



CMGS AUDIT

Genetics Service
Guy's Hospital
Level 7, Borough Wing
Great Maze Pond
London
SE1 9RT

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Clinical Molecular Genetics Society Audit 2009-10

The Clinical Molecular Genetics Society (CMGS) produce an annual audit of the genetic testing activity undertaken by the laboratories of its members. The audit has been running since 1993 and the laboratories represented comprise nearly all of the UK Regional Molecular Genetic Services plus some specialist services.

The data presented includes the number of samples received and extracted, the number and type by disease of postnatal, prenatal and predictive test reports, the number and type of tests that are sent for analysis at a different network laboratory, reporting times for specific diseases by service level, staff numbers and a comparison of the range in staffing level and workload across the different laboratories. Since 2005, the audit includes the names of the participating laboratories but the individual laboratory codes remain anonymous and change each year.

The data has proved a useful reference source for estimating activity subtypes at national level, for trend analysis particularly with respect to overall volume of activity and reporting times, for the UK professional bodies concerned with the use of genetic tests for insurance purposes and for the individual laboratories for bench marking of their own service.

Challenges faced in the collation and presentation of data include the difficulty for the participant laboratories in collecting the information, establishing and evaluating appropriate standard measures of activity and responding to the changing scope of services and range of providers. This year's audit data from 2009-10, shows a continued growth in national activity for each of the postnatal, prenatal and predictive testing categories, an improvement in routine reporting times and encouraging data on the capturing and utility of activity using the proposed new national unit of activity (MoIU).

Many thanks again to the participant laboratories for providing the core data.

Gail Norbury
CMGS Audit Sub-committee Chair

Participants

Note the laboratories below are listed alphabetically and the anonymous laboratory codes used on the charts are allocated on a random basis and are changed each year. Most but not all participants submitted data for all parts of the audit.

Aberdeen Molecular Genetics Laboratory
All Wales Molecular Genetics Laboratory (Cardiff)
Belfast Regional Molecular Genetics Laboratory
Birmingham Children's Hospital
Bristol Genetics Laboratory
Cheshire & Merseyside Molecular Genetics Laboratory (Liverpool)
Dundee Molecular Genetics Laboratory
East Anglian Molecular Genetics Laboratory (Cambridge)
Exeter Regional Genetics Centre
London North East Thames Molecular Genetics Laboratory (Great Ormond Street)
London North West Thames Regional Genetic Centre (Kennedy Galton)
London South East Regional Genetic Centre (Guys & St Thomas')
London South West Thames Molecular Genetic Diagnostic Laboratory (St Georges)
London UCL Hospitals, Clinical Biochemistry
London UCL Hospitals, Haemoglobinopathy Genetics Centre
London UCL Hospitals, Institute of Neurology (NHNN)
National Haemoglobinopathy Reference Laboratory (Oxford)
International Blood Group Reference Laboratory, Bristol
North West Regional Genetics Centre (Manchester)
Northern Regional Genetics Centre (Newcastle)
Nottingham Molecular Genetics Laboratory
Oxford Regional Genetic Centre
Retinoblastoma Laboratory (Barts)
South East Scotland Genetics Service (Edinburgh)
The Doctors Laboratory (London)
Wessex Regional Genetic Centre (Salisbury)
West Midlands Regional Genetic Centre (Birmingham)
West of Scotland Regional Molecular Genetics Laboratory (Glasgow)
Yorkshire Regional Genetic Centre (Leeds)

1. SAMPLES RECEIVED AND EXTRACTED 2009-10

	Category	Total	Range		Labs	Fails	
		Sum	min	max	Count	Number	%
Samples	Blood	143651	46	17935	28		
	Amnio/CVS	12715	3	6265	25		
	Buccal	1391	2	453	21		
	Archive	5676	3	1559	19		
	Other	13012	8	4124	24		
	DNA	33451	3	4059	28		
	Research	15413	9	4742	19		
Extracts	Blood auto	110093	232	17685	23	842	0.8%
	Blood manual	23974	14	4622	19	213	0.9%
	Amnio & CVB auto	4627	1	2293	10	0	0.0%
	Amnio & CVB manual	8839	3	6265	19	6	0.1%
	Buccal auto	766	9	475	6	2	0.3%
	Buccal manual	413	2	136	13	1	0.2%
	Archive auto	3288	3	1559	10	157	4.8%
	Archive manual	2099	2	1291	10	121	5.8%
	Other auto	7864	17	2248	14	51	0.6%
	Other manual	4380	3	3460	16	29	0.7%
	Research auto	4349	1	1838	9	78	1.8%
	Research manual	1110	6	942	6	72	6.5%
	Totals	Service samples	209896	96	25578	28	
Service extracts		178028	56	24215	28	1411	0.8%
Research samples		15413	9	4742	19		
Research extracts		9246	7	3787	12	221	2.4%

1.1 Table Sample and Extraction summary

Notes

As there was not always clear distinction between the use of a zero and no available submitted data, only positive figures were included in the analysis to determine minimum and fail rate values.

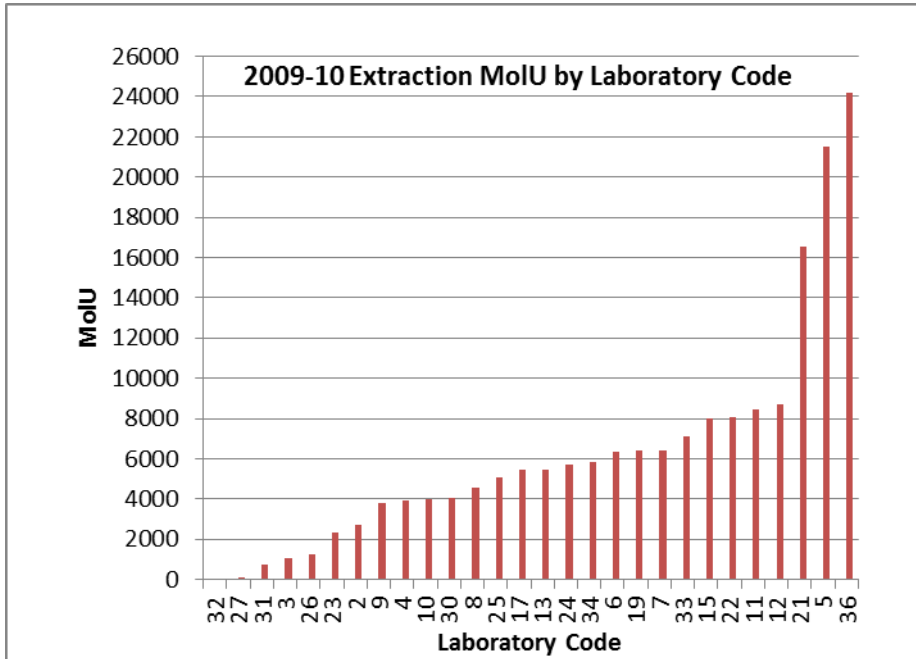
The total number of service samples increased by 16% on last year, (209896 v 181009) that compares to an apparent 19% increase the previous year.

The total number of service extractions increased by 30% (178028 v137154) compared to the 13% increase in the previous year.

Note

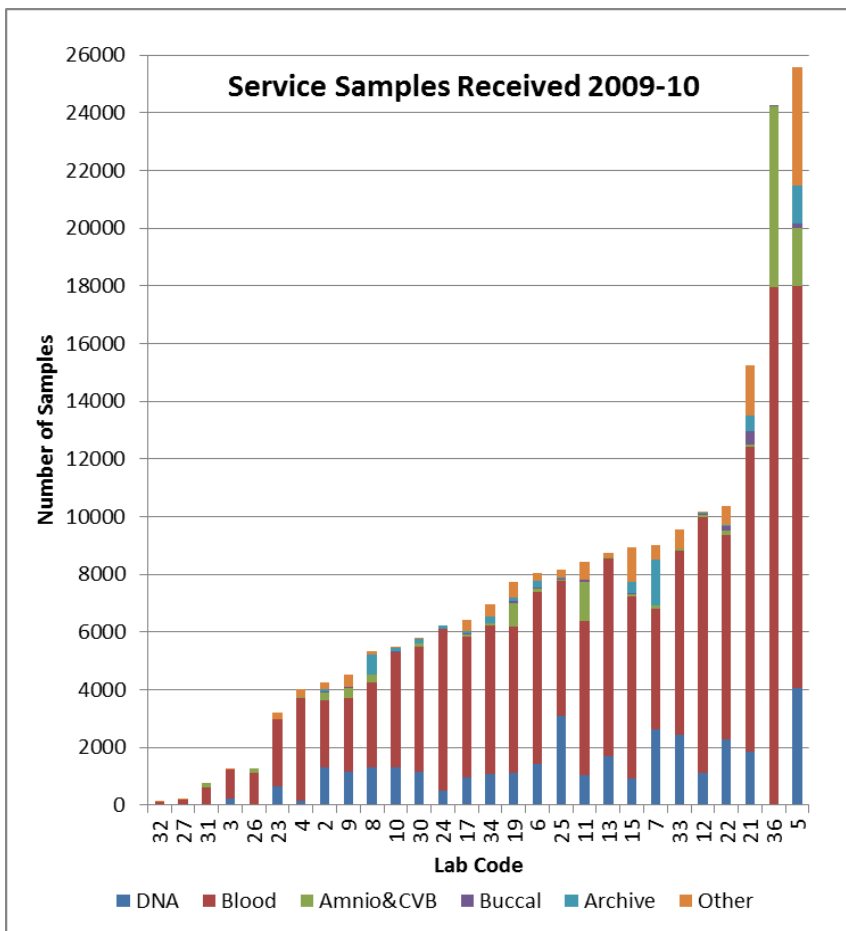
Sample number and extraction number represent different measures of activity; sample received may include aliquots of the same sample being counted multiple times if analysis involves more than one laboratory whilst number of extraction may result in a sample only being counted once despite involving a number of different investigations. Overall, as shown in the table above, extracted DNA accounts for ~16% service samples.

Under the new MoIU workload system, a single unit of activity is accrued for extraction (and storage).

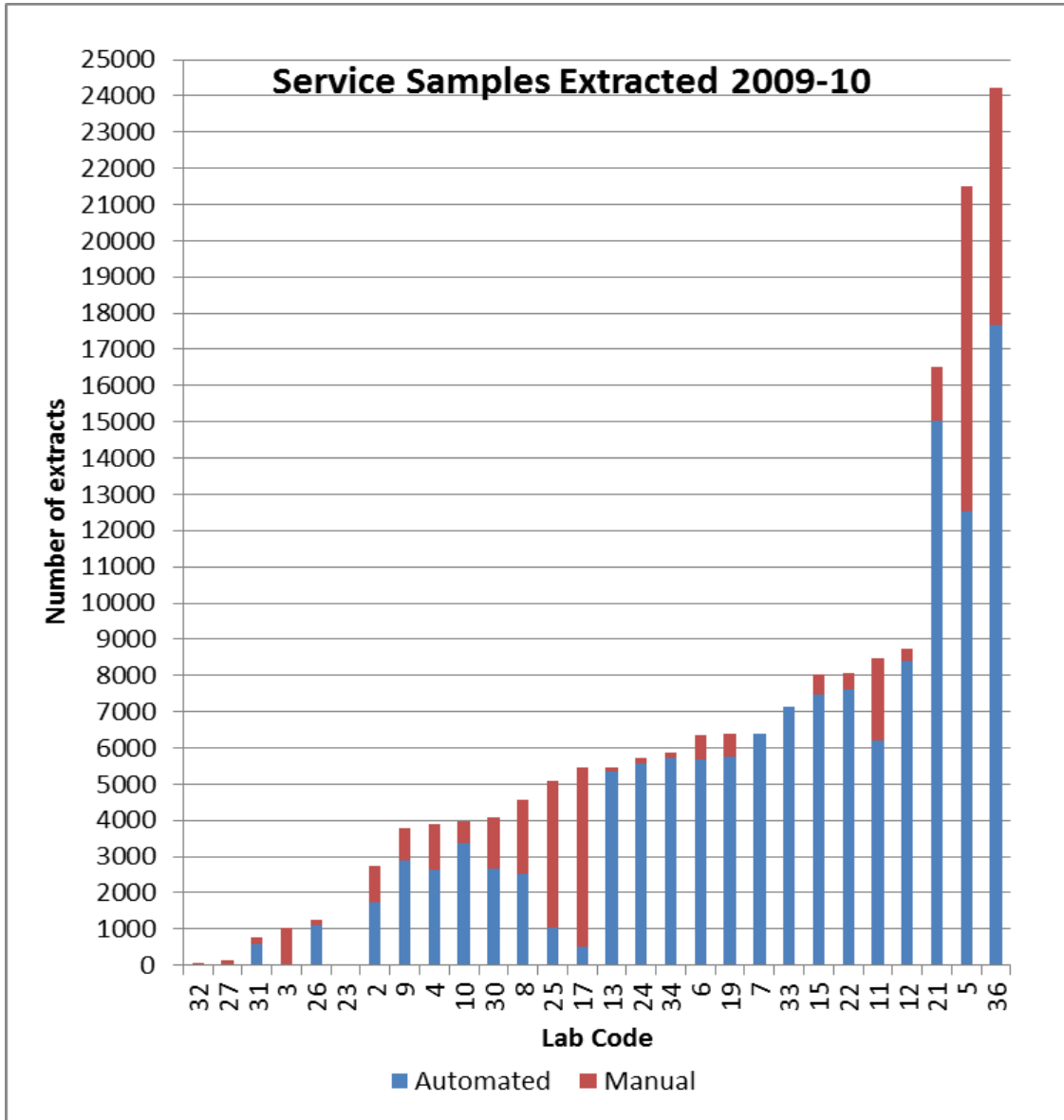


1.2 Chart of Service sample extraction activity by MoIU

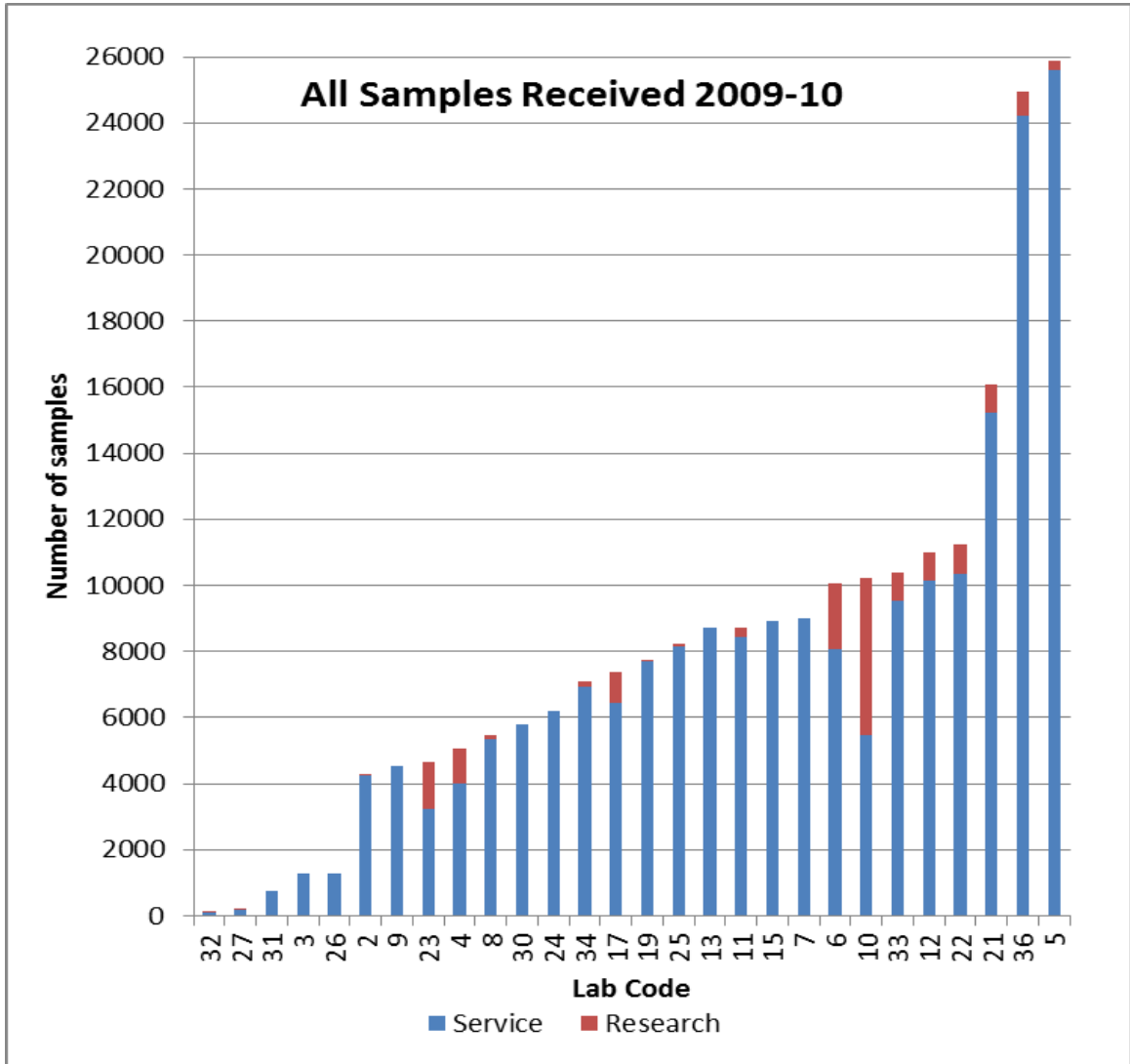
No data was available for Lab 35



1.3 Chart showing samples received by each laboratory, ranked by the total number of samples and broken down by the different sample type. No data was available for Lab 35



1.4 Chart of number of extractions (manual and automated) performed by each laboratory, ranked by total number of extractions. No data was available for Lab 35 and no breakdown in data for Lab 23



1.5 Chart of Grand total samples (service & research) received by each laboratory, ranked by total number of samples received. No data was available for Lab 35

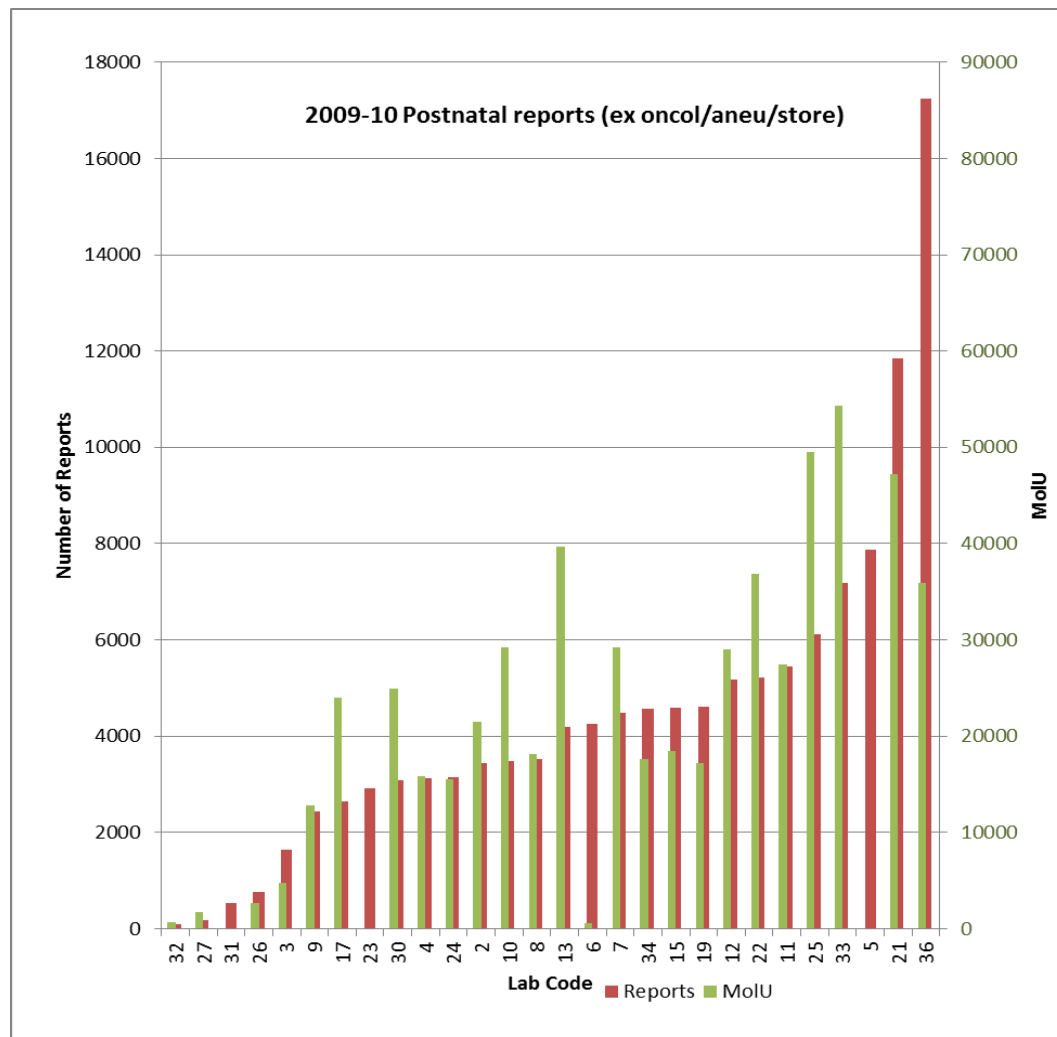
2 POSTNATAL REPORTS 2009-10

The report data as usual is presented separately for postnatal and prenatal reports. The totals are presented with the inclusion and exclusion of activity for acquired oncology work as this large volume work is not undertaken by all laboratories and is not part of the core definition set for medical genetics.

2009-10	Total	Range
Grand total	138,211	87-17,235
Total –acquired oncology/aneuploidy	124,314	87-17,235
Total – acquired oncology/ aneuploidy/ store	123,750	87-17,235

The postnatal activity for 2009-10 shows a 29% increase in the grand total number of reports and a 31% increase in single gene disorder reports i.e. total excluding oncology/chimerism/aneuploidy/store work.

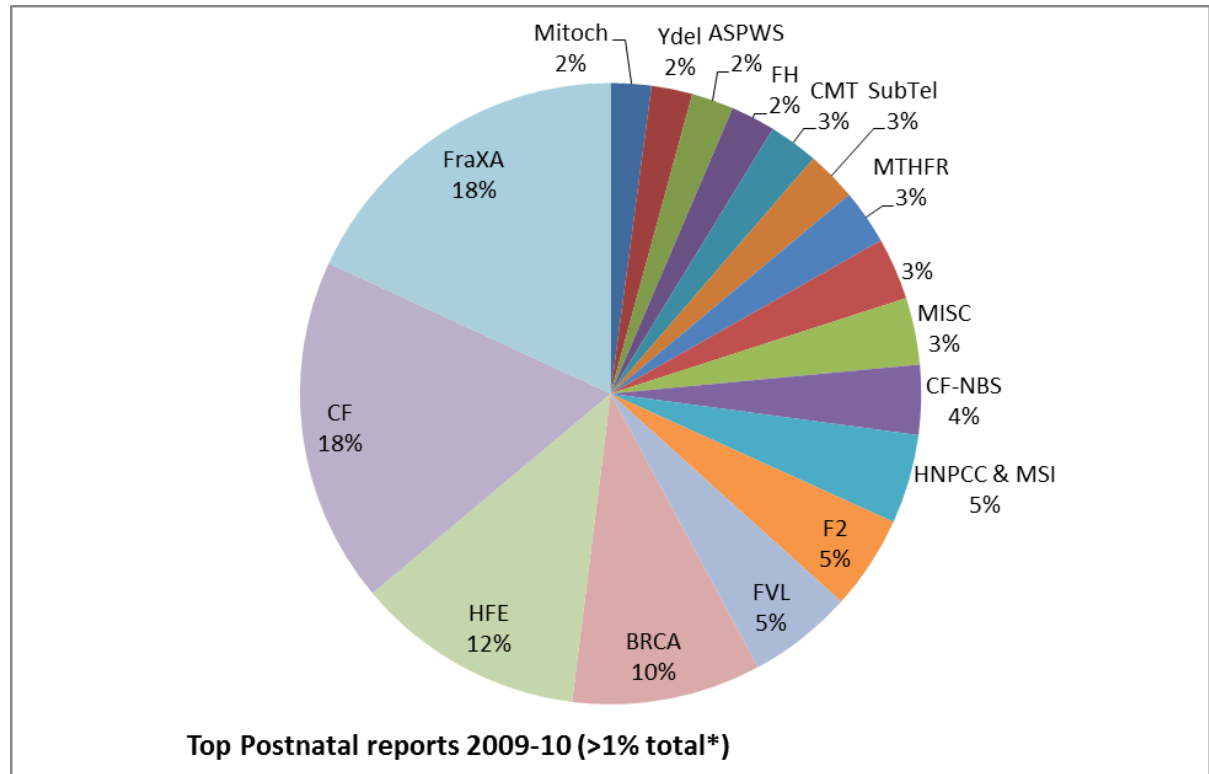
These changes reflect the inclusion of a new large volume provider.



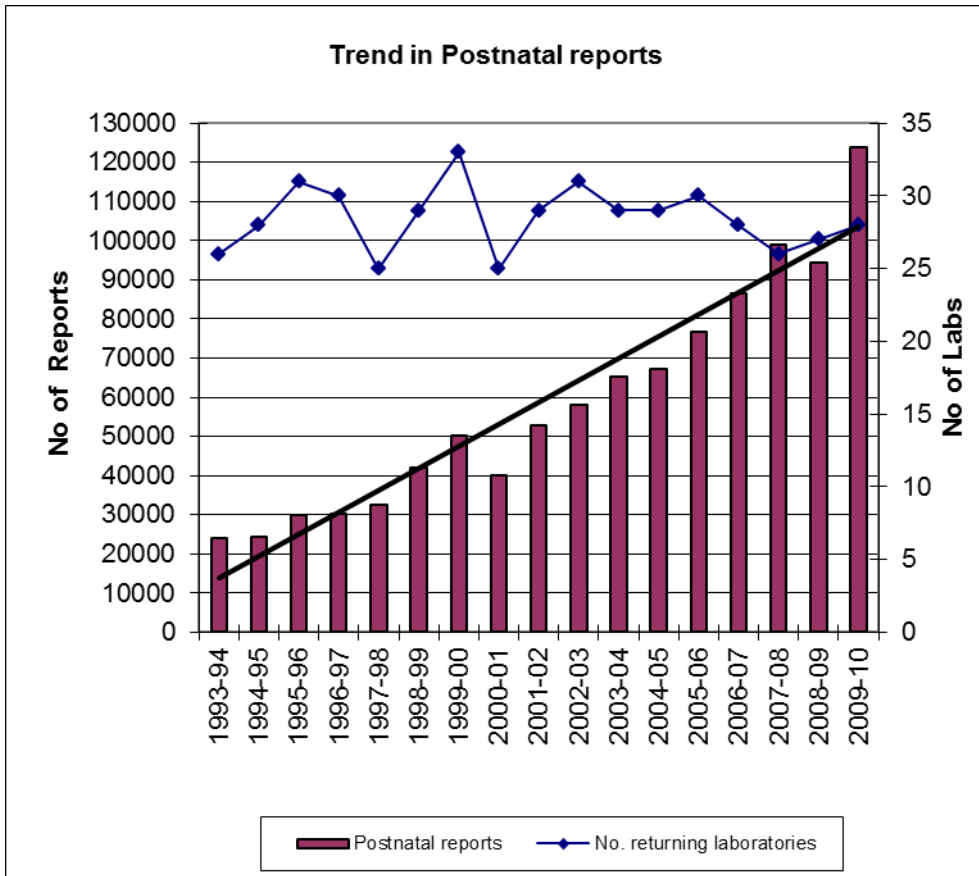
2.1 Total number of postnatal reports and postnatal MolU for each laboratory, ranked by sum of report numbers (excluding acquired oncology, aneuploidy and identifiable bank/store only activity). No MolU data was available for labs 31, 23, 6 or 5.

Range of postnatal reports

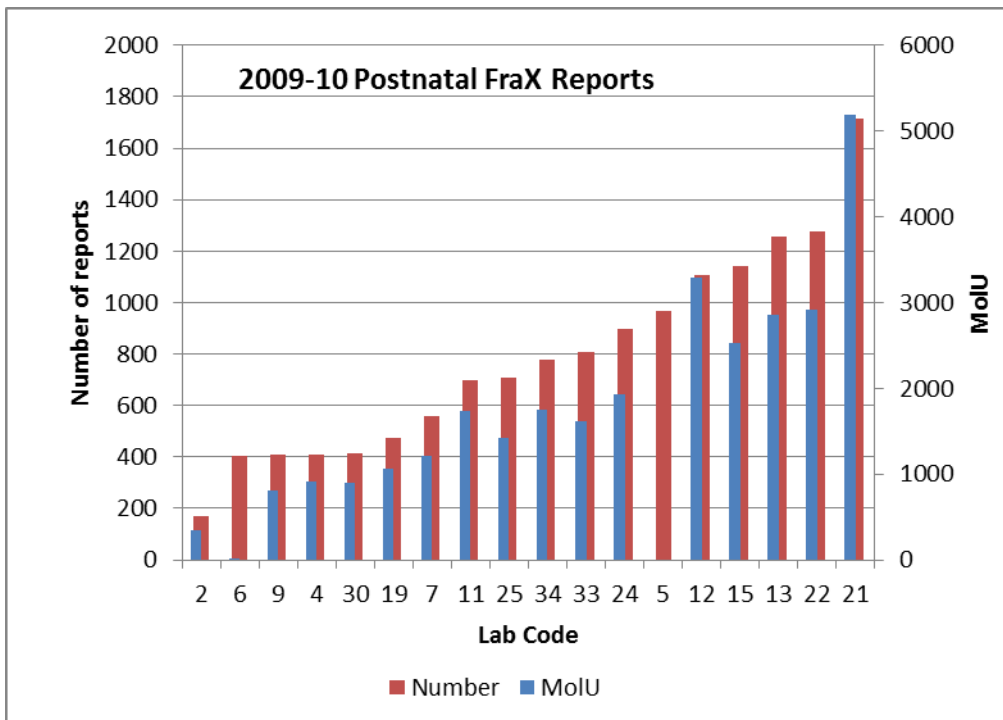
In 2009-10, overall around 465 different categories of postnatal report were recorded. Note this includes a list of “miscellaneous/others” and some laboratories combined certain categories because of the test design e.g. for mitochondrial, PWS/AS, SCAs, CMT. The range in number of providers per test was 1 to 19 laboratories as for the previous year.



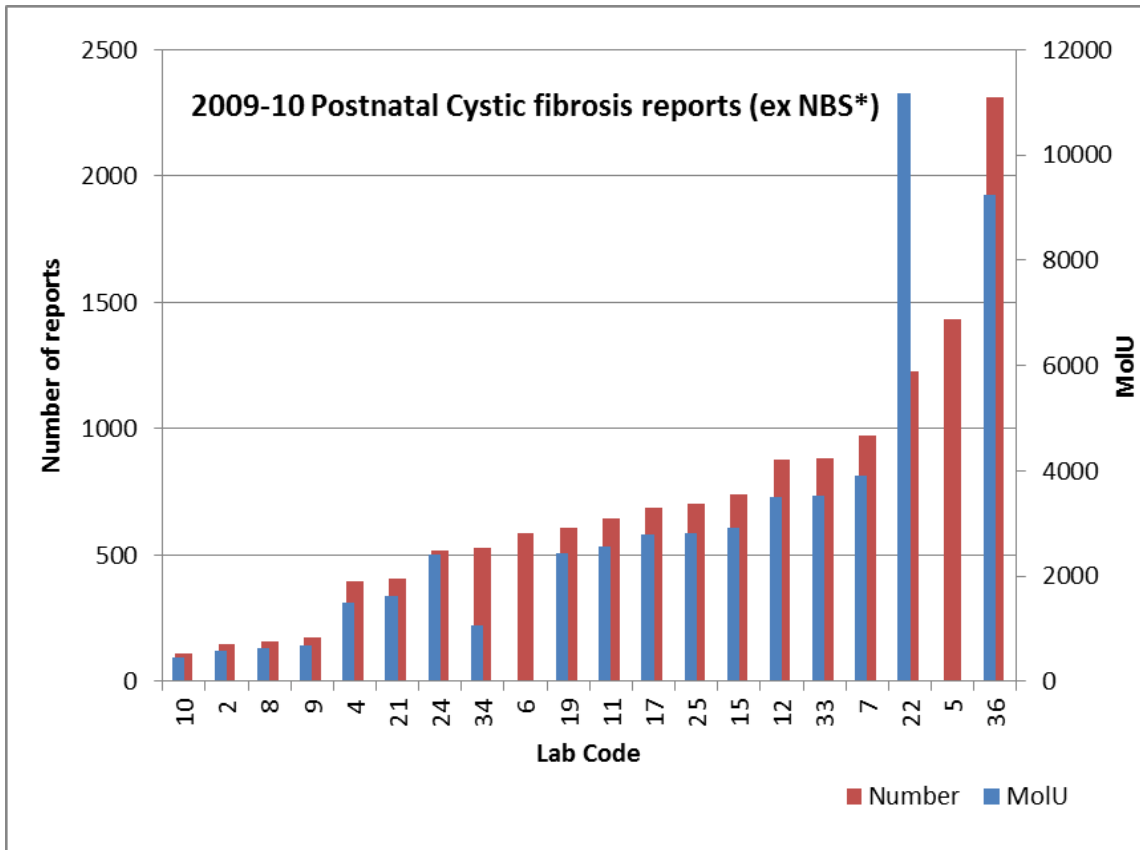
2.2 Pie chart of disease categories that account for more than 1.0% of total annual activity *excluding acquired oncology/aneuploidy activity (total of 123,750 reports)



2.3 Chart showing the trend in postnatal report number with corresponding number of laboratories submitting data over past 17 years



2.4 Fragile X postnatal report activity
 Mean number was 789 reports across the 18 providers and the mean MoIU per report was 2.3. The mode MoIU per report was 2.



2.4 Cystic fibrosis postnatal report activity

*Excludes new-born screening (NBS) activity where identified

Mean number was 706 reports pa across the 20 providers and the mean & mode MoIU per report was 4.0.

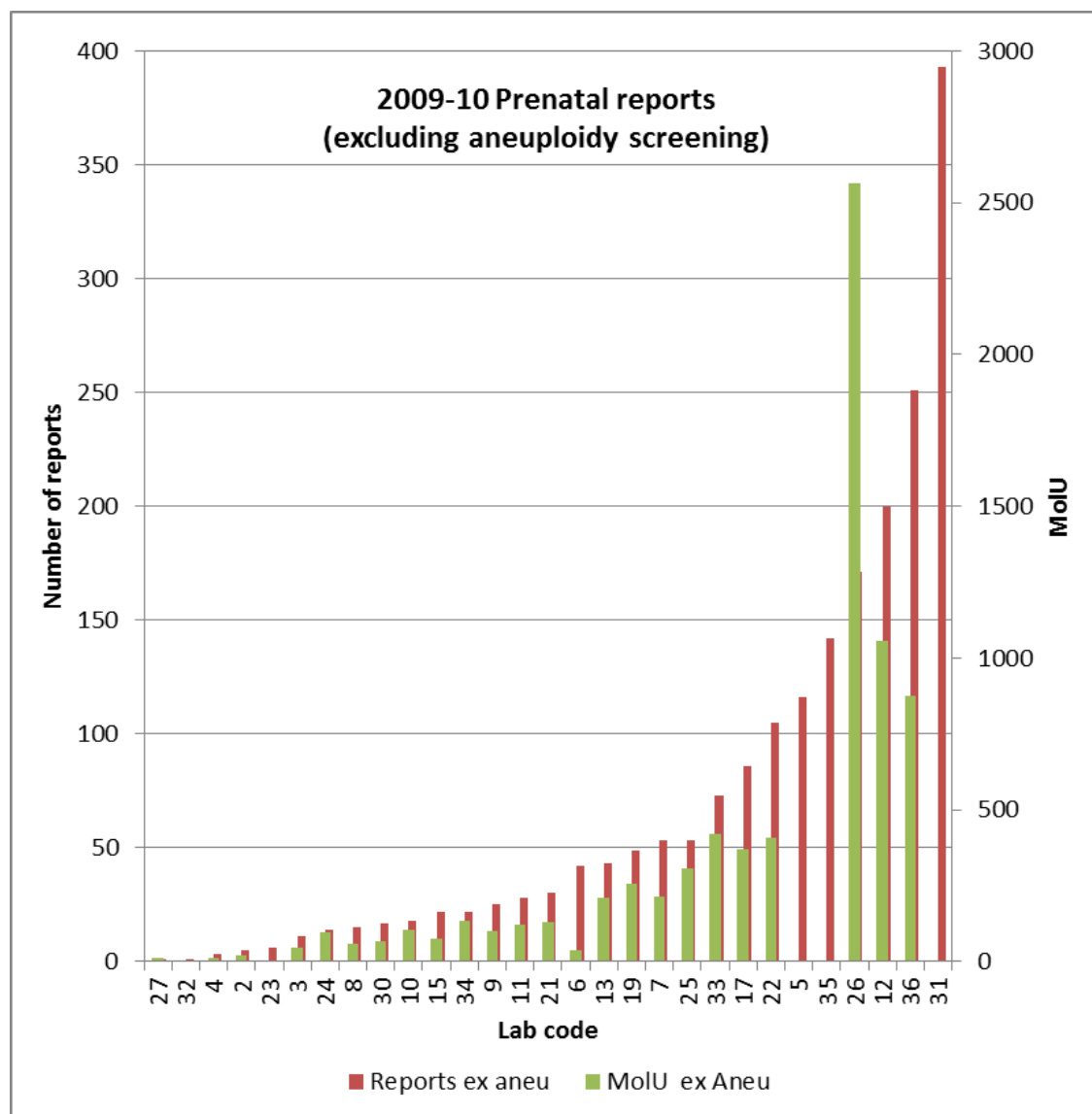
3. PRENATAL REPORTS for 2009-10

Prenatal diagnosis reports were recorded for 120 different disorders by 29 labs.

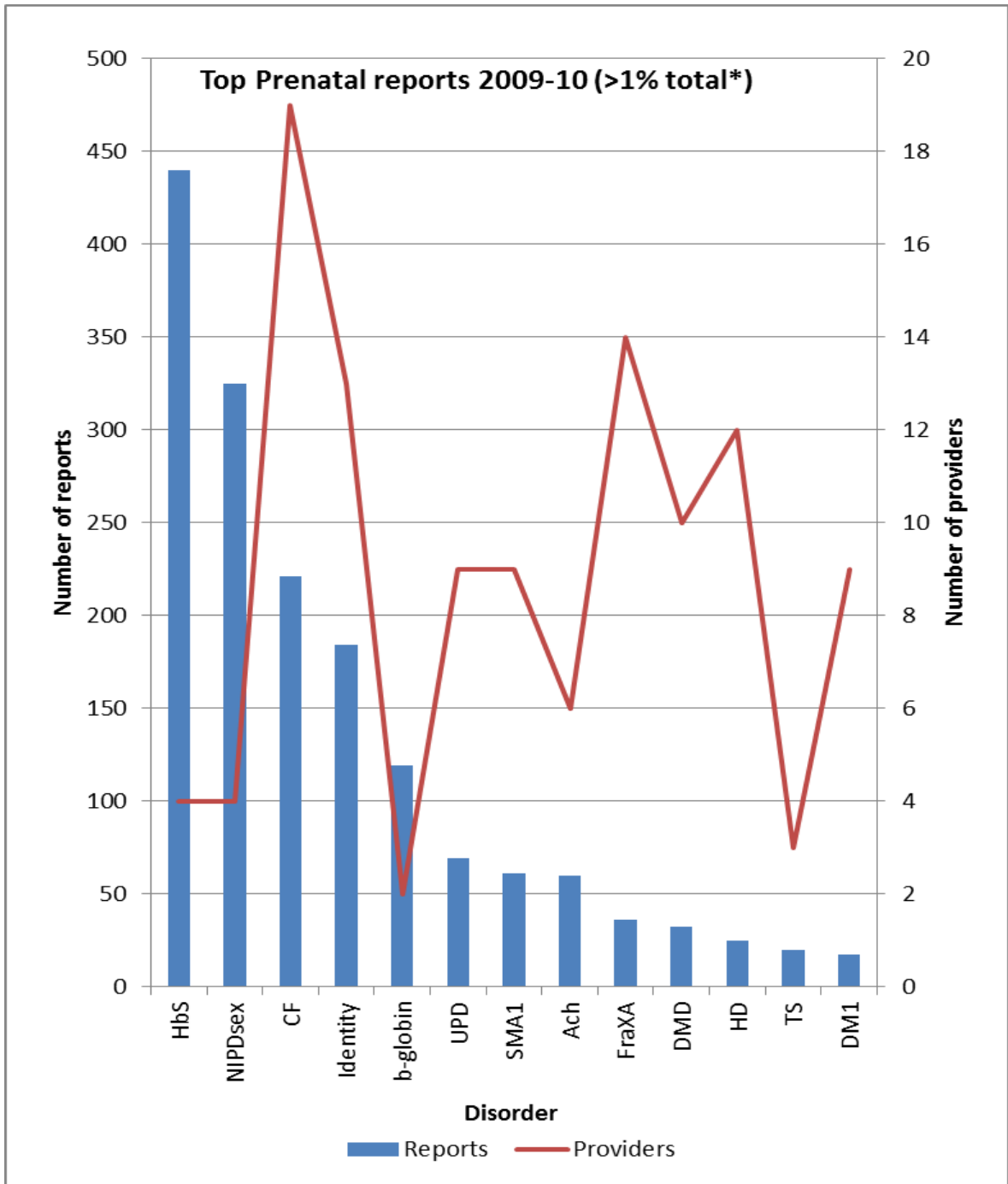
The largest activity (86%) was for the common aneuploidies reported by 6 laboratories.

The grand total for prenatal reports was 12389, up 47% on last year of (8399). Excluding aneuploidy, the total was 1744 that was also up 10% on the corresponding total for the previous year. The increase in aneuploidy activity reflects a change in the participants contributing data to the audit.

Non-invasive testing fetal sexing was reported by 4 laboratories and accounted in total for 19% of the non-aneuploidy screening reported prenatal activity.



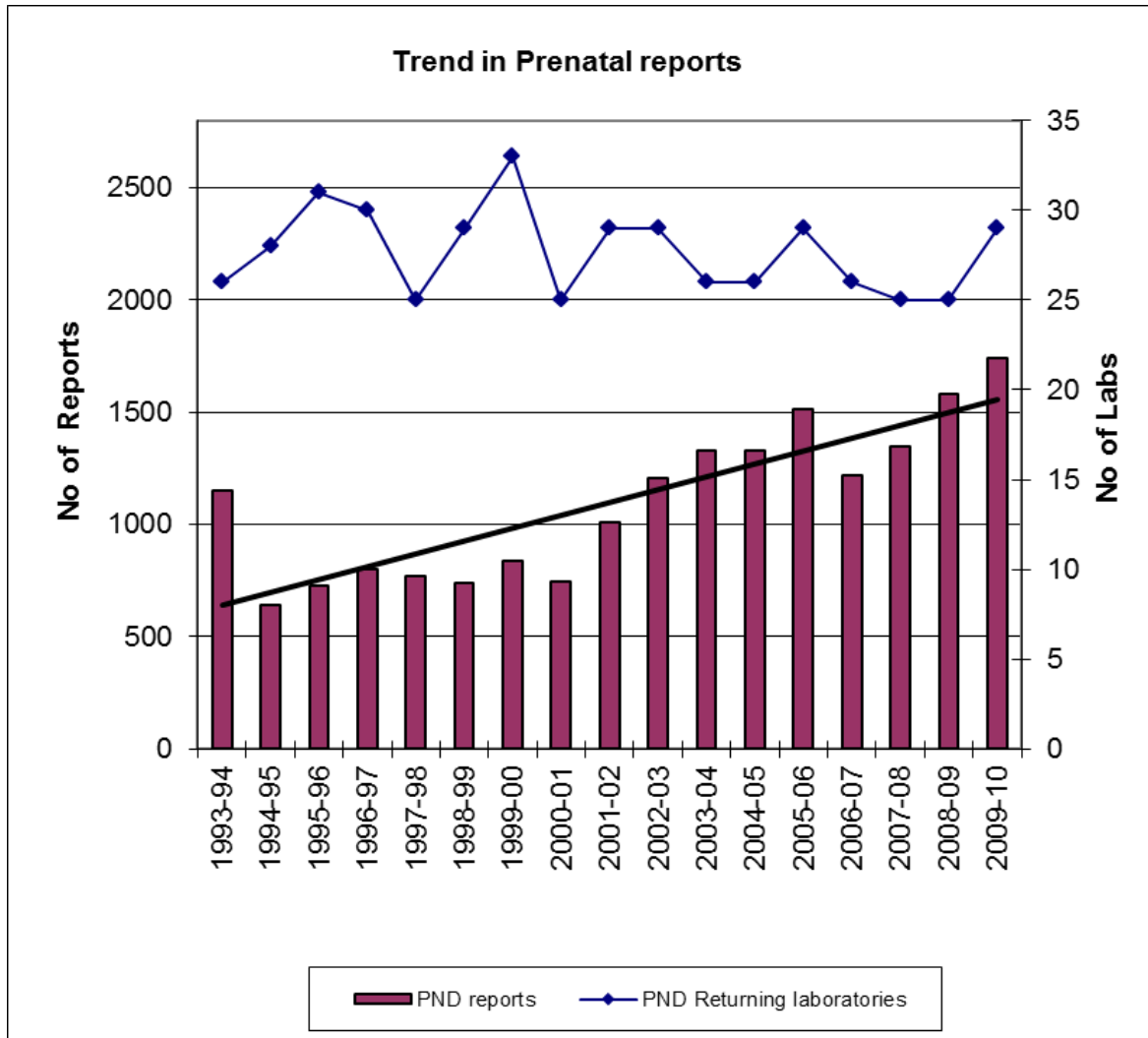
3.1 Chart of prenatal report activity excluding Qf-PCR (aneuploidy) MoLU data was not available from all laboratories.



3.2 Top (>1% activity excluding screening) prenatal requests together with the corresponding number of laboratories reporting that activity.

Year	Original total	No. of submitting CMGS labs	FREE FETAL DNA -sexing	SICKLE CELL ANEMIA	Corrected CF prenatals (1998-99 to 2007-08) included some FEEB tests	BETA THALASSAEMIA	SPINAL MUSCULAR ATROPHY, TYPE I; SMA1	ACHONDROPLASIA	PHENYLKETONURIA	RETARDATION 1 GENE; EMB4	DYSTROPHY, DUCHENNE TYPE;	UPD all	HUNTINGTON DISEASE	CRANIOSYNOSTOSIS	DYSTROPHIA MYOTONICA 1	ALPHA THALASSAEMIA	TUBEROUS SCLEROSIS	POLYCYSTIC KIDNEY DISEASE; AUTOSOMAL RECESSIVE	THANATOPHORIC DYSPLASIA; TD	ADRENAL HYPERPLASIA, CONGENITAL, DUE TO 21-HYDROXYLASE DEFICIENCY	AS/PWS	TREACHER COLLINS	ADRENOLEUKODYSTROPHY; ALD	HEMOPHILIA A	ABNORMAL HAEMOGLOBINS	
1993-94	1154																									
1994-95	640																									
1995-96	726																									
1996-97	802																									
1997-98	769																									
1998-99	729	29		48	144	37		10	37	70	9	35			30	6		3		21	11			4		
1999-00	840	33		53	137	52	68	14	42	63	10	25	9		22	9				19	10			8		
2000-01	746	25			190		39	15	41	74	8	25	4		36	5		9		13	13		1	2		
2001-02	1011	29			184		27		56	65		26			40						11					
2002-03	1208	29		168	192	77	51	36	47	74	20	16	7		30	4	1	15	6	13	17		3	4		
2003-04	1615	26		194	147	43	56	41	46	93	28	36	13		36	4	7	21	10	11	23	2	3	1		
2004-05	1327	26		226	141	95	32	37	45	85	22	24	8		27	8	6	16	17	21	17		4	8	40	
2005-06	1511	29		303	212	112	45	37	43	65	34	39	17		24	7	10	19	10	29	10	2	10	3	79	
2006-07	1247	26	60	107	219	48	65	44	42	53	49	27	13		37	10	7	21	13	19	16	6	1	2		
2007-08	1347	25	149	102	305	45	67	42	50	38	54	32	14		38	6	15	16	12	19	8	7	8	2		
2008-09	1571	25	358	199	159	74	62	51	48	48	37	26	19		16	16	14	13	12	12	11	9	7	2		
2009-10	1744	29	325	440	221	119	61	60	36	32	68	25	9		17	12	20	14	7	13	11	2	4	0	0	

Table summarising prenatal diagnosis activity for single gene disorders



3.4 Chart to show the trend in prenatal report numbers (excluding aneuploidy screening) with corresponding number of laboratories submitting data over past 17 years. This was up 10% on previous year of 2008-9 that may reflect the difference in the participating labs and the wider use of non-invasive prenatal diagnosis using cell free fetal DNA.

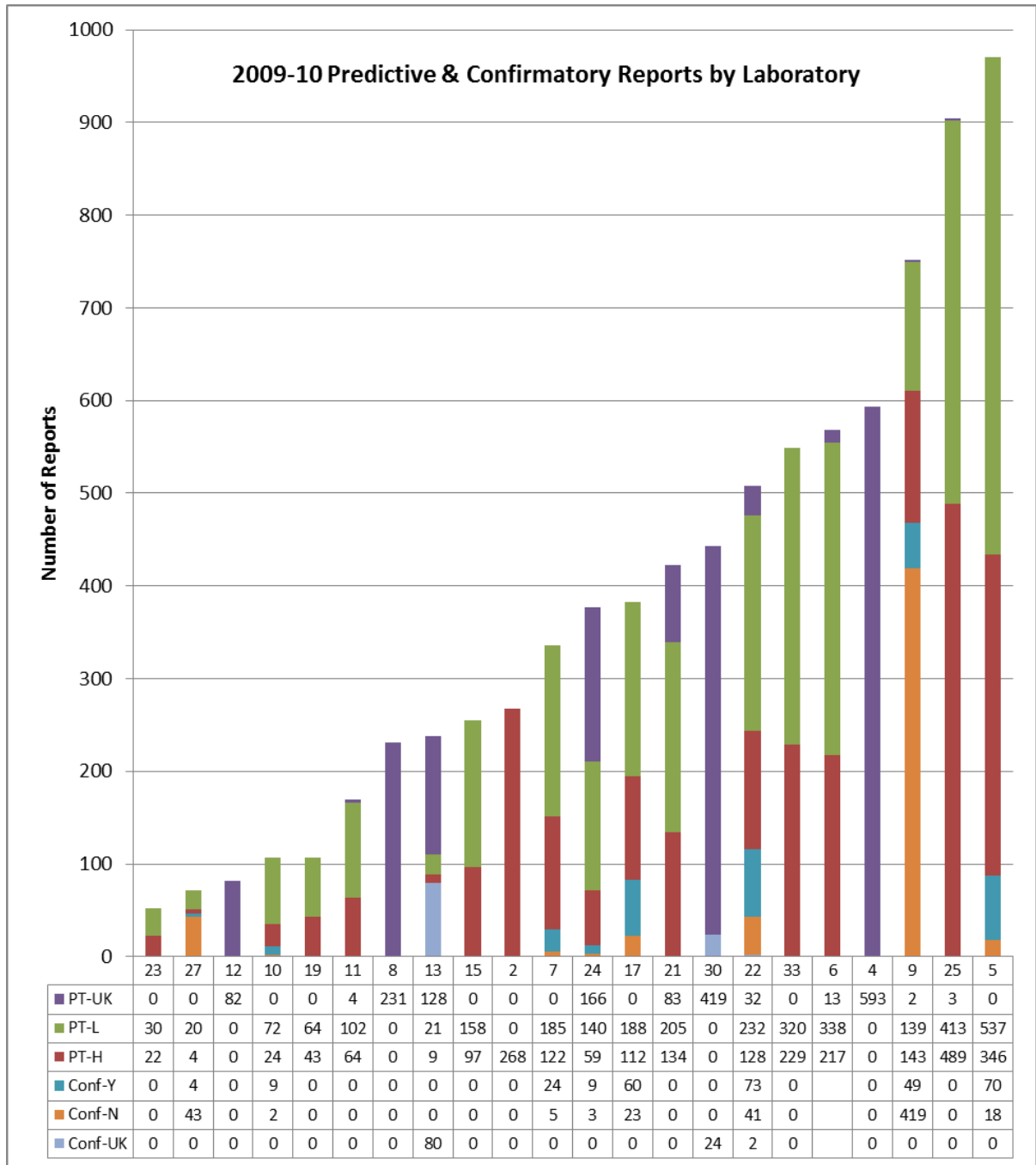
4. PREDICTIVE TESTS 2009-10

The CMGS Laboratories have historically shared data on the number of predictive and confirmatory tests with various professional bodies in support of the moratorium on the use of genetic tests for insurance. These bodies include the DH, Association of British Insurers (ABI), Genetics and Insurance Committee (GAIC) and the Human Genetics Commission - Monitoring Group on Genetics and Insurance (MGGI).

Some laboratories were unable to provide a full break down of the data. Categories are presented by predictive tests undertaken in asymptomatic relatives and confirmatory tests undertaken in symptomatic relatives to confirm the molecular diagnosis within the family. It is possible that some of the data in the last category includes some misclassified with segregation and exclusion tests.

2009-10	2008-9	Var	Category
22	21	5%	Number of lab returns
128	129	-1%	Number of disorders
1756	1404	25%	Unknown outcome of predictive tests
106	261	-59%	Unknown outcome of confirmatory tests
2510	1945	29%	High risk predictive
3164	2738	16%	Low risk predictive
288	577	-50%	Yes-Confirmatory test
521	484	8%	No-Confirmatory test
8345	7409	13%	Grand Total (predictive & confirmatory)

4.1. Summary table of predictive and confirmatory tests in 2009-10



4.2 Breakdown by returning labs

Key

PT-UK is the unknown outcome of predictive tests.

PT-L refers to low risk (mutation absent) predictive/presymptomatic test

PT-H refers to high risk (mutation present) predictive/presymptomatic test

Conf-Y refers to the confirmed presence of the mutation

Conf-N refers to the mutation being absent i.e. not present

Conf-UK is the unknown outcome of confirmatory tests

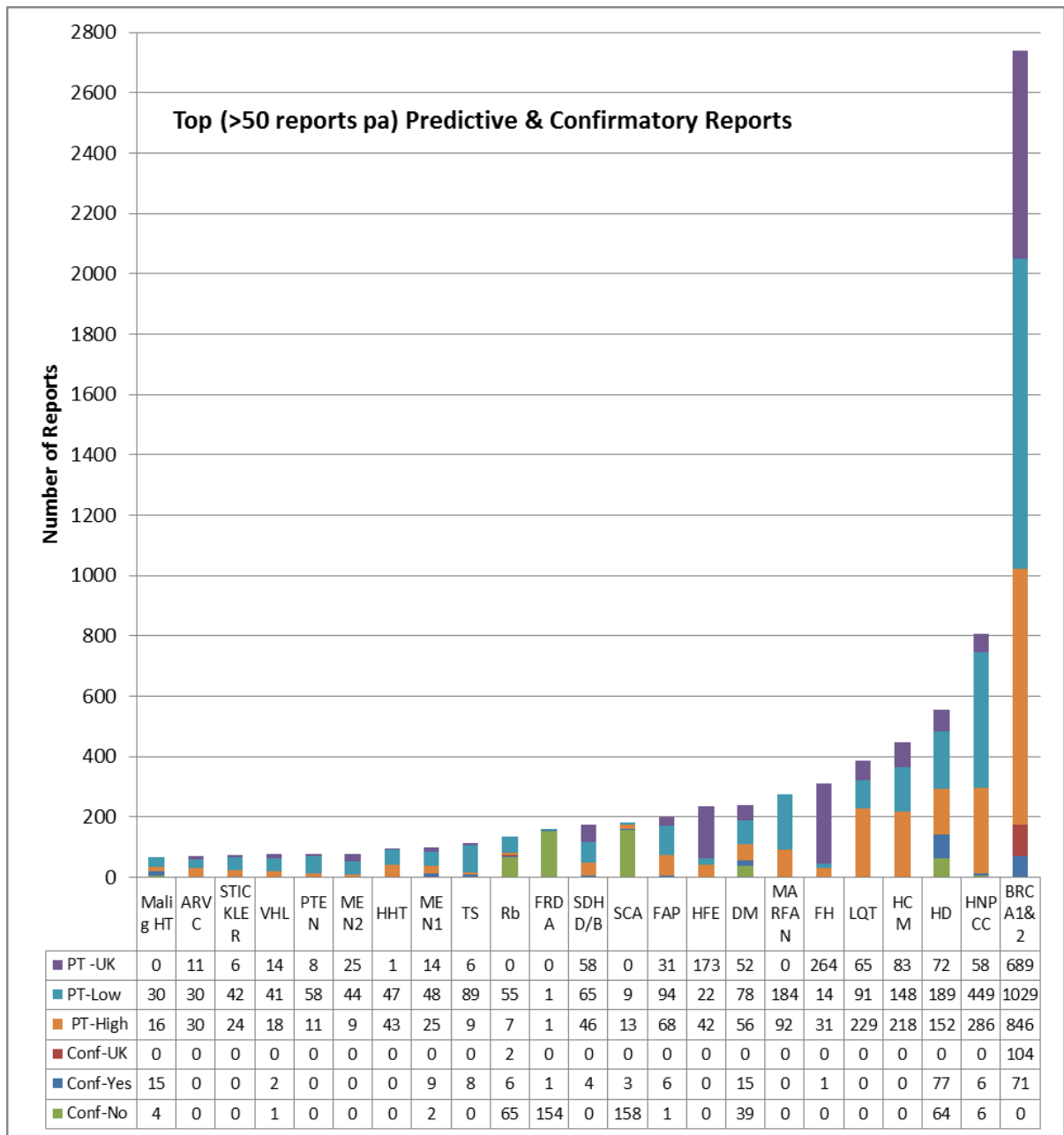


Figure 4.3
Breakdown of top (>50 pa) predictive/confirmatory tests by disorder

Disease codes are: Malig HT (malignant hypertension, ARVC (Arrhythmogenic Right Ventricular Cardiomyopathy), STICKLER, VHL (Von Hippel Lindau syndrome), PTEN (Phosphatase and Tensin Homolog deleted On Chromosome 10), MEN2 (multiple endocrine neoplasia type 2, HHT Hereditary haemorrhagic telangiectasia, MEN1 (multiple endocrine neoplasia type 1), TS (tuberous sclerosis), Rb (retinoblastoma), FRDA (Friedreich ataxia), SDHD/B (succinate dehydrogenase subunit complex B), SCA (spinocerebellar ataxia), FAP (familial adenomatous polyposis coli), HFE (hemochromatosis), DM (myotonic dystrophy), Marfan syndrome, FH (familial hypercholesterolemia), LQT (long QT), HCM (hypertrophic cardiomyopathy), HD (Huntington's disease), HNPCC (hereditary non polyposis coli), BRCA1/2 (breast & ovarian cancer),

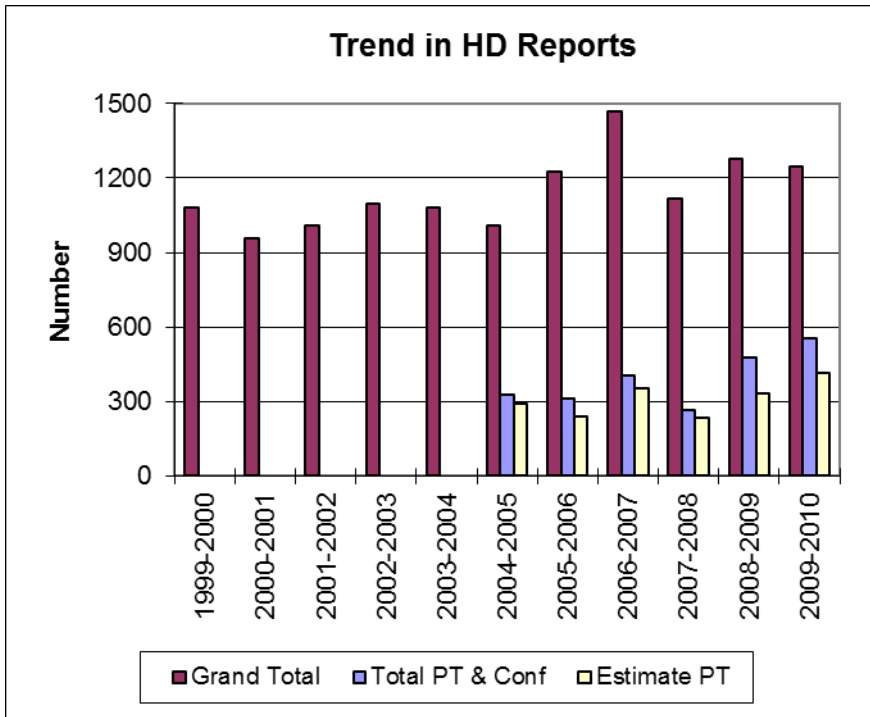
Comparison with activity in previous years

CMGS data	1999 2000	2000 2001	2001 2002	2002 2003	2003 2004	2004 2005	2005 2006	2006 2007	2007 2008	2008 2009	2009 2010
No. HD lab returns					17	15	13	11	13	10	9
No. BRCA lab returns					18	18	13	13	14	10	15
Grand Total HD tests	1080	956	1010	1099	1079	1009	1226	1469	1118	1278	1245
Total PT&Conf for HD						328	311	407	268	478	554
Grand Total BRCA tests	1740	1889	2469	3142	4635	4455	5309	6906	9048	7041	7733
Total PT&Conf for BRCA						1124	1746	2548	2068	2141	2739
Estimate predictive of total HD tests (%)						328 (33%)	242 (20%)	354 (24%)	237 (21%)	330 (26%)	413 (33%)
Estimate predictive of total BRCA tests (%)					20%	939 (21%)	1021 (19%)	1393 (20%)	1949 (21%)	1890 (27%)	2564 (33%)

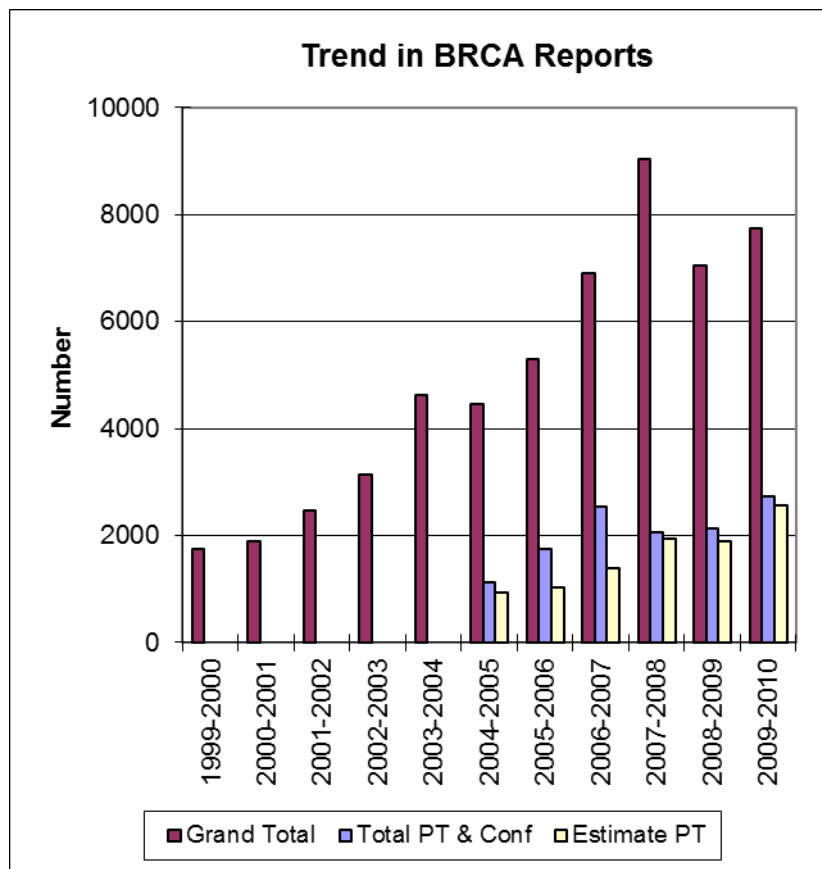
Comments

The 2009-10 shows an increase in predictive HD reports, up 25% on previous year. The estimated percentage of predictive tests of total HD reports shows an increase to around 33%.

The total number predictive BRCA test reports is also up 35% on last year and the estimated percentage of predictive tests of total BRCA reports also shows an increase to around 33%.

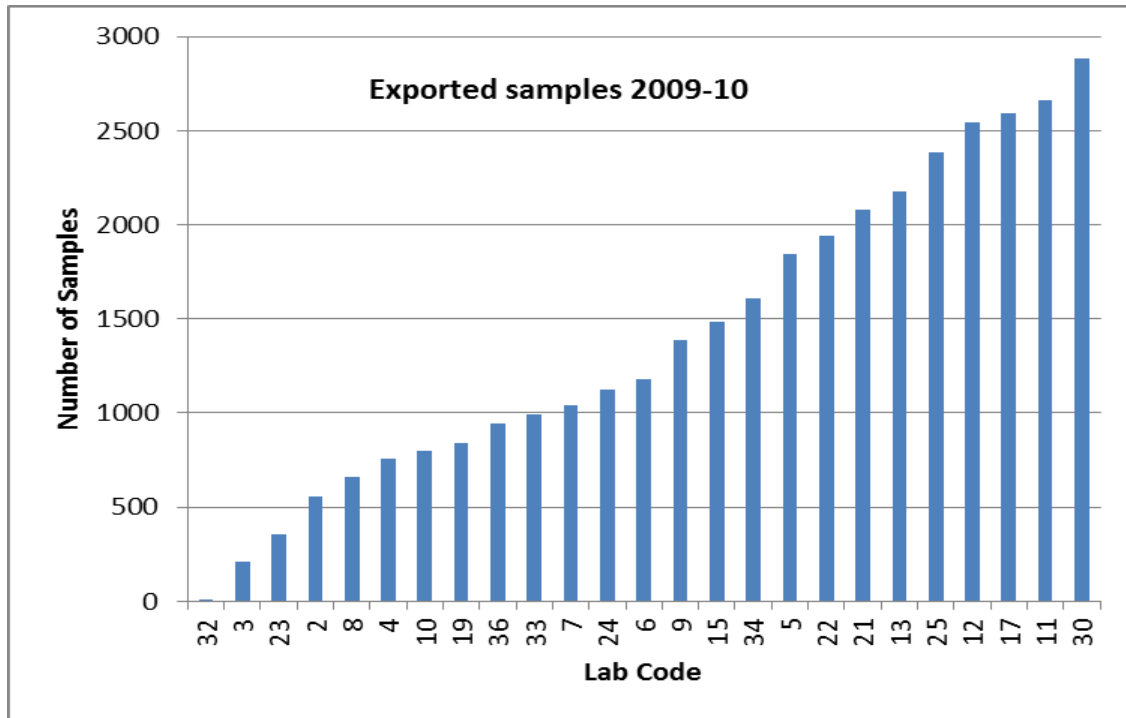


4.3 Trend in test activity for Huntington disease to show total (grand total), number of predictive (PT) and confirmatory (Conf) tests

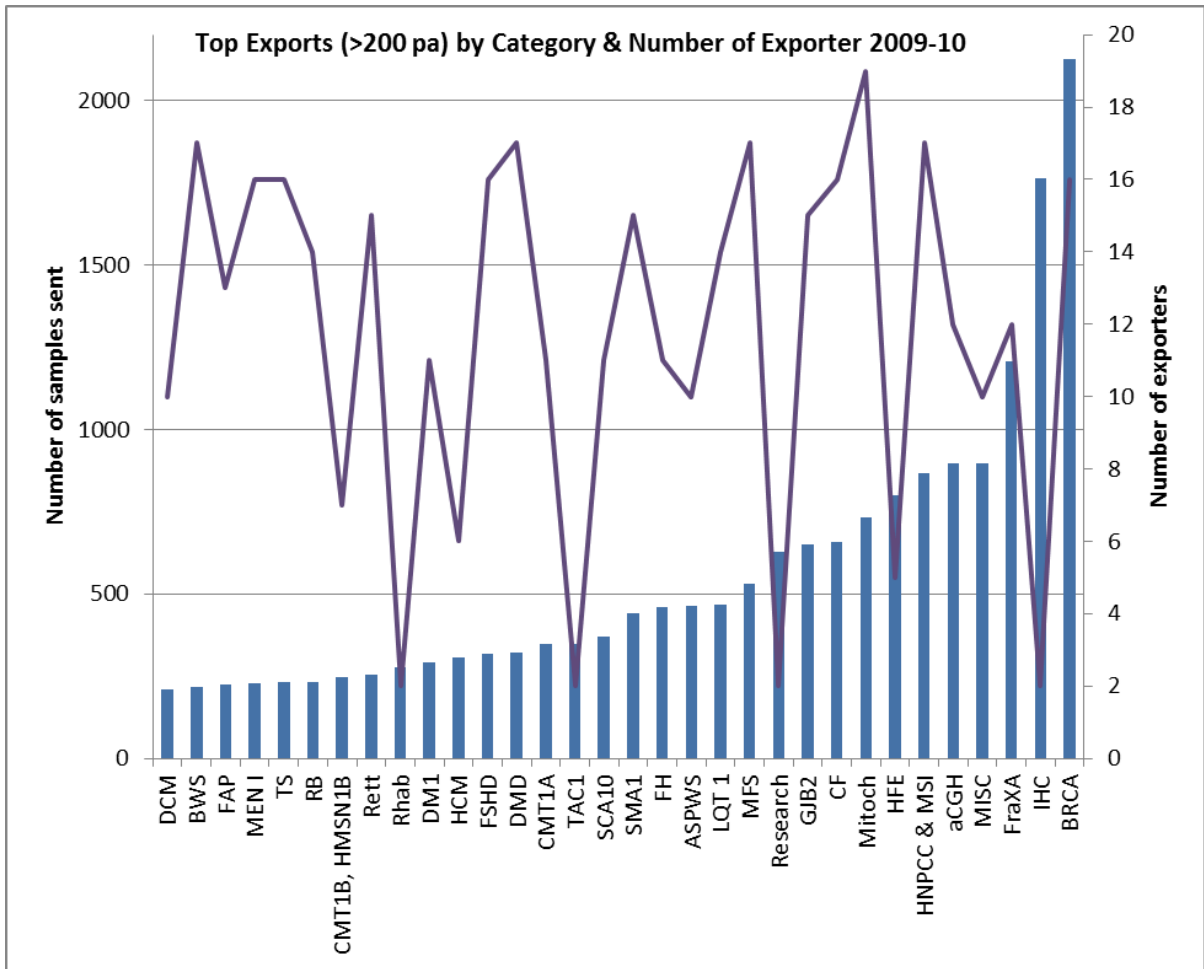


4.5 Trend in test activity for breast/ovarian cancer to show total (grand total), number of predictive (PT) and confirmatory (Conf) tests

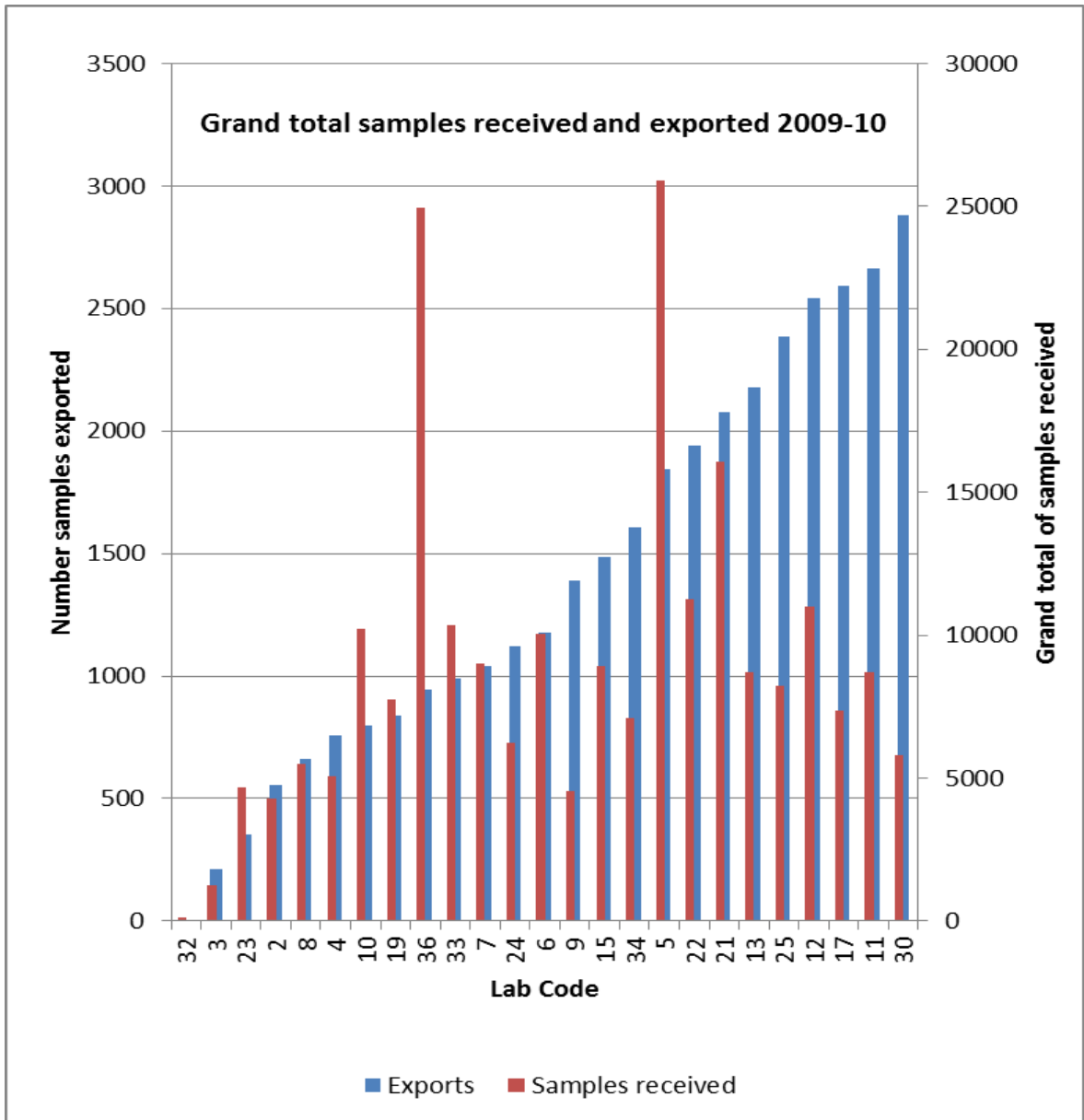
5. EXPORTS



5.1 Total exports reported was 35036 that was 23% more than for the previous year. This ranged from 1 to 2880 sends per lab as reported by 25 respondents.

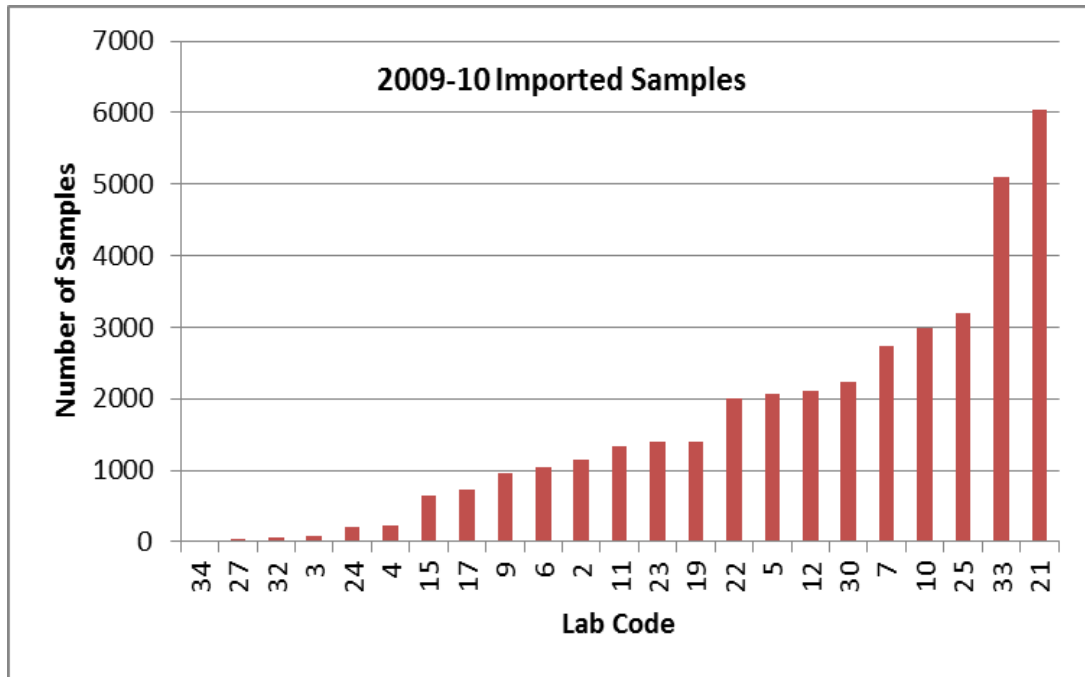


5.2 Number of top exports (sends), over 200 per year by disease showing number of laboratories exporting that test.
 Note most but not all of the above 25 laboratories in chart 5.1 provided a breakdown by disease of the export activity.



5.3 Chart to show combined export and sample received activity by individual laboratories ranked by number of exported samples

6. IMPORTS 2009-10



6.1 Summary of total samples received from other laboratories (imports) that ranges from 2 to 6041 samples across the individual 23 responding labs and extends over 342 different tests.

The total declared imports is 37777, similar to the previous year and compares with a reported sum of 34091 exported samples.

Note that some samples will be sent and received from outside the CMSG labs, both within and outside the UK

7 REPORTING TIMES

The data is presented in the table below that focuses on the larger volume tests and those tests provided by several centres for which there is more meaningful data. Some of the work provided by the specialist labs is included to allow these laboratories to benchmark their reporting times.

The data includes a summary of the average reporting times and the percentage compliance of reports for each of the three White Paper reporting time targets.

Note

Zero figures in the returns were excluded as it was unclear if this referred indicated no compliance or lack of data.

Some laboratories have used calendar rather than working days.

Some Laboratories may have categorized complex and routine analyses according to different criteria

7.1 Summary table of average reporting times between labs for top referrals from 2006/7 to 2009/10

Disease	gene	Average															%											
		Routine complex	Routine complex	Routine complex	Routine complex	Routine complex	Routine simple	Routine simple	Routine simple	Routine simple	Routine simple	Urgent	Urgent	Urgent	Urgent	Urgent	Routine complex	Routine complex	Routine complex	Routine complex	Routine simple	Routine simple	Routine simple	Routine simple	Urgent	Urgent	Urgent	Urgent
		Median	Min	Max	Mean	count	Median	Min	Max	Mean	count	Median	Min	Max	Mean	count	Median	Min	Max	Mean	Median	Min	Max	Mean	Median	Min	Max	Mean
CYSTIC FIBROSIS	CFTR	28	5	41	24	5	6	2	13	6	16	2	1	4	2	17	82	43	100	78	95	70	100	93	100	79	100	96
2008-9		20	6	38	23	5	6	3	15	7	17	2	2	4	3	13	99	59	100	91	96	1	100	90	97	1	100	87
2007-8		54	24	124	64	4	6	3	19	7	18	3	1	4	3	15	58	38	80	58	93	1	100	80	86	1	100	73
2006-7		22	9	32	21	3	8	4	34	9	15	3	2	6	3	15	100	81	100	94	94	36	100	87	82	31	100	80
2005-6		13	8	35	19	3	9	4	48	13	19	4	2	14	5	18												
2004-5		28			27	4	12			11		3			4	19												
DYSTROPHIA MYOTONICA 1	DMPK	29	8	158	56	4	8	6	12	9	11	2	2	3	2	9	100	8	100	80	86	61	99	85	100	1	100	87
2008-9		23	11	46	26	6	9	6	16	9	9	3	2	8	3	9	89	33	100	78	87	44	98	77	100	50	100	83
2007-8		27	16	101	38	9	9	6	39	13	11	3	1	7	3	8	74	1	100	57	65	1	100	63	50	0	100	55
2006-7		23	21	51	28	5	10	6	20	11	9	3	2	6	3	7	86	50	98	82	62	13	91	60	97	50	100	84
2005-6		37	23	55	38	6	19	6	11	26	11	6	2	19	7	10												
2004-5		24			24	2	21			26	17	9			11	12												
FRAGILE SITE MENTAL RETARDATION 1 GENE; FMR1	FMR1	33	20	109	39	11	10	6	16	10	15	7	2	15	7	6	80	29	97	76	84	25	96	80	100	10	100	100
2008-9		29	18	42	29	12	11	6	20	11	13	3	3	10	5	4	84	1	100	74	11	6	20	11	100	1	100	72
2007-8		30	15	137	39	15	12	4	16	11	15	3	2	9	4	6	72	0	100	55	59	0	100	53	100	9	100	50
2006-7		31	14	76	41	13	13	4	22	13	13	4	3	7	4	5	71	26	100	68	61	1	99	52	84	33	100	75
2005-6		43	13	86	45	12	21	5	58	23	13	9	5	24	10	11												
2004-5		51			52	4	20			24	17	8			11	13												
HUNTINGTON DISEASE	IT15	38	38	38	38	1	9	7	18	10	10	2	1	3	2	8	100	100	100	1	90	23	100	80	100	1	100	88
2008-9		8	0	15	8	2	8	5	43	11	12	2	1	14	4	8	98	98	98	98	82	1	100	73	100	0	100	60
2007-8		13	9	13	11	3	10	3	19	10	13	2	2	5	3	8	100	0	100	50	74	1	100	62	100	0	100	76
2006-7		12	12	12	12	1	11	8	15	11	12	2	1	4	2	8	100	100	100	100	71	1	95	60	100	40	100	93
2005-6		16	166	166	166	1	16	7	38	17	15	4	2	13	5	10												
2004-5		20			20	2	18			19	16	8			10	14												

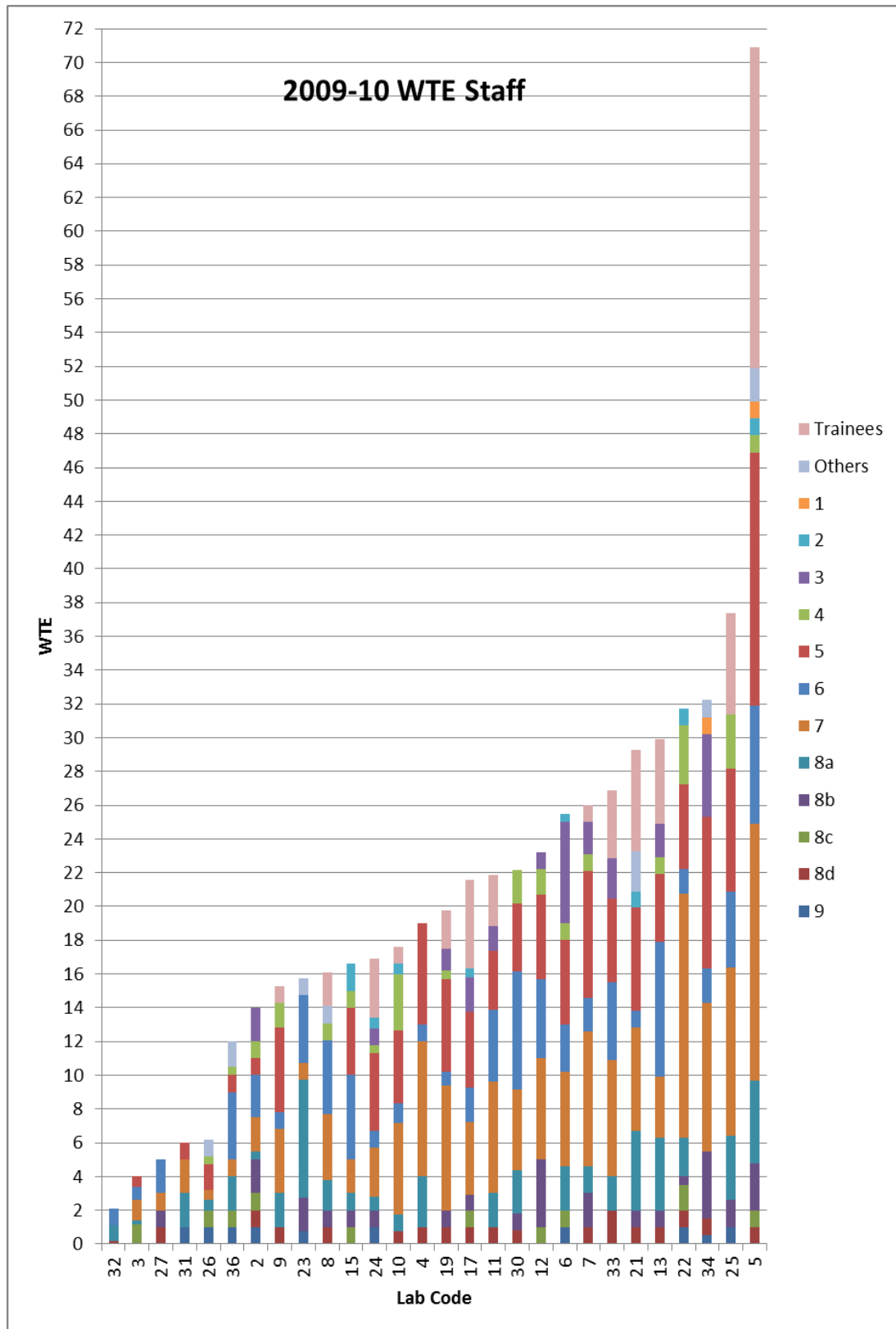
Disease	gene	Average															%												
		Routine complex	Routine complex	Routine complex	Routine complex	Routine complex	Routine simple	Routine simple	Routine simple	Routine simple	Routine simple	Urgent	Urgent	Urgent	Urgent	Urgent	Routine complex	Routine complex	Routine complex	Routine complex	Routine simple	Routine simple	Routine simple	Routine simple	Urgent	Urgent	Urgent	Urgent	
		Median	Min	Max	Mean	count	Median	Min	Max	Mean	count	Median	Min	Max	Mean	count	Median	Min	Max	Mean	Median	Min	Max	Mean	Median	Min	Max	Mean	
MUSCULAR DYSTROPHY, DUCHENNE TYPE; DMD	Dystr ophin	30	28	55	38	3	11	6	33	15	10	3	1	4	3	8	86	31	94	70	78	19	100	67	90	60	100	86	
2008-9		35	12	76	39	4	12	8	20	12	10	3	1	5	3	9	72	6	90	60	57	27	88	58	100	0	100	86	
2007-8		53	15	143	71	5	11	1	28	12	12	2	1	4	2	10	37	0	88	41	64	1	100	61	100	3	100	79	
2006-7		67	34	602	168	5	16	7	33	17	9	2	1	5	3	10	59	33	96	62	46	24	87	48	100	33	100	82	
2005-6		42	18	319	99	5	23	7	55	26	13	4	1	7	4	11													
2004-5		74			86	6	26			35	14	3			5	13													
AS&PWS (all)	SNRP N	49	40	59	49	2	15	8	35	18	4	2	2	2	2	1	53	53	53	53	71	50	96	72	100	10	0	100	100
2008-9		30	4	33	23	5	11	6	22	12	9	3	3	4	3	2	75	1	97	65	71	1	95	63	1	0	25	9	
2007-8		26	9	56	29	6	11	4	20	11	10	8	2	9	7	4	72	1	100	62	53	1	95	53	100	10	0	100	100
2006-7		44	29	53	42	3	13	7	29	14	11	4	2	9	5	4	81	69	83	77	56	29	94	58	100	25	100	81	
2005-6		58	34	372	122	8	17	6	27	16	17	3	3	19	8	5													
2004-5		44			52	4	25			46	26	8			11	15													
HEMOCHROMA TOSIS; HFE	HFE						12	6	14	11	11										87	34	99	78					
2008-9							10	8	20	11	10										79	36	97	74					
2007-8		23	23	23	23	1	11	5	21	11	12						99	99	99	99	75	0	98	63					
2006-7							8	6	17	10	7										86	27	95	72					
2005-6							13	5	27	16	17	13	13	13	13	1													
2004-5							14			17	12																		
CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 1A; CMT1A	PMP 22	24	22	39	28	3	13	10	25	15	5						92	50	100	80.7	68	41	93	65					
2008-9		27	40	32	4	83	12	7	36	15	6	19	14	23	19	2	83	66	100	83	43	3	88	47	50	0	100	50	
2007-8							10	7	16	11	5										70	40	92	65					
2006-7		29	23	36	30	2	16	14	32	20	5						63	41	85	63	39	17	56	37					
2005-6							21	12	54	26	8	7	3	12	7	2													
2004-5		79			80	4	26			31	11	15			12	4													

Disease	gene	Average															%												
		Routine complex	Routine complex	Routine complex	Routine complex	Routine complex	Routine simple	Routine simple	Routine simple	Routine simple	Routine simple	Urgent	Urgent	Urgent	Urgent	Urgent	Routine complex	Routine complex	Routine complex	Routine complex	Routine simple	Routine simple	Routine simple	Routine simple	Urgent	Urgent	Urgent	Urgent	
		Median	Min	Max	Mean	count	Median	Min	Max	Mean	count	Median	Min	Max	Mean	count	Median	Min	Max	Mean	Median	Min	Max	Mean	Median	Min	Max	Mean	
SPINAL MUSCULAR ATROPHY, TYPE I; SMA1	SMN	25	25	25	25	1	9	7	11	9	7	3	2	5	3	9	100	100	100	100	84	63	100	81	93	50	100	87	
2008-9		61	61	61	61	1	8	6	12	8	7	3	2	5	3	6					81	56	92	79	100	14	100	81	
2007-8		30	27	32	30	2	8	5	15	9	9	4	2	6	4	7	89	78	100	89	78	50	96	74	70	6	100	63	
2006-7		23	20	27	23	2	10	6	11	10	6	3	2	6	3	7	100	100	100	100	69	47	98	70	94	13	100	72	
2005-6		46	46	46	46	1	12	4	22	12	8	4	3	8	5	7													
2004-5		18			18	2	13			14	10	5			11	8													
FAMILIAL BREAST & OVARIAN CANCER	BRCA 1, BRCA 2	48	22	617	110	10	11	7	26	12	12	2	2	2	1	55	1	100	51	77	1	94	66	100	10	0	100	100	
2008-9		42	20	356	89	10	12	6	13	11	10					72	7	100	61	74	1	95	63	62		62	62		
2007-8		37	23	381	107	9	11	6	62	15	12					41		100	39	59		100	62						
2006-7		83	24	357	105	10	12	8	17	12	10	1	1	19	7	3	36	9	100	42	64	29	87	62	100	10	0	100	100
2005-6		17	8	76	575	256	11	20	7	63	24	13	7	5	10	7	2												
2004-5		12	3			197	10	24			97	9	17			32	8												
FAMILIAL ADENOMATOUS POLYPOSIS	APC	36	31	142	53	7	9	5	55	14	10					50	19	88	54	91	0	100	75						
2008-9		40	8	238	68	6	11	7	17	12	7					66	6	100	57	83		95	78						
2007-8		63	22	334	130	6	11	8	44	15	10					45	6	100	49	64		100	63						
2006-7		53	34	309	125	6	12	6	25	13	7	2	2	2	2	1	62	20	100	61	55	3	100	55	100	10	0	100	100
2005-6		19	6	50	131	4	322	8	25	6	76	28	10	9	7	12	9	2											
2004-5		89			110	8	27			48	7	17				17	4												

Disease	gene	Average															%													
		Routine complex	Routine complex	Routine complex	Routine complex	Routine complex	Routine simple	Routine simple	Routine simple	Routine simple	Routine simple	Urgent	Urgent	Urgent	Urgent	Urgent	Routine complex	Routine complex	Routine complex	Routine complex	Routine simple	Routine simple	Routine simple	Routine simple	Urgent	Urgent	Urgent	Urgent		
		Median	Min	Max	Mean	count	Median	Min	Max	Mean	count	Median	Min	Max	Mean	count	Median	Min	Max	Mean	Median	Min	Max	Mean	Median	Min	Max	Mean		
COLORECTAL CANCER, HEREDITARY NONPOLYPOSIS; HNPCC	MSH2 MLH1 MSH6 PMS2 MSI	46	22	180	72	10	9	3	50	4	62	11					84	7	100	68	85	9	100	74						
2008-9		47	37	220	72	8	9	7	18	10	8						77	2	100	63	78	17	97	69						
2007-8		57	29	361	111	9	11	7	98	24	10						62	0	91	48	50	1	97	54						
2006-7		95	32	224	117	8	12	8	16	12	7	12	12	12	12	1	48	5	77	37	48	21	92	54	100	100	100	100		
2005-6		24	4	60	408	236	8	22	7	72	25	10																		
2004-5		15	2			179	7	76			154	6	17			44	5													
Combined SCAs		SCAs	29	29	29	29	1	16	12	89	30	6						84	84	84	84	51	36	94	58					
2008-9							19	19	19	19	1										55	55	55	55						
2007-8	45		30	60	45	2	14	14	14	14	1						93	86	100	93	75	75	75	75						
2006-7	20		20	20	20	1	14	10	30	16	5						55	9	100	55	50	20	89	54						
2005-6	30		30	30	30	1	37	10	11	8	48	8	19	11	14	13	3													
2004-5	97					142	6	20			26	24	11			11	6													
FACTOR V DEFICIENCY	F5	11	11	11	11	1	9	6	14	9	6	5	5	5	5	1	93	93	93	93	87	58	99	83	33	33	33	33		
2008-9							10	7	20	11	2										86	51	93	79						
2007-8							8	6	19	10	6										64	34	100	68						
2006-7							11	6	27	14	3										54	23	94	57						
2005-6							11	6	56	19	5	12	12	12	12	1														
2004-5							12			14	5																			

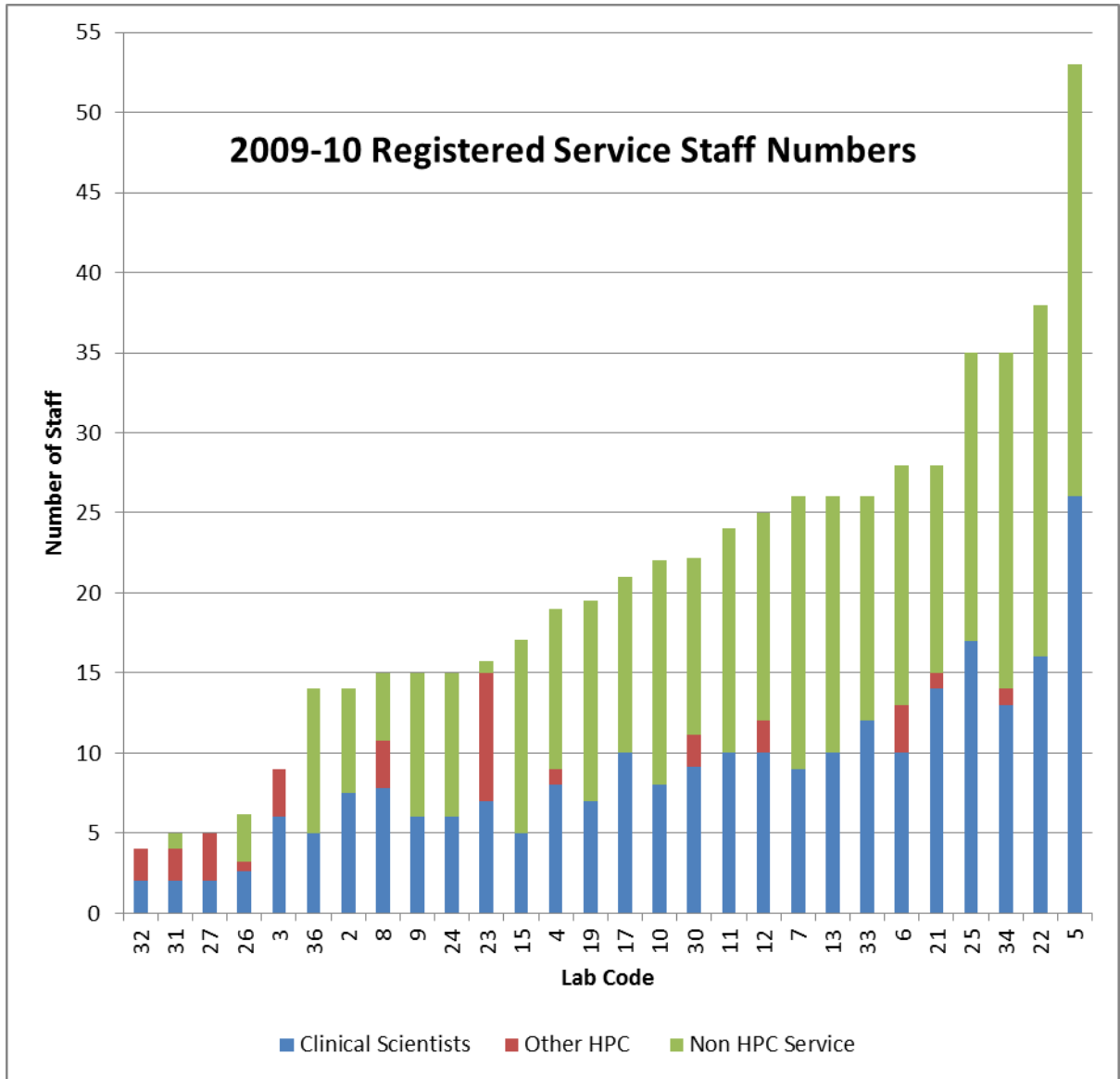
Disease	gene	Average															%											
		Routine complex	Routine complex	Routine complex	Routine complex	Routine complex	Routine simple	Routine simple	Routine simple	Routine simple	Routine simple	Urgent	Urgent	Urgent	Urgent	Urgent	Routine complex	Routine complex	Routine complex	Routine complex	Routine simple	Routine simple	Routine simple	Routine simple	Urgent	Urgent	Urgent	Urgent
		Median	Min	Max	Mean	count	Median	Min	Max	Mean	count	Median	Min	Max	Mean	count	Median	Min	Max	Mean	Median	Min	Max	Mean	Median	Min	Max	Mean
GAP JUNCTION PROTEIN, BETA-2	Cx26	32	18	35	28	7	12	8	25	14	5	2	2	2	2	1	86	1	99	75	70	38	100	69	100	100	100	
2008-9		26	20	31	25	4	18	8	22	17	4	1	1	1	1	1	89	1	99	66	19	0	100	33	1	1	1	1
2007-8		36	22	56	38	5	11	9	48	21	5	3	3	3	3	1	53	1	99	55	6	0	75	30	100	100	100	100
2006-7		70	32	128	75	4	33	13	89	42	4						59	7	85	52	44	11	73	43				
2005-6		11	6	72	159	116	2	50	6	13	8	61	4															
2004-5		11	1			111	2	55			61	5	5		5	1												
MITOCHONDRIAL MYOPATHY	Mt	33	27	38	33	2	16	9	42	22	3	3	3	3	3	1	93	92	94	93	25	21	89	45	100	100	100	100
2008-9		63	30	97	63	2	11	8	15	11	2						55	25	85	55	53	25	82	53				
2007-8		29	25	36	30	3	19	14	19	18	3	2	2	2	2	1	91	70	98	86	37	20	50	36	100	100	100	100
2006-7		48	48	48	48	1	16	6	18	56	4	5	5	5	5	1	47	47	47	47	39	29	50	39				
2005-6		33	14	53	33	2	18	12	92	35	4	10	10	10	10	1												
2004-5		56			56	2	55			53	4	4			4	2												
ABNORMAL HAEMOGLOBIN	a-globin, b-globin	22	15	28	22	2											93	87	100	93								
2008-9		25	25	25	25	1											87	87	87	87								
2007-8		26	26	26	26	1											79	79	79	79								
2006-7		28	28	28	28	1											82	82	82	82								
BETA THALASSAEMI A	b-globin	19	17	20	19	2	7	7	7	7	1	4	4	5	4	2	93	88	98	93	93	93	93	93	48	37	59	48
2008-9		24	24	24	24	1	17	4	30	17	2						80	80	80	80	71	53	88	71				
2007-8		25	25	25	25	1						3	3	3	3	1	85	85	85	85					64	64	64	64
2006-7		19	19	19	19	1											92	92	92	92								
RETINOBLASTOMA; RB1	RB1	46	35	57	46	2	7	6	8	7	2	2	2	12	5	3	47	21	72	47	89	86	92	89	94	89	100	94
2008-9		33	33	33	33	1	9	9	9	9	1	2	1	3	2	2	68	68	68	68	80	80	80	80	100	100	100	100
2007-8		65	14	108	63	4	16	11	20	16	2	3	2	3	3	2	44	13	100	34	51	22	79	51	83	67	100	83
2006-7		14	6	120	173	146	2	13	10	17	13	2	4	2	6	4	2	15	8	22	15	56	33	79	56	55	9	100

8. Staff



8.1 Staff configuration in whole time equivalents (WTE) and agenda for change (AfC) band for the 28 Laboratories providing data

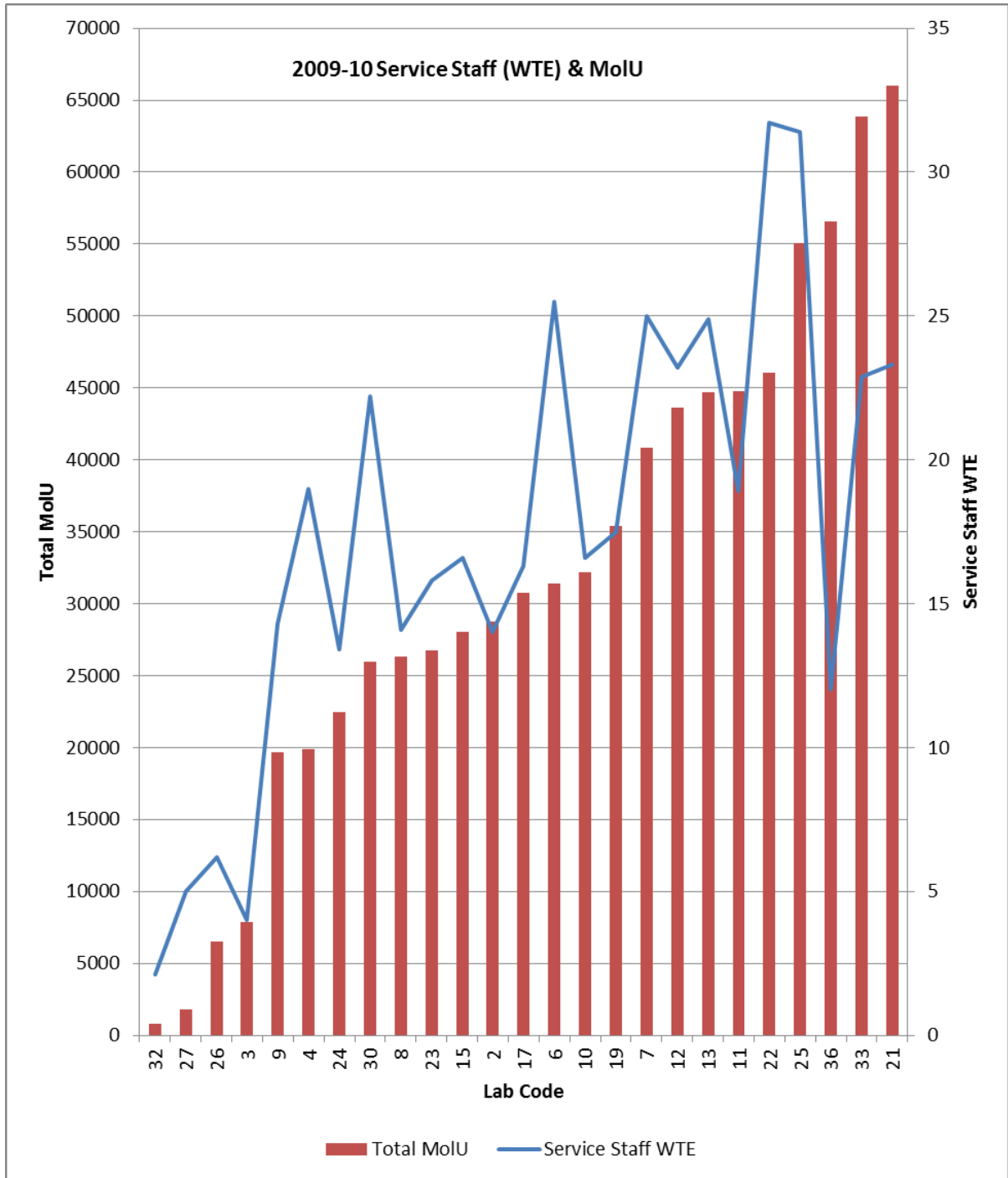
NOTE Trainee staff may be shared across both Molecular and Cytogenetic Laboratories



8.2 Configuration of Health Professions Council (HPC) registered staff of total number service staff

Registered staff accounted for 49% of the workforce that appears to be a 9% increase on previous year.

Note this excludes trainees and is the individual number not whole time equivalent of staff



8.3 Total laboratory activity by MoIU against service staff in WTE activity

25 Laboratories provided **pilot data** for the new MoIU activity units that show a median MoIU per lab of 30729 (range 781 to 66023).