/c.388-19T/A,
			5	1	7	253G>T/199T>C
Warwickshire	J					
	K					
	L					
Birmingham East and North	N					
	P					
South Birmingham	R					
Heart of Birmingham Teaching	S					
Coventry Teaching	T					
Dudley	U					
Sandwell	W					
Solihull	X					
Walsall Teaching	Y					
Wolverhampton City	Z	1	6	2	14	985A>G/250C>T

Core standard:	100% positive screening results available **and clinical referral initiated** within 4 working days of sample receipt
Developmental standard:	100% positive screening results available **and clinical referral initiated** within 3 working days of sample receipt

100% compliance with core standard

100% compliance with developmental standard

Clinical referral guidelines

98% of babies should have a clinical referral initiated by 14 days of age

100% compliance with core standard

12.11 STANDARD 8 – Timely identification of babies for whom the child health records department has not received notification of specimen received in laboratory, screening test result or decline

This data is to be provided by the Child Health Record Departments

12.12 STANDARD 9 - Completeness of uptake

This data is to be provided by the Child Health Record Departments

12.13 Screening For Sickle Cell Disorders

PCT	Code	Significant disorders					Non-significant		Carriers reported			
		FS	FSC	FS-other	FE	F only	FC	FD	FAS	FAC	FAD	FAE
Worcestershire	A								5	1	8	
	C								1			2
	D								1	1		
Herefordshire	B										1	1
Telford and Wrekin Shropshire County	E								13	2	1	3
South Staffordshire	F								7	1		3
	H					1			2	1	3	3
North Staffordshire & Stoke on Trent	G					1			26	7	4	5
Warwickshire	J								3	2	2	2
	K							1	8		1	1
	L								9	1		2
Birmingham East and North	N	1	2						45	5	9	25
	P			1			1		27	6	4	4
South Birmingham	R	4				1		2	75	22	18	11
Heart of Birmingham Teaching	S	10			1	1			95	32	12	12
Coventry Teaching	T	2	1						72	7	6	3
Dudley	U								19	6	11	2
Sandwell	W	2	1						54	19	21	5
Solihull	X				1				6		1	1
Walsall Teaching	Y	1			1		1		22	9	4	5
Wolverhampton City	Z	1	1					1	60	22	6	
Totals		**21**	**5**	**1**	**3**	**4**	**2**	**4**	**550**	**144**	**112**	**90**

Key to haemoglobin patterns and presumptive, unconfirmed classification:

FS Sickle cell disease or S HPFH
FSC SC type sickle cell disease
FS-other SD, SE, SV
FE haemoglobin E disease
F only B thalassaemia major
FC haemoglobin C disease
FD haemoglobin D disease

FAS Sickle carrier
FAC Hb C carrier
FAD Hb D carrier
FAE Hb E carrier

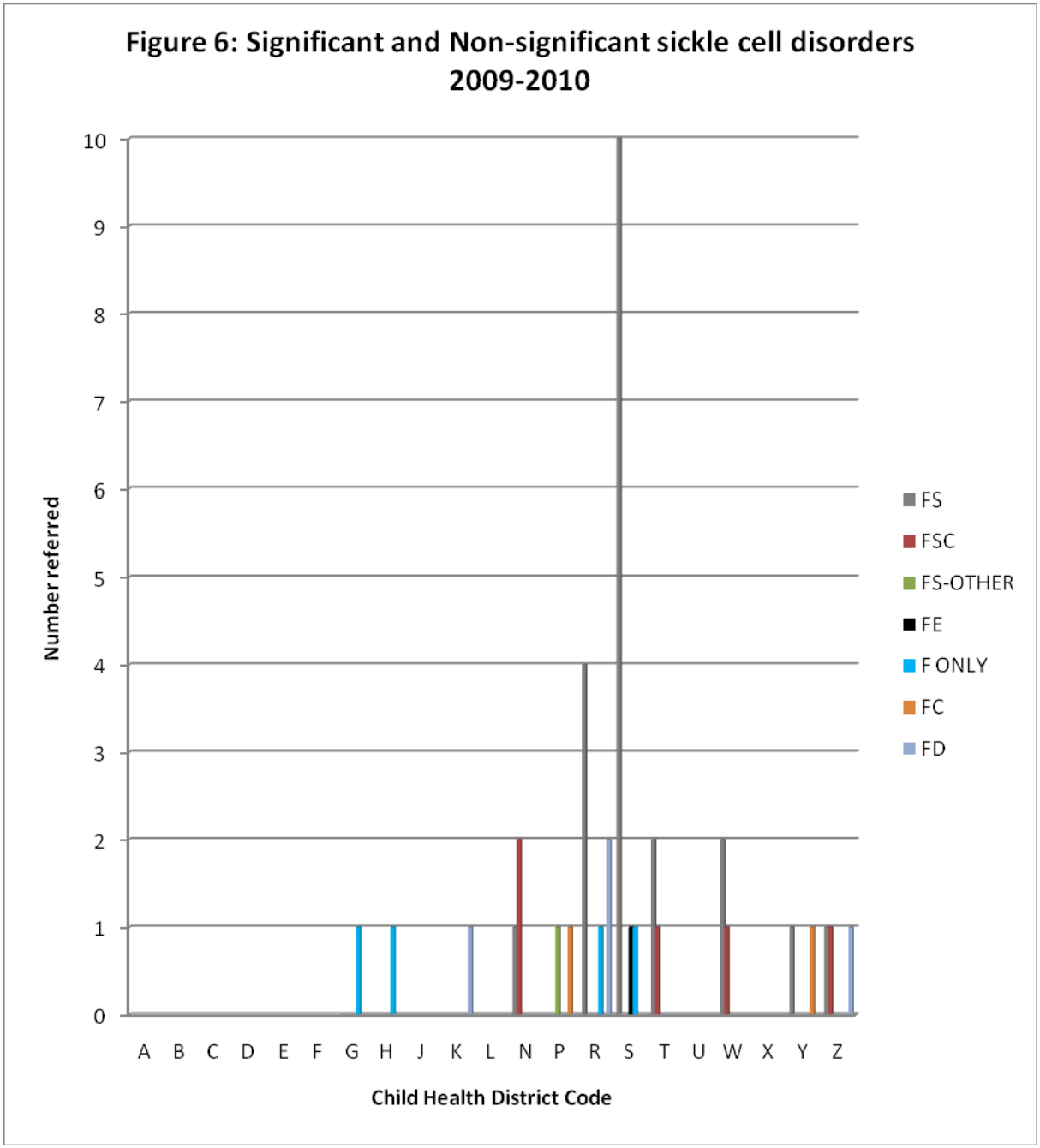
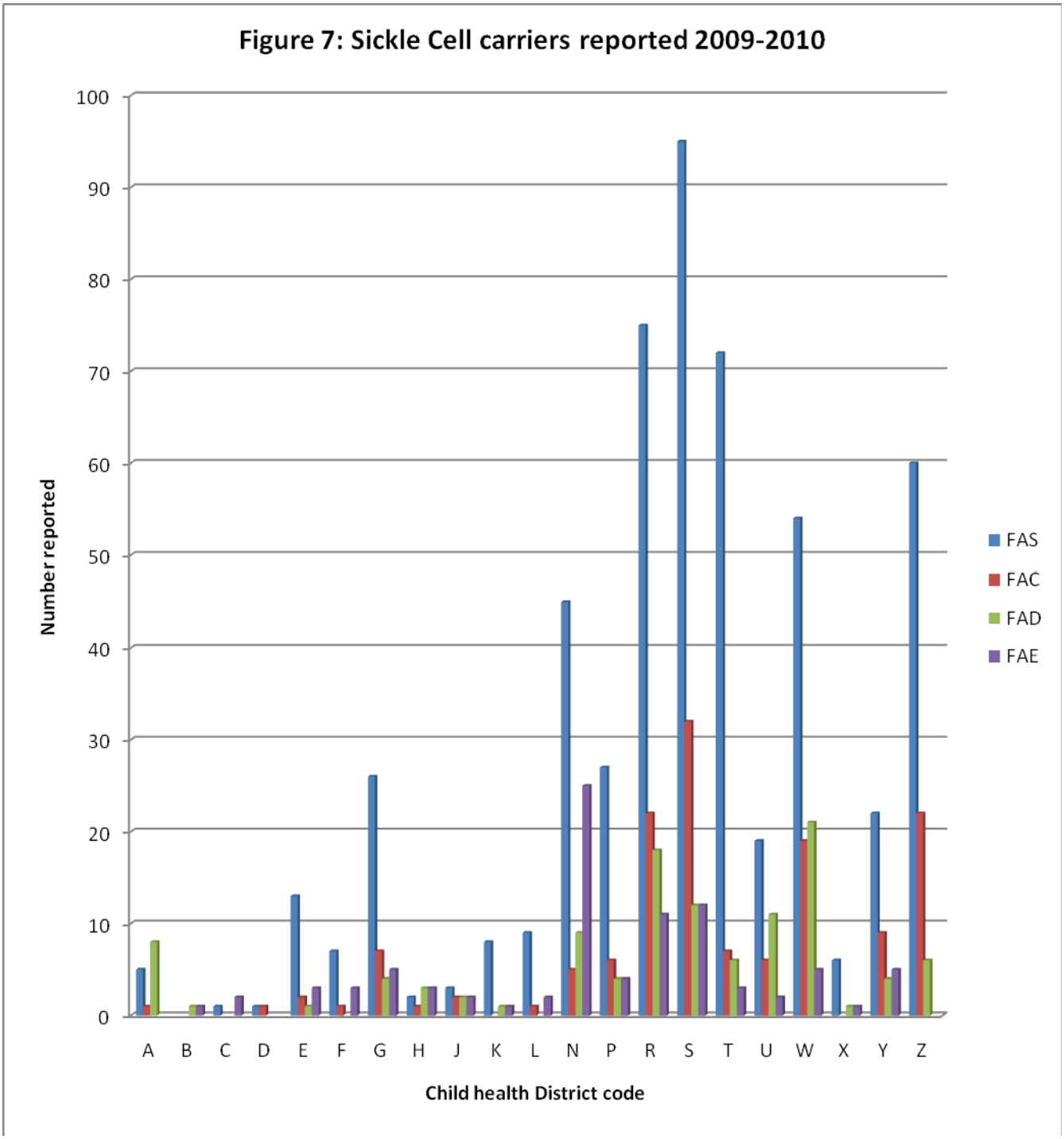


Figure 6: Significant and Non-significant sickle cell disorders 2009-2010
Number referred
10
9
8
7
6
5
4
3
2
1
0
A B C D E F G H J K L N P R S T U W X Y Z
Child Health District Code
FS
FSC
FS-OTHER
FE
F ONLY
FC
FD



Note: Repeat testing organised by haemoglobinopathy counsellors at 6 months of age for the unusual haemoglobin carriers identified stopped during 2009 for all Birmingham and the majority of West Midland babies. Current analytical technology had been proven to be significantly accurate that confirmatory testing was agreed not to be required.

12.14 Performance Standards for process of sickle screening

Hb Status	Age at nurse visit	Age at first clinic appt
FS	4 weeks 4 days	6 weeks 2 days
FS	4 weeks 1 day	5 weeks 4 days
FS	4 weeks 5 days	7 weeks 3 days
FS	5 weeks 2 days	7 weeks 3 days
FS	cord blood taken	6 weeks 4 days
FS	4 weeks	5 weeks 3 days
FS	4 weeks 1 day	5 weeks 1 day
FS	4 weeks 3 days	7 weeks 2 days
FS	4 weeks	7 weeks 1 day
FS	3 weeks 4 days	4 weeks 3 days
FS	3 weeks 2 days	4 weeks 1 day
FS	4 weeks 4 days	7 weeks 2 days
FS	3 weeks 3 days	5 weeks
FS	4 weeks 5 days	6 weeks 6 days
FS	pre-natal diagnosis	pre-natal diagnosis
FS	3 weeks 4 days	5 weeks 3 days
FS	4 weeks 1 day	8 weeks 2 days
FS	Coventry follow-up (no visit from BCH CNS)	7 weeks
FS	4 weeks	6 weeks
FS	6 weeks	6 weeks 6 days
FS	5 weeks 2 days	5 weeks 6 days
FSC	2 weeks 6 days	5 weeks 2 days
FSC	4 weeks 4 days	4 weeks 5 days
FSC	4 weeks 1 day	4 weeks 5 days
FSC	Coventry follow-up (no visit from BCH CNS)	6 weeks
FSC	3 weeks 6 days	4 weeks 4 days
FSD	known family seen in Sib clinic	7 weeks 4 days
FE	5 weeks	6 weeks
FE	4 weeks 2 days	5 weeks
FE	5 weeks 6 days	7 weeks
F	5 weeks	7 weeks 1 day
F	3 weeks	6 weeks 1 day
F	known family seen in Sib clinic	4 weeks 1 day
F	5 weeks	5 weeks 2 days
FC	4 weeks 6 days	7 weeks 6 days
FC	4 weeks 6 days	7 weeks 1 day
FD	4 weeks 4 days	4 weeks 5 days
FD	4 weeks 4 days	4 weeks 5 days
FD	4 weeks 4 days	5 weeks 5 days
FD	3 weeks 4 days	5 weeks 2 days

- **Standard P4: 95% of parents of infants with a positive screening result notified in person by nominated health care professional by 4 wks of age.**

If using cut off as <4wks
Out of 40 positive sickle screening results, 34 were visited by the Clinical Nurse Specialist from Birmingham Children's Hospital. The other 6 cases, 2 were seen in the sib clinic as they were known families, 1 was a cord blood and 2 were seen by haemoglobinopathy counsellors in Coventry. Of the 34 cases, 11 were notified by a health care professional by 4 wks of age **(32%)**

If using cut off as <5wks
Out of 40 positive sickle screening results, 34 were visited. Of these 34, 30 were notified by a health care professional by <5 wks of age **(88%)**

Whilst not achieving the standard, it should be noted that babies are protected by foetal haemoglobin up to the age of 3 months.

- **Standard P4: 95% attend local clinic by 3 months of age**

Out of 40 positive sickle screening results, 39 were seen in clinic and 1 was a pre-natal diagnosis. Of these 39, 39 attended the local clinic by 3 months of age **(100%)**

- **Standard P4: 95% registered with clinic by 8 wks of age**

Out of 40 positive sickle screening results, 39 were seen in clinic. Of these 39, 39 attended the local clinic by 8 wks of age **(100%)**

13 Clinical Governance

13.1 External Quality Assurance (EQA)

The laboratory participates in the following EQA schemes :

- UK NEQAS for blood spot phenylalanine and tyrosine (PKU screening and phenylalanine and tyrosine monitoring)
- UK NEQAS for blood spot thyroid stimulating hormone (CHT screening)
- UK NEQAS scheme for abnormal blood spot haemoglobins (SCD screening)
- DG KC German scheme for blood spot TSH (CHT screening)
- US CDC Quality Control Scheme for blood spot:
  - Phenylalanine
  - Tyrosine
  - Acyl carnitines (C0 and C8)
  - IRT (CF)
- US CDC Proficiency Testing Scheme for blood spot:
  - Tandem Mass Spectrometry
  - Inborn Errors of Metabolism
  - Sickle Cell Disease and Other Hemoglobinopathies.
  - IRT (CF)
  - Cystic Fibrosis Mutation Detection
- Pilot UK scheme for C8 and C10 (MCADD screening)

We are assessed for accuracy of numerical results in comparison with other participating laboratories and also the concordance of our result interpretation compared with that of other laboratories.

Points of note are as follows:-

- Performance in all schemes was considered to be satisfactory.

13.2 Accreditation

The Newborn Screening Laboratory, as part of the Clinical Chemistry Department at Birmingham Children's Hospital, is accredited to CPA standards following the latest assessment in January 2009 (reference number 1288).The surveillance visit is due at the end of November 2010. A user satisfaction survey will be developed and circulated in the next financial year.

13.3 Items of concern

Incorrect NHS numbers

The errors identified in NHS numbers recorded on bloodspot cards either by hand or even on barcoded sticky 'baby' labels continue on and are serious. Incorrect NHS numbers on bloodspot cards are nearly always *someone else's* and not due to transposition of numbers. Erroneous NHS numbers can include the baby's mother's, an older sib's, the other twin's and sometimes someone completely different even in another part of the country. Whilst some of these errors are due to lack of attention on NNUs or in the home removing labels from sib's red books, some are due to poor understanding of the process of generation of NHS numbers from the national issuing service and incorrect practice in Maternity Units on the occasions when NHS numbers are not generated. Some maternity software systems permit the production of 'baby' labels for mothers. Whilst the laboratory has been able to trap most of these errors before results are sent back against the wrong individual, some incidents have occurred where this has not been possible.

Staff in both Child Health and Maternity involved with NHS number generation at birth need to have a full understanding of the task, their role and correct procedures need to be followed. Further training may be required.

Incorrect NHS numbers

PCT	Code	Old Child Health District	Associated main Maternity Hospital	Number of Incorrect NHS numbers
Worcestershire	A	North Worcestershire	Alexandra	2
	C	Kidderminster	Kidderminster (community midwifery, no deliveries)	1
	D	South Worcestershire	Worcester Royal	3
Herefordshire	B	Hereford	Hereford County	1
Telford and Wrekin Shropshire County	E	Shropshire	Shrewsbury	2
South Staffordshire	F	Mid.Staffordshire	Stafford	4
	H	S.E. Staffordshire	Queen's Burton	6
North Staffordshire & Stoke on Trent	G	North Staffs	North Staffs	6
Warwickshire	J	N.E. Warwickshire (Rugby)	Walsgrave	1
	K	North Warwickshire	George Eliot Nuneaton	1
	L	South Warwickshire	Warwick	2
Birmingham East and North	N	East Birmingham	Heartlands	7
	P	North Birmingham	Good Hope	1
South Birmingham	R	South Birmingham	Birmingham Womens'	3
Heart of Birmingham Teaching	S	West Birmingham	City	2
Coventry Teaching	T	Coventry	Walsgrave	4
Dudley	U	Dudley	Russells Hall	4
Sandwell	W	Sandwell	Sandwell	3
Solihull	X	Solihull	Solihull	0
Walsall Teaching	Y	Walsall	Walsall Manor	1
Wolverhampton City	Z	Wolverhampton	New Cross	1
Total				**55**

14 Audit

14.1 Referral Guidelines and Standards

The Newborn Screening Laboratory actively participates in multidisciplinary audit of its service and follow up of patients jointly with paediatricians and other health professionals.

Congenital Hypothyroidism

Year	2004-5	2005-6	2006-7	2007-2008	2008-2009	2009-2010
Number of positives followed up	44	36	40	40	81	50

Sickle Cell Disorders

The sickle programme is audited by a joint laboratory audit group with Clinical Haematology at BCH.

14.2 Information provision

The laboratory provides laboratory address labels. Information leaflets for 'Newborn blood spot screening for your baby' are provided by the UKNSPC.

15 Training and Education

During the year, our Newborn Screening Training Co-ordinators have held training for midwives and other health professionals involved in the collection of dried bloodspots throughout the West Midlands. These sessions have either been held at the Children's Hospital, or locally as requested. They have consisted of teaching both the theoretical and the practical aspects of dried blood spot collection and an overview of CHT, PKU, SCD, CF and MCADD.
The following table shows details of the sessions that have taken place.

Venue	No. of sessions	No. of attendees
Allens Croft Children's Centre	1	9
Birmingham Children's Hospital	11	105
City Hospital	4	58
Coventry	2	43
Millenium Health Centre	1	15
Newcross Hospital	4	53
North Staffs	2	37
Russells Hall Hospital	2	7
Shelton Health Centre	1	12
Total	**28**	**339**

PKU schools

PKU school was held in the Easter Holidays 2010. These schools give children and young people the opportunity to learn about PKU, have fun, meet others with PKU and to learn about their diet. The groups are generally 4-7yrs, older ages 8-11 yrs. For those older than 11yrs there are different activities. The groups are varied but what works best is that the 4-7yr are a mixed group and then there are two separate groups for girls and boys aged 8-11yrs.

16 Research and Development

i) Screening for Tyrosinaemia Type 1 by measurement of succinyl acetone in bloodspots. A method for newborn screening using the measurement of succinylacetone in blood spots by tandem mass spectrometry has been developed.

ii) Development of HPLC method for pterins in dried blood spots.

iii) Continuing involvement with planning for an expanded Newborn Screening project. A report on reviewing the evidence of expanded newborn screening has been commissioned by the NIHR-Collaboration for Leadership in Applied Health Research and Care- South Yorkshire. The report concluded that expanded newborn screening for 5 conditions should be supported and recommended a pilot programme. It is hoped that a pilot study is carried out in 4 centres in England to include 5 additional disorders: maple syrup urine disease, homocystinuria, glutaric aciduria type I, isovaleric aciduria and long chain acyl-coA dehydrogenase deficiency. Birmingham has been put forward as one of the pilot sites.

17 References and publications

UK NSPC Standards and Guidelines for Newborn Blood Spot screening in the UK, August 2008.

NHS Sickle Cell and Thalassaemia Screening Programme, Standards for the linked antenatal and newborn screening programme, November 2006.

A Laboratory guide to Newborn Screening in the UK for MCADD, July 2008, UK Newborn Screening Programme Centre, Dr P.E. Goddard major contributor.

Streetly A, Latinovic R, Hall K, Henthorn J Implementation of universal newborn bloodspot screening for sickle cell disease and other clinically significant haemoglobinopathies in England: screening results for 2005–7 J Clin Pathol. 2009; 62(1): 26–30.

Presentations
(Kate Hall)

27 May 2009 Screening newborns for sickle cell disease. Workshop: Trinity Biotech Primus Scientific Meeting, Northampton.

12-13 June 2009. Practical aspects of newborn screening. Conference: Neonatal Screening – a Chance to Life. Bucharest, Romania.

8 July 2009. A quick guide to newborn screening. Regional Neonatal SHO Study Day, Birmingham Women's Hospital.

2 November 2009 Exclusion of biopterin metabolism disorders – the Birmingham service and its development. At national PKU Guidelines Group meeting, Birmingham Children's Hospital.

11 November 2009. Timely identification, completeness of uptake and results to parents. Regional Newborn Screening meeting. Acton Trussell, Shropshire.

10 February 2010 A quick guide to newborn screening. Regional Neonatal SHO Study Day, Birmingham Women's Hospital.

17 March 2010 Completeness of uptake and normal results to parents. Regional Newborn Screening meeting. Birmingham.