ACGS Quality Subcommittee

ACGS Audit
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28/02/2014

Association of Clinical Genetics Science Audit of Data 2012-13

The Clinical Molecular Genetics Society (CMGS) have produced an annual audit of the genetic testing activity undertaken by the laboratories of its members since 1993. This includes data from the UK Regional Molecular Genetic Services plus some specialist services. This year's audit under the now merged CMGS and Association of Clinical Cytogenetics, ACGS, includes submissions from all the Regional Molecular Genetic Laboratories and the aneuploidy screening data from the Regional Cytogenetic Laboratories. The final submission was received on the 12th November 2013

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 and reporting times for specific diseases by service level,. Since 2005, the audit has included the names of the participating laboratories but the individual laboratory codes remain anonymous and change each year.

The collection and collation of data represents a considerable effort and many thanks are due to all the participating laboratories for the work involved in submitting this data and also for their willingness to share the information.

Gail Norbury
ACGS Quality Sub Committee

Participants in 2012-13 Audit

Aberdeen Regional Genetics Laboratory

All Wales Regional Genetics Laboratory (Cardiff)

Belfast Regional Genetics Laboratory

Birmingham Children's Hospital

Bristol Regional Genetics Laboratory

Cheshire & Merseyside Regional Genetics Laboratory (Liverpool)

Dundee Regional Genetics Laboratory

East Anglian Regional Genetics Laboratory (Cambridge)

Exeter Regional Genetics Laboratory

Leicester Cytogenetics Laboratory

London North East Thames Regional Genetics Laboratory (Great Ormond Street)

London North West Thames Regional Genetics Laboratory (Kennedy Galton)

London South East Regional Genetics Laboratory (Guys & St Thomas')

London South West Thames Regional Genetics Laboratory (St Georges)

London UCL Hospitals, Clinical Biochemistry Laboratory

London UCL Hospitals, Haemoglobinopathy Genetics Laboratory

London UCL Hospitals, Neurogenetics Laboratory (NHNN)

London Kings College Hospital

London Retinoblastoma Laboratory (Barts)

London The Doctors Laboratory

National Haemoglobinopathy Reference Laboratory (Oxford)

International Blood Group Reference Laboratory (Bristol)

North West Regional Genetics Laboratory (Manchester)

Northern Regional Genetics Laboratory (Newcastle)

Norfolk & Norwich Cytogenetics Laboratory

Nottingham Regional Genetics Laboratory

Oxford Regional Genetics Laboratory

Sheffield Regional Genetics Laboratory

South East Scotland Regional Genetics Laboratory (Edinburgh)

Wessex Regional Genetics Laboratory (Salisbury)

West Midlands Regional Genetic Centre (Birmingham)

West of Scotland Regional Genetics Laboratory (Glasgow)

Yorkshire Regional Genetics Laboratory (Leeds)

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.

1. SAMPLES RECEIVED AND EXTRACTED 2012-13

	Category	Total	Range		Labs	Fails	
		Sum	min	max	Count	Number	%
Samples	Blood	195127	2	19388	36		
	Amnio/CVS	18194	2	5266	34		
	Buccal	2032	1	486	24		
	Archive	32958	2	16847	24		
	Other	26116	12	4508	36		
	DNA	47225	6	3821	32		
	Research	22042	4	8007	20		
Extracts	Blood auto	178073	2	16213	29	1272	0.7%
	Blood manual	17430	34	3862	22	112	0.6%
	Amnio & CVB auto	5320	1	1382	16	12	0.2%
	Amnio & CVB manual	11776	2	5266	27	4	0.0%
	Buccal auto	2513	12	1125	14	19	0.8%
	Buccal manual	648	1	173	16	3	0.5%
	Archive auto	23571	1	16847	12	48	0.2%
	Archive manual	8929	1	5071	13		
	Other auto	15312	8	3850	21	75	0.5%
	Other manual	7467	4	2342	22	294	3.9%
	Research auto	7461	3	5041	10	15	0.2%
	Research manual	442	5	175	5	5	1.1%
Totals	Service samples	321652	103	33417	38		
	Service extracts	271039	36	30521	34	1859	0.7%
	Research samples	22042	4	8007	20		
	Research extracts	7903	9	5041	10	20	0.3%

1.1 Table Sample and Extraction summary

Notes

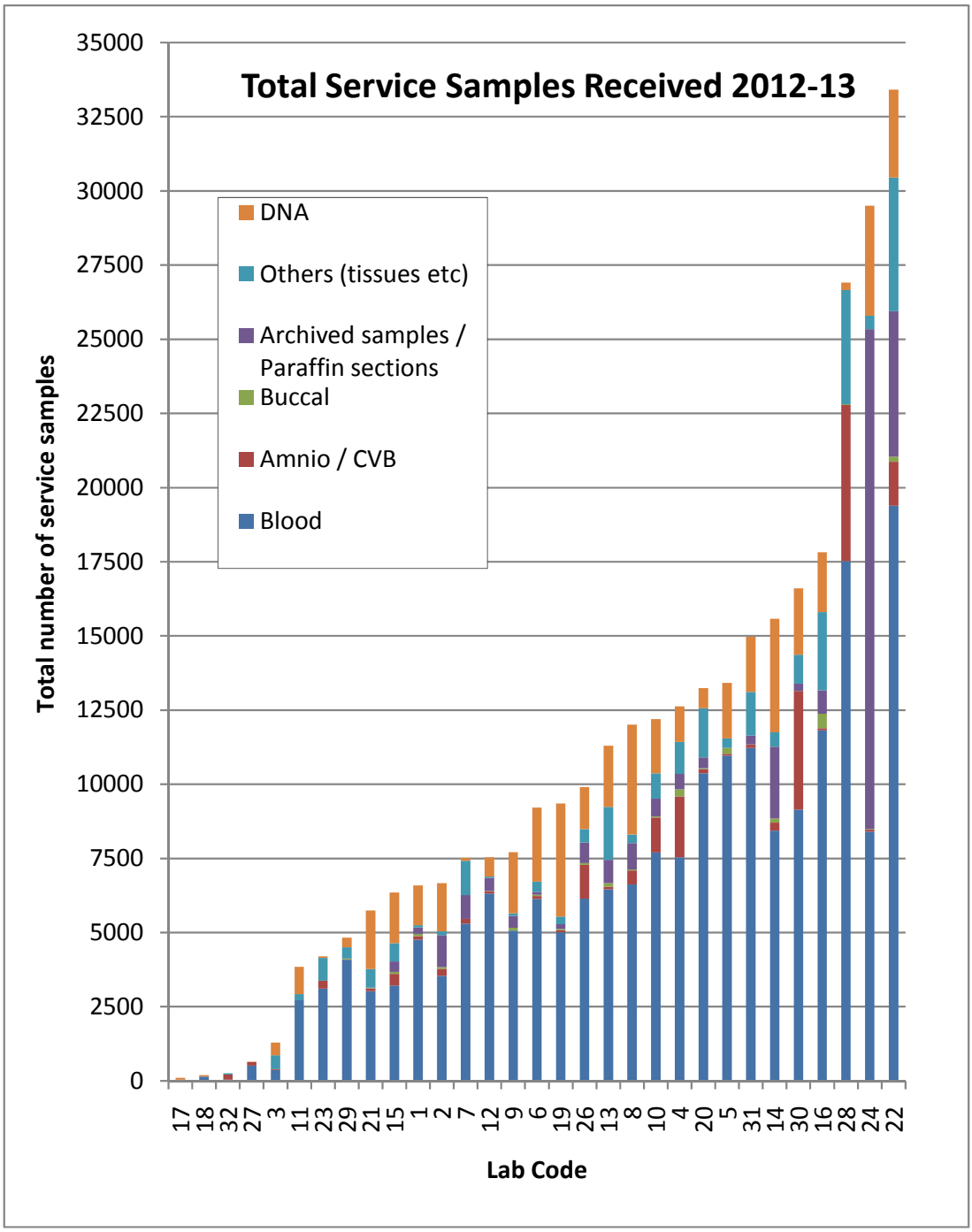
As there was not a clear distinction between the uses of a zero and no available submitted data, only positive figures were included in the analysis to determine minimum and fail-rate values.

The total number of service samples increased by 20%* on last year (321652 v 265600) that compares to an apparent 18% increase on the previous year.

The total number of service extractions increased by 23% (271039 v 220029) compared to the apparent 16% increase in the previous year.

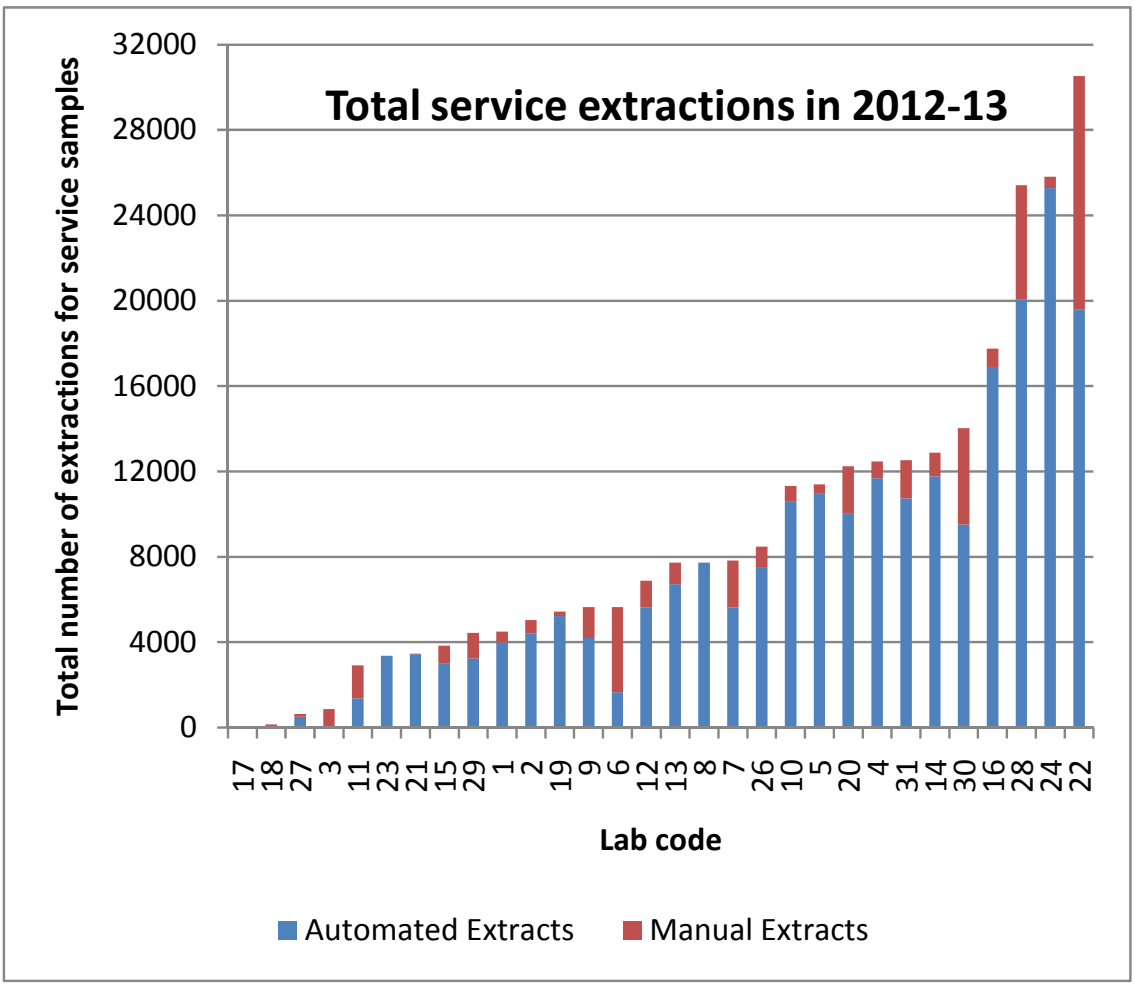
The major increase in activity reflects the increased samples (160%) and extraction (84%) of archive tissue for tumour analysis. There is also an approximate 50% increase in number of CVS/Amnio samples reflecting the inclusion of aneuploidy testing performed in cytogenetic laboratories. This activity has not previously been captured.

*excluding the additional aneuploidy activity included in this audit, the on year increase in sample number was 18% and in extraction number was 20%).



1.2 Chart showing total service samples received by each laboratory, ranked by the total and broken down by the different sample type.

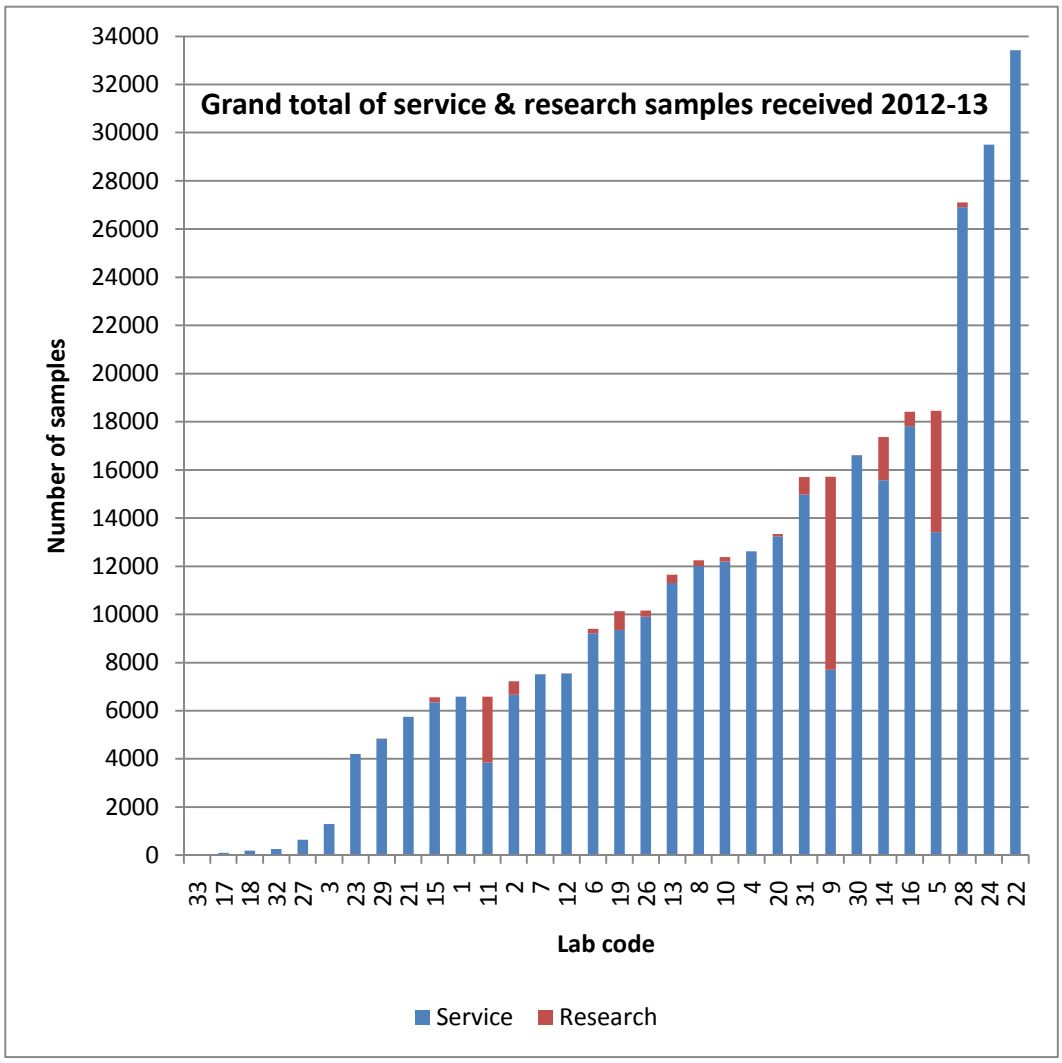
The separate returns for cytogenetic, molecular genetic laboratories of the same Genetics Centre have been combined for consistency.



1.3 Chart of total number of service sample extractions performed by manual and automated processes

There was a relative increase (503%) in the number of automated extractions of archive material. However, because the audit this year included the CVS/amnio samples from Cytogenetic Laboratories that tend to be extracted manually, the overall proportion of automated extractions remained at 83%, as for the previous year.

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, receipt of extracted DNA continues to account for ~15% service samples. Under the GenU workload system, a single unit of activity is accrued for extraction (and storage).



1.4 Chart of Grand total samples (service & research) received by each centre,

Service samples accounted for 94% of all samples as for the last three years

2 POSTNATAL REPORTS 2012-13

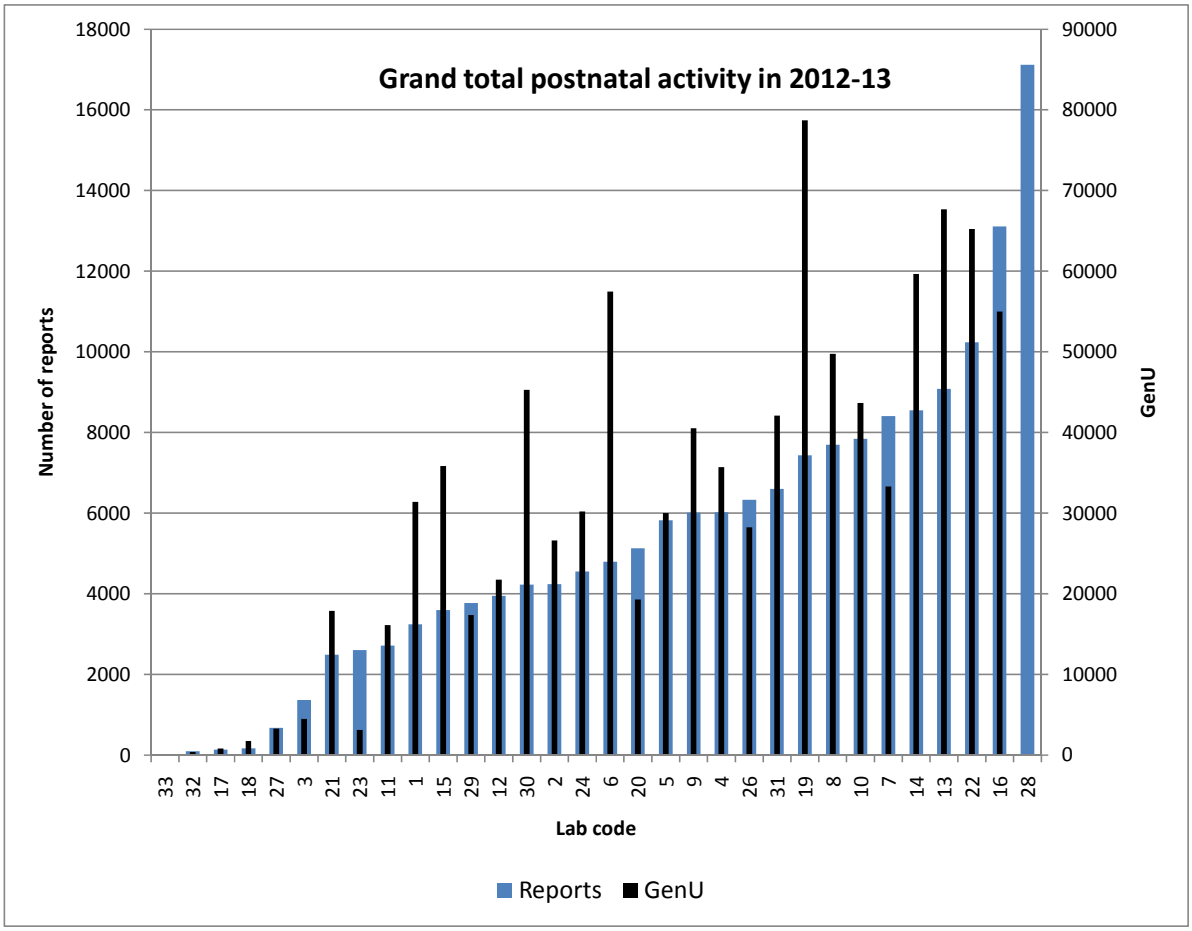
As in previous year's presents, postnatal and prenatal report activity is presented separately. The postnatal data is presented with the inclusion and exclusion of activity for acquired oncology, viral testing, therapeutic drug monitoring (TDM), tumour marker work and new-born screening (NBS). This is because this large volume work is not undertaken by all laboratories, is not part of the specialised service definition set for medical genetics and so skews the comparative analysis.

2012-13	**Total number of reports**	**Range**
Grand total	170,830	18-17,119
Excluding acquired oncology/viral/TM/HLA typing & research & storage	147,212	18-12,509
Excluding acquired oncology/viral/TM/ HLA typing research/storage/newborn screening & postnatal aneuploidy	138,928	0-12,374

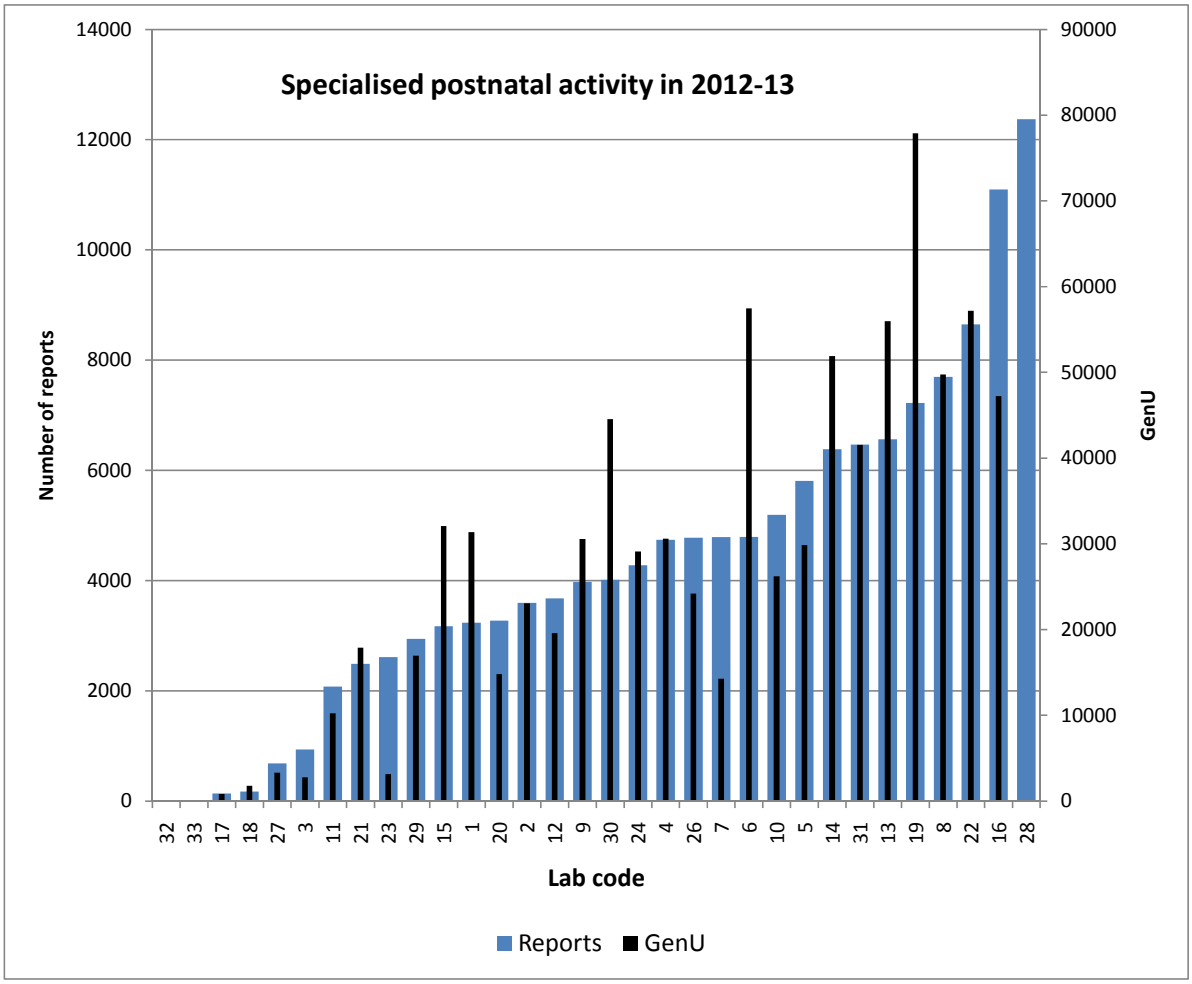
The postnatal activity for 2012-13 shows a 3% increase in the grand total number of reports from 2011-12(165,805). The more specialist activity* shows a smaller, approximately1.7% increase from 2011-12 (136,514).

* i.e. after excluding acquired disorders, viral, therapeutic monitoring, research, storage only, cystic fibrosis newborn screening and post natal aneuploidy screening tests.

In 2012-13 around 732 different categories of postnatal report were recorded of which 691 were for specialised genetic disorders. Note this is approximate as it 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.
This compares to around 626 different specialised disorders for which reports were issued the previous year.
The range in number of providers for individual test type ranged from 1 to 21 laboratories.



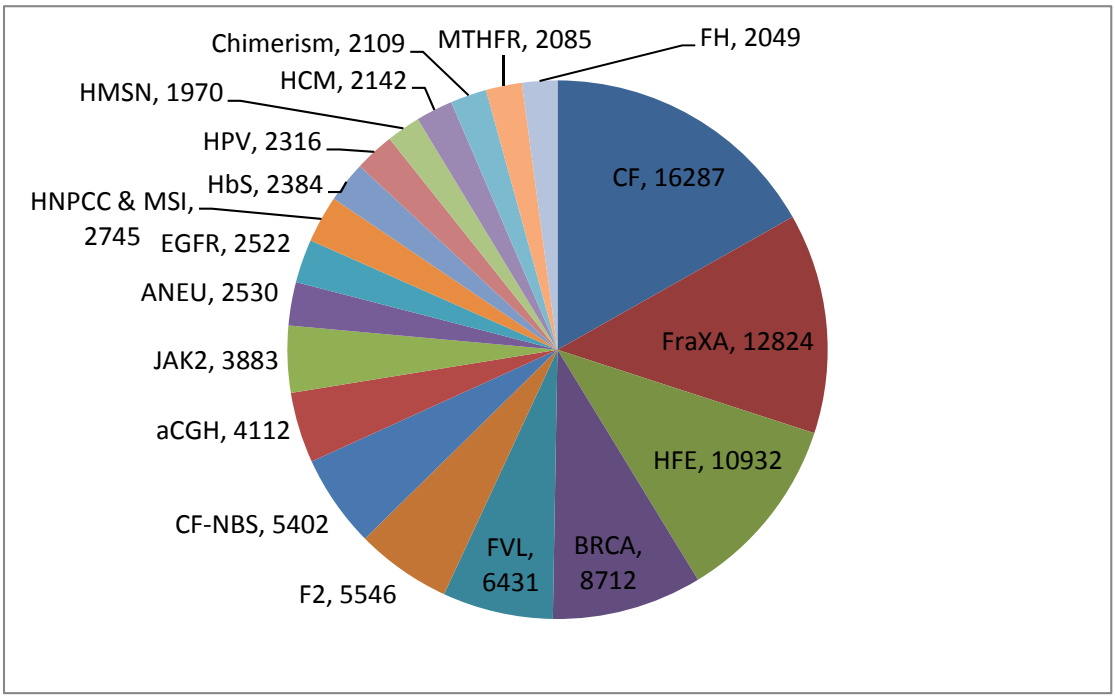
2.1 Grand total number of all postnatal reports and postnatal GenU for each laboratory, ranked by sum of report numbers.
No GenU data was available for Lab 28.



2.2 Total number of postnatal reports and postnatal GenU for each laboratory relating to specialised genetic tests only i.e. excluding acquired oncology/viral/TM/ HLA typing research/storage/newborn screening & postnatal aneuploidy.

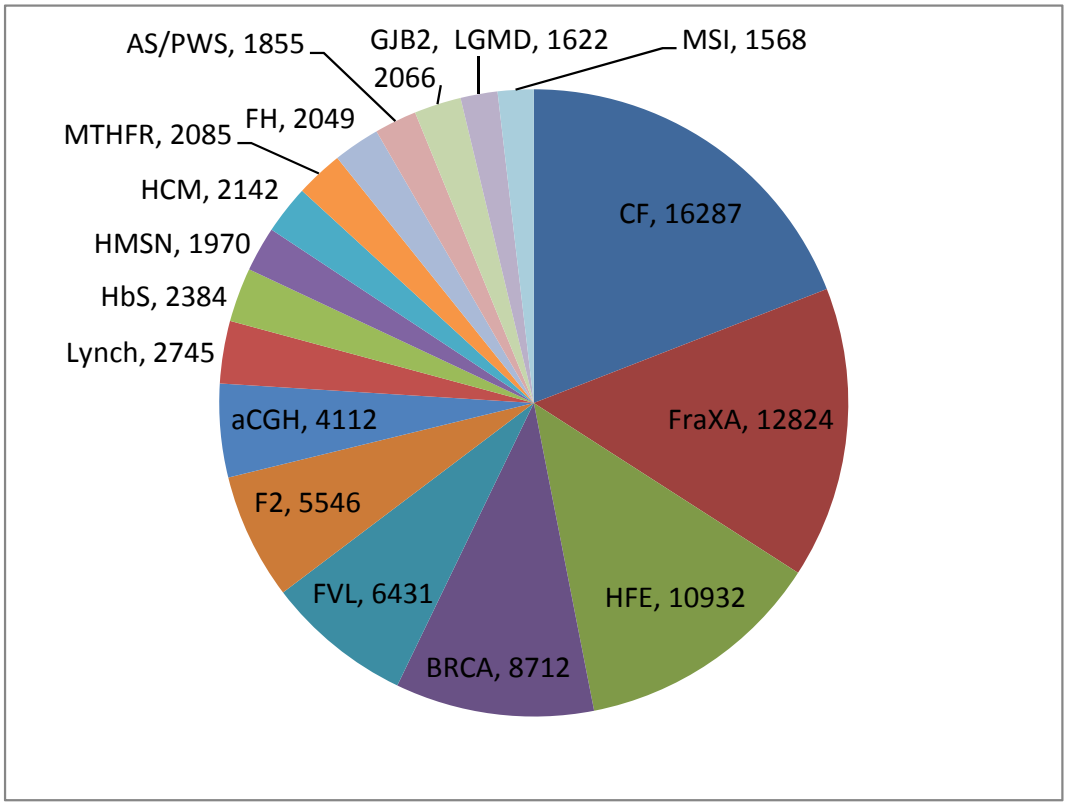
There was no GenU activity for Lab 28

Top (>2000) Postnatal Reports by Disorder Issued in 2012-13

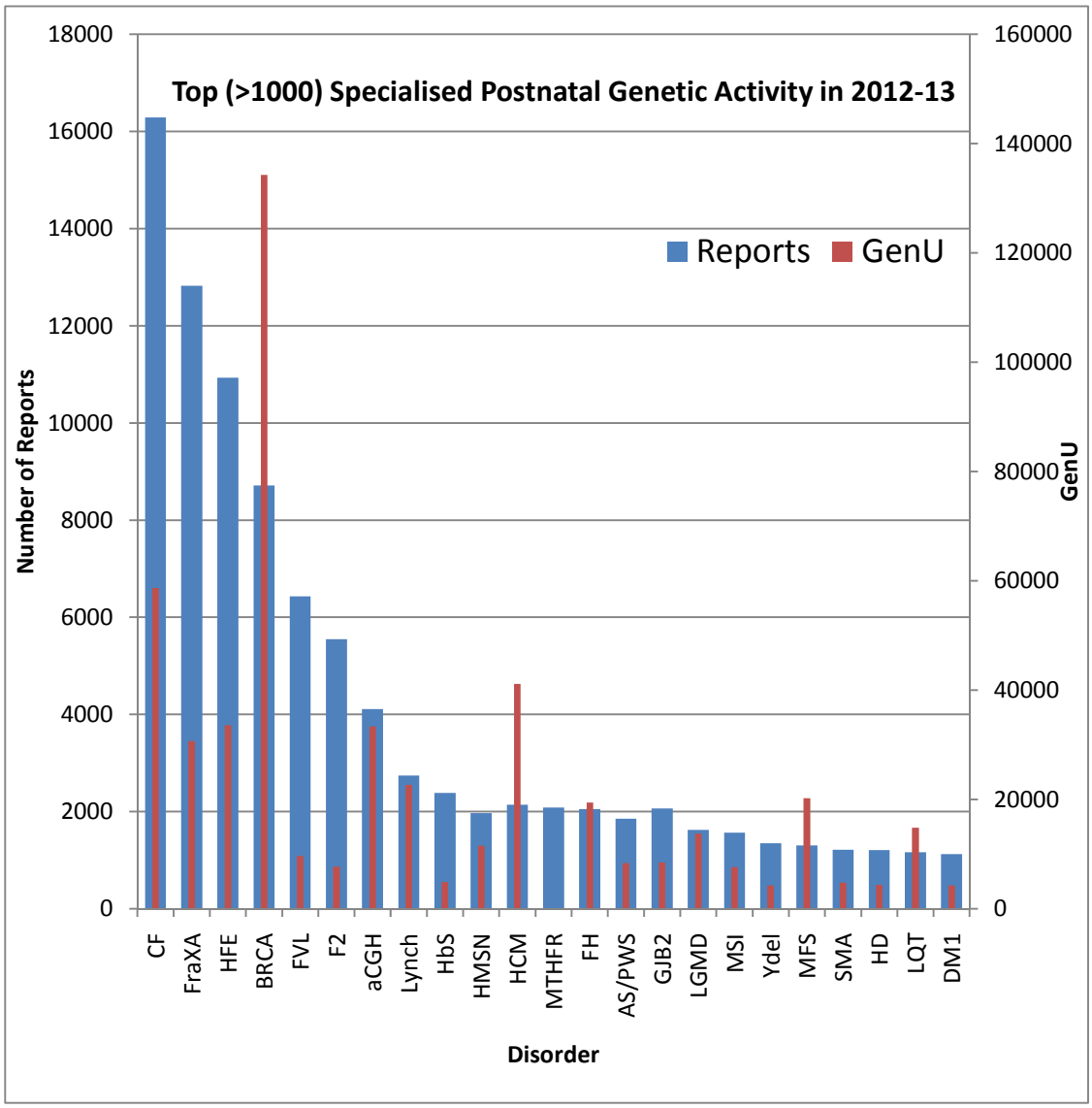


2.3 Pie chart of individual test categories that account for more than a total of 2000 annual reports.
Cystic fibrosis (CF), Fragile X (FraX) and haemachromatosis (HFE) are the top three report types as in 2011-12.

Top (>2000) Specialised Postnatal Reports by Disorder Issued in 2012-13

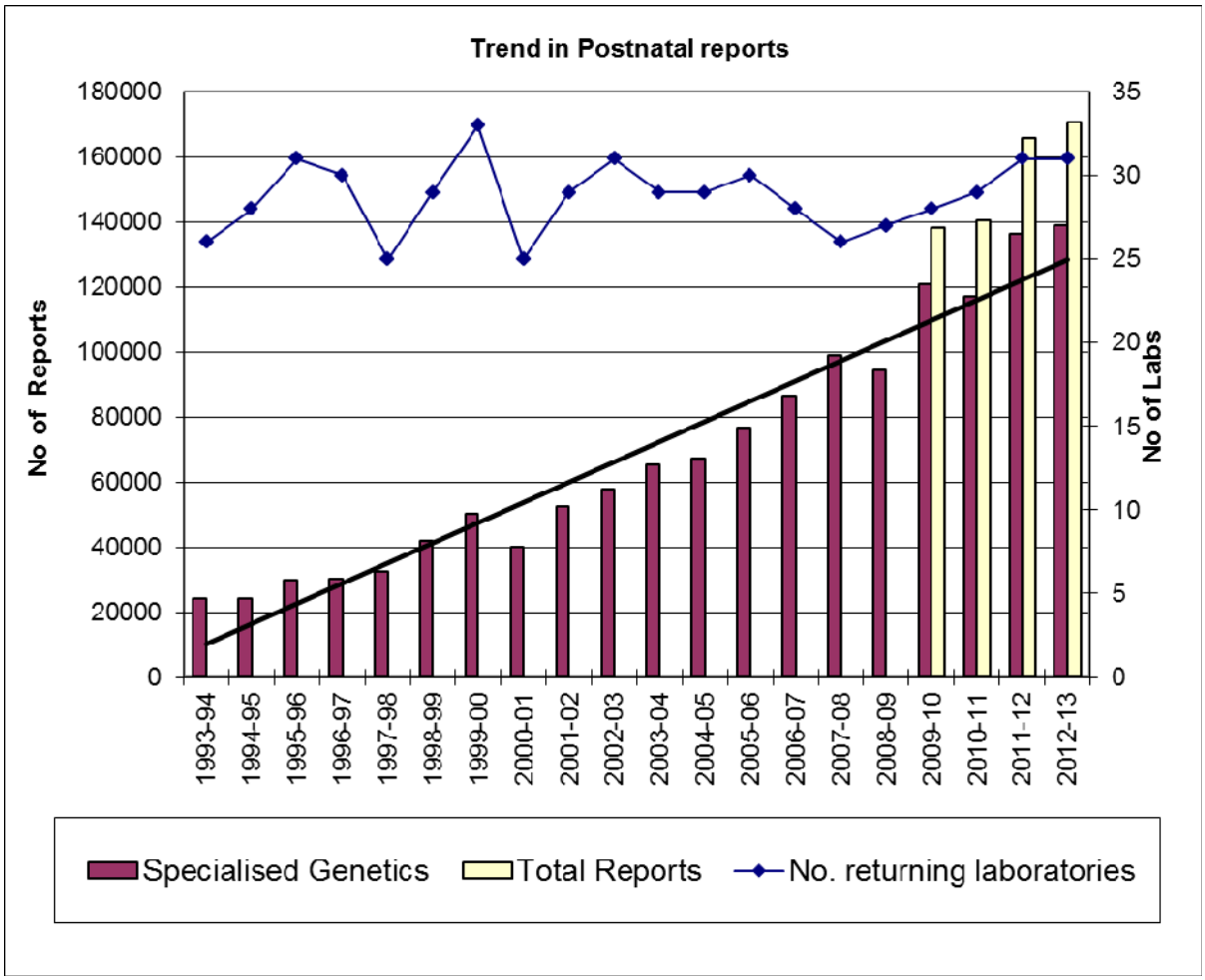


2.4 Pie chart of individual specialised genetic test categories that account for more than a total of 1000 reports.

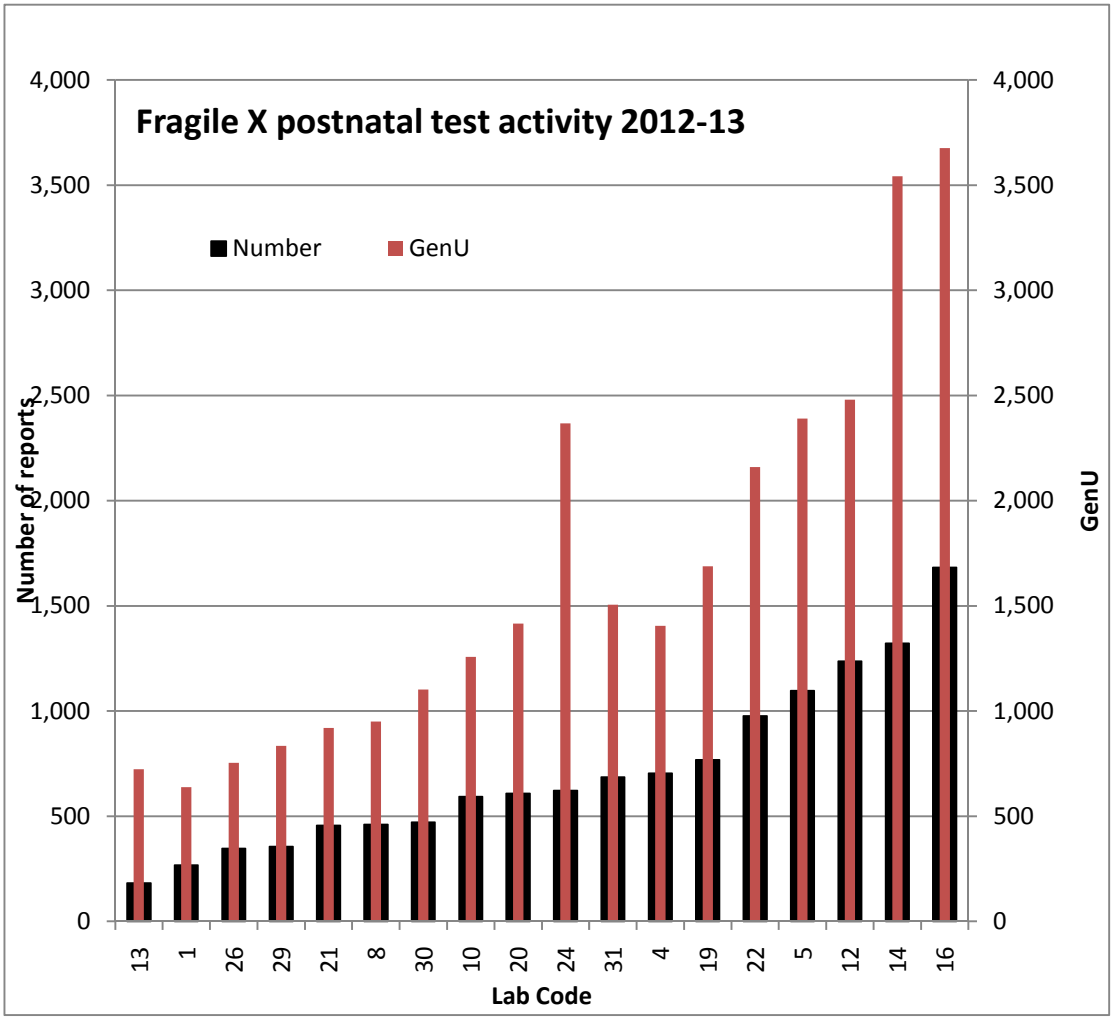


2.5 Chart of individual specialised genetic test categories that account for more than 1000 reports (number of reports) together with the associated workload (GenU)
Note the aCGH activity is only from 4 molecular providers so should not be included in the comparative analysis.

By report number, the top five disorders (CF, FraX, HFE, BRCA & FVL) are the same as last year.
By GenU, the top four disorders (BRCA, CF, HCM, HFE, FraX) are the same but there has been further increase in activity for cardiac tests (HCM).

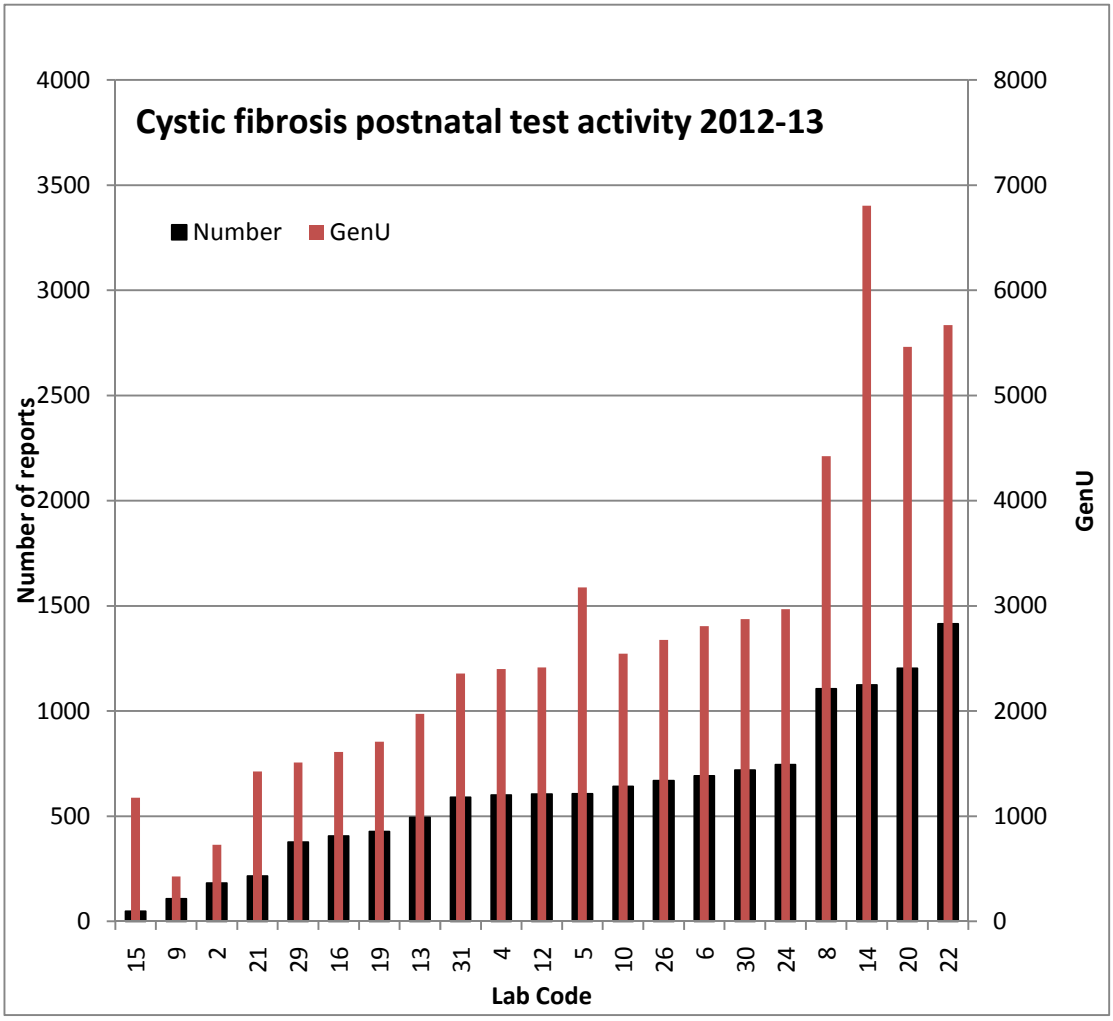


2.6 Chart showing the trend in postnatal report number with corresponding number of laboratories submitting data over past 20 years



2.7 Fragile X postnatal report activity

Mean number of reports was 712 reports across the 18 providers. The total number of postnatal fragile X reports was down to 90% of that in 2011-12.
The mean GenU per report was 2.4.
(A FraX PCR scores 2 GenU and a Southern blot 4 GenU).



2.8 Cystic fibrosis postnatal report activity

*Excludes new-born screening (NBS) activity where identified
No GenU data was provided by Lab 28

Mean number was 740 annual reports across the 22 providers.
The mean GenU per report was 5.3
Targeted CF mutation screen scores 4 GenU.
(Lab 15 had a mean GenU per report of 25, excluding lab 15, the mean was 4.3 GenU per report)

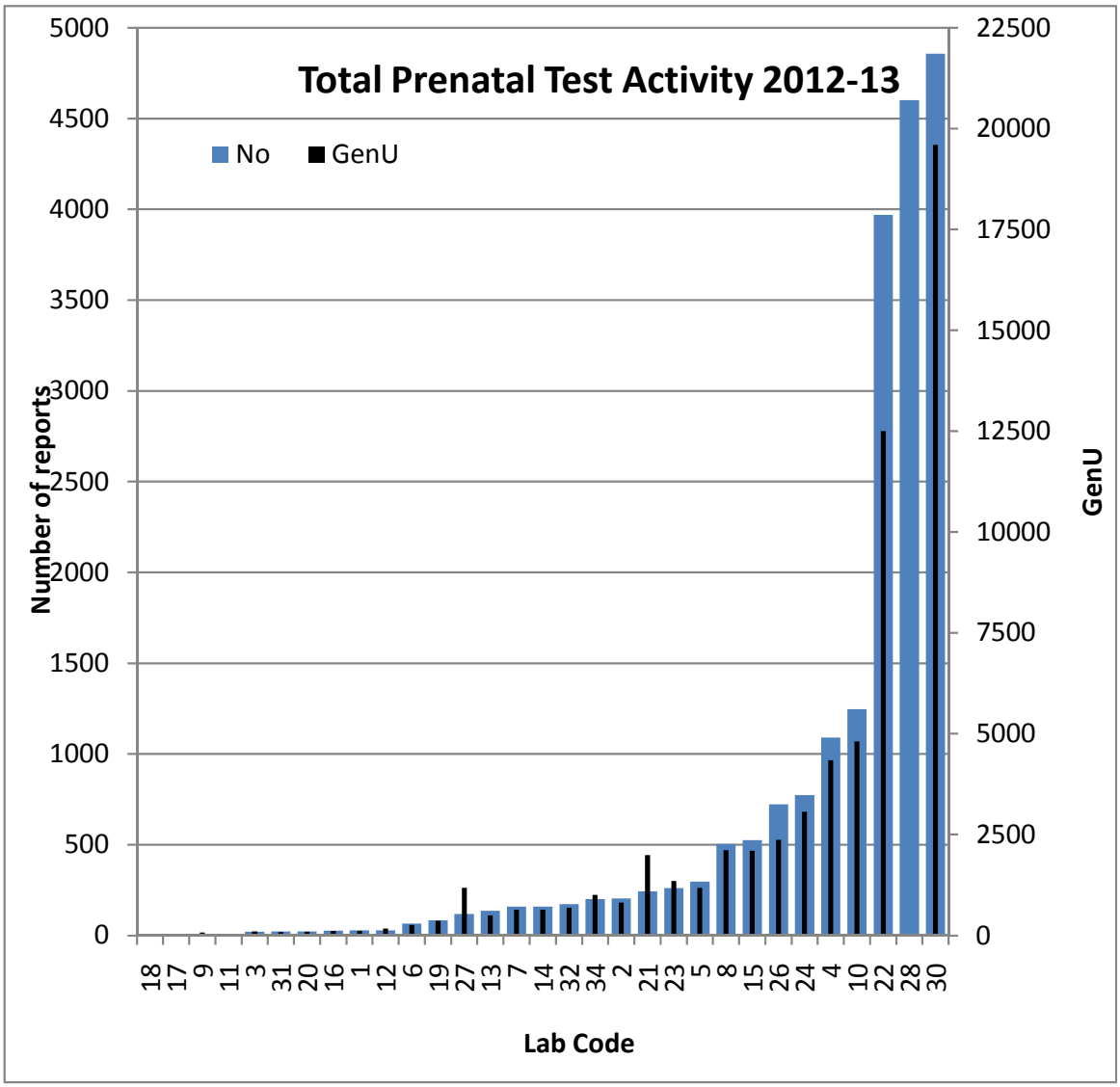
3 PRENATAL REPORTS for 2012-13

Prenatal diagnosis reports were recorded for 190 named different disorders (compared to 183 last year across 33 of the total 34 provider labs.

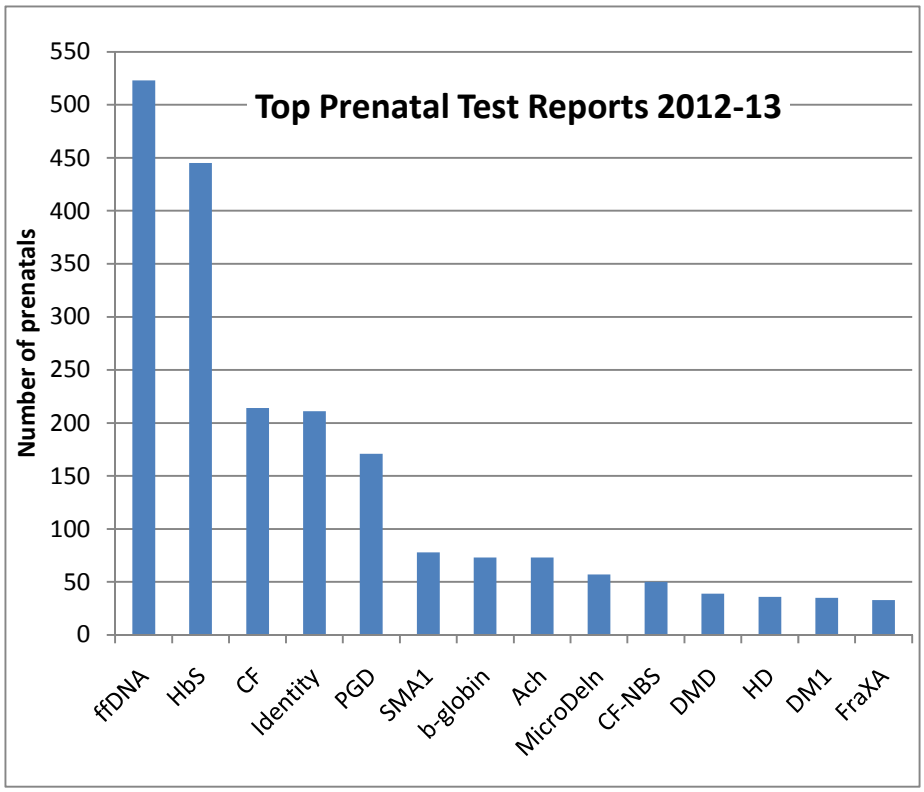
The largest activity (83% grand total) was screening for the common aneuploidies reported by 12 laboratories.

The grand total for prenatal reports was 20,557 up 50% on last year (13,258), reflecting the inclusion of aneuploidy screening activity from the non-molecular genetics (ACC) laboratories.
Excluding aneuploidy & sub telomere MLPA & microdel the total was 2642 that was similar to last year (2635).

Non-invasive testing fetal sexing was reported by 5 laboratories and accounted in total for 19% of the non-aneuploidy screening reported prenatal activity as last year.



3.1 Chart of prenatal report activity including Qf-PCR aneuploidy screening
GenU data was not available from Lab 28



Top (>30 reports) prenatal test activity excluding Qf-PCR aneuploidy screening by number of reports

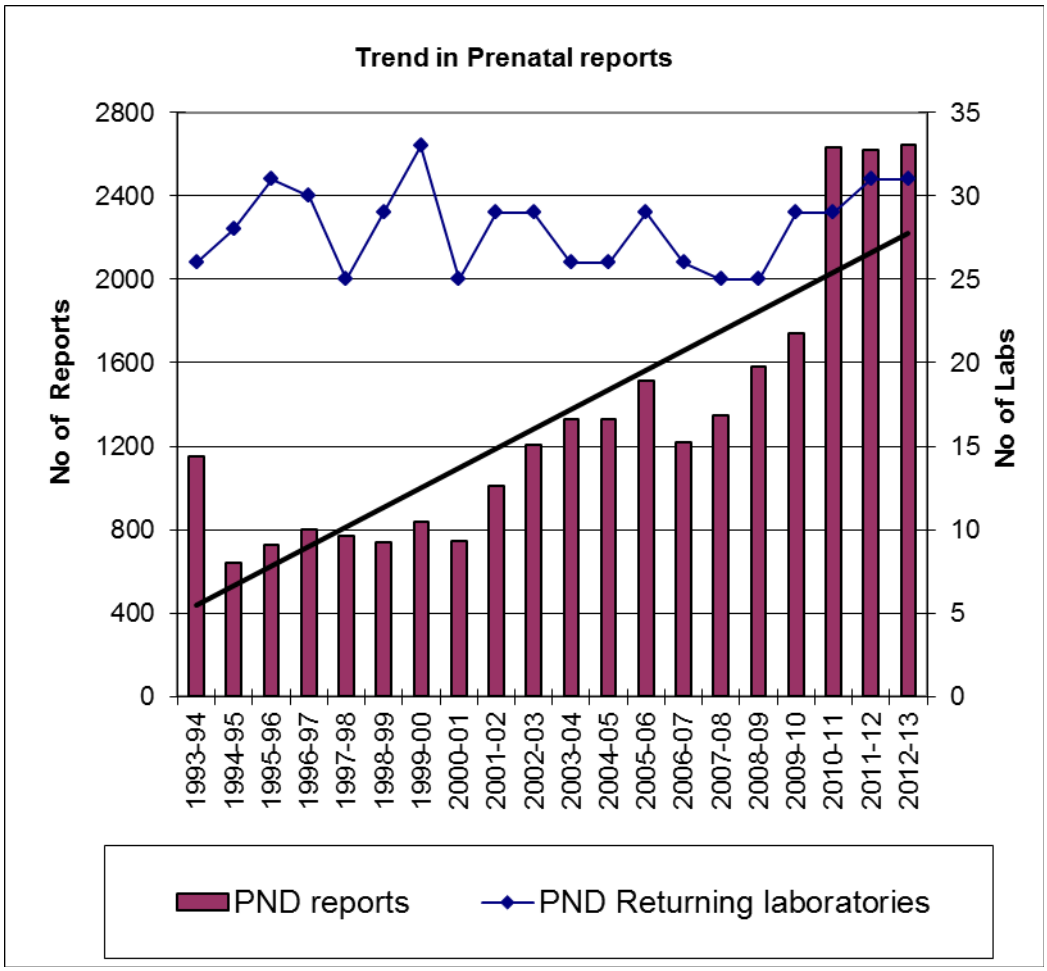
Pre-implantation diagnosis (PGD) and free fetal DNA sexing (ffDNA) is not broken down by the disease type.

The total number of prenatal aneuploidy screening reports for 2012-13 was 17,001 performed by QfPCR and 307 by FISH.

Year	Original total	No. of submitting CMGS labs	FREE FETAL DNA -sexing	SICKLE CELL ANEMIA	Cystic fibrosis*	BETA THALASSAEMIA	SPINAL MUSCULAR ATROPHY	ACHONDROPLASIA	FRAGILE SITE MENTAL RETARDATION 1	MUSCULAR DYSTROPHY, DMD&BMD	UPD all	HUNTINGTON DISEASE	CRANIOSYNOSTOSIS	DYSTROPHIA MYOTONICA 1	ALPHA THALASSAEMIA	TUBEROUS SCLEROSIS	POLYCYSTIC KIDNEY DISEASE, RECESSIVE	THANATOPHORIC DYSPLASIA; TD	CAH TO 21-HYDROXLASE DEFICIENCY	AS/PWS	TREACHER COLLINS	ADRENOLEUKODYSTROPHY; ALD	HEMOPHILIA A	ABNORMAL HAEMOGLOBINS	Di George
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		1
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	3
2010-11	2635	28	432	578	288	154	50	65	49	51	98	39	14	28	11	10	12	6	16	12	5	5	0		81
2011-12	2621	31	491	415	197	187	58	67	32	44	23	30	13	27	20	11	11	14	9	9	11	8	4	300	75
2012-13	2642	31	523	445	214	73	78	73	33	39	27	36	16	35	14	8	15	29	12	10	4	3	4	79	22

3.3 Table summarising prenatal diagnosis activity for single gene disorders

*Corrected CF prenatals (1998-99 to 2007-08 originally included some FEB tests



3.4 Chart to show the trend in prenatal report numbers (excluding aneuploidy screening) with corresponding number of laboratories submitting data over past 20 years.

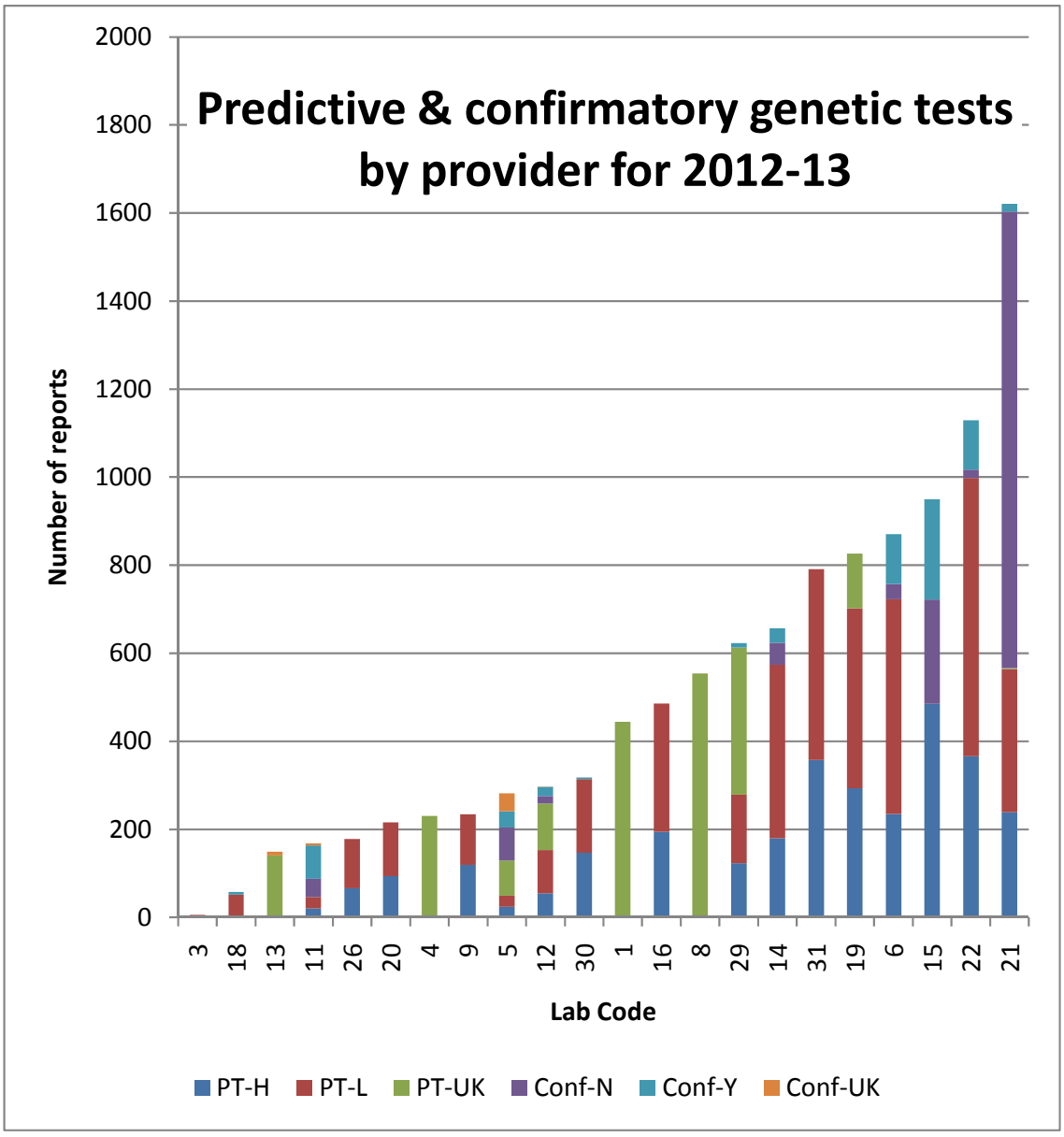
3 PREDICTIVE TESTS 2012-13

The ACGS member 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 as predictive tests undertaken in asymptomatic relatives and subsequent confirmatory tests undertaken in symptomatic relatives to confirm the molecular diagnosis within the family (exclude phenocopy). The total number of tests (table 4.4) includes the diagnostic tests undertaken to make the diagnosis in an individual meeting the appropriate clinical criteria.

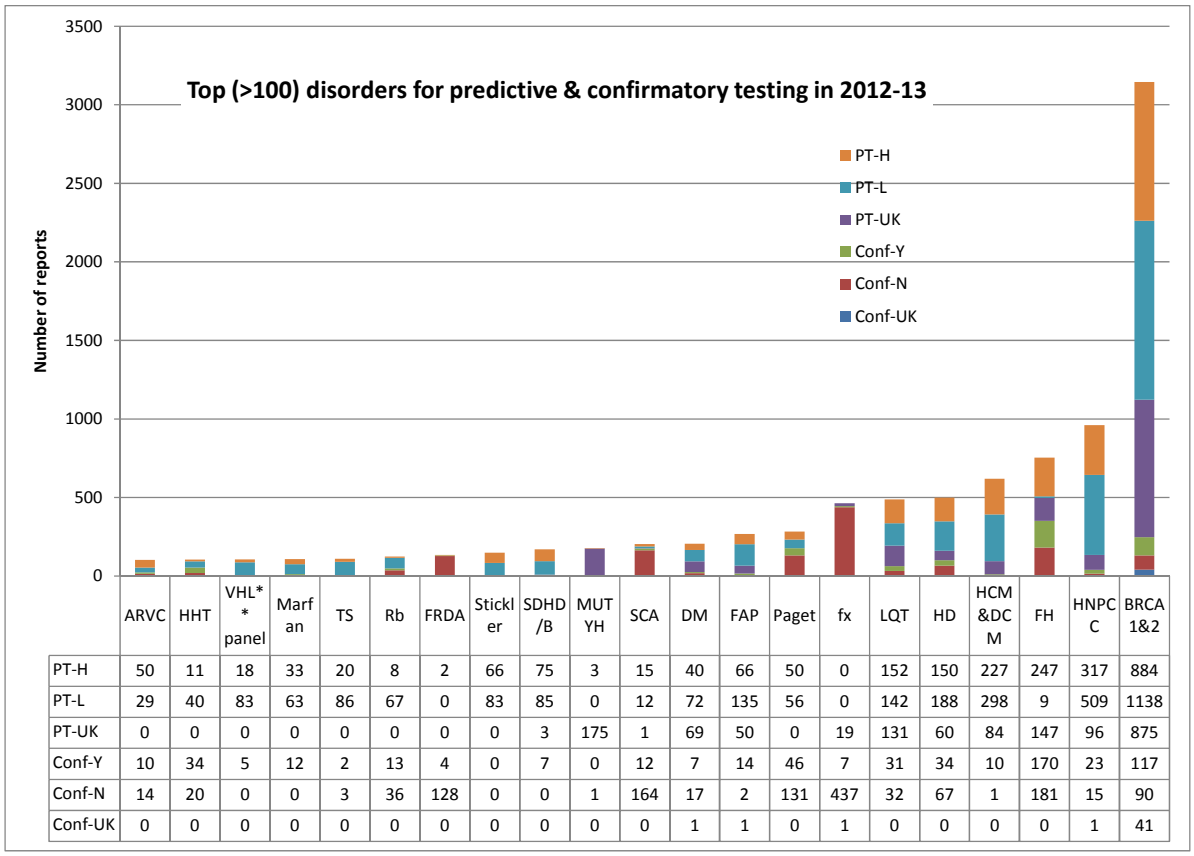
2012-13	2011-12	2010-11	2009-10	2008-9	Category
22	24	22	22	21	Number of lab returns
127	138	116	128	129	Number of specified disorders
2011	3037	1128	1756	1404	Unknown outcome of predictive tests
57	207	2	106	261	Unknown outcome of confirmatory tests
3002	3193	2397	2510	1945	High risk predictive
3852	3781	3624	3164	2738	Low risk predictive
795	719	430	288	577	Yes-Confirmatory test
1511	1565	558	521	484	No-Confirmatory test
11228	**12502**	**8139**	**8345**	**7409**	**Grand Total (predictive & confirmatory)**

4.1. Summary table of predictive and confirmatory tests for the last 5 years.



4.2 Breakdown for the 21 provider labs

Key
PT-UK refers to the predictive tests where the outcome has not been provided
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 refers to the confirmatory tests where the outcome has not been provided



	ARVC	HHT	VHL** panel	Marfan	TS	Rb	FRDA	Stickler	SDHD/B	MUTYH	SCA	DM	FAP	Paget	fx	LQT	HD	HCM &DCM	FH	HNPCC	BRCA 1&2
PT-H	50	11	18	33	20	8	2	66	75	3	15	40	66	50	0	152	150	227	247	317	884
PT-L	29	40	83	63	86	67	0	83	85	0	12	72	135	56	0	142	188	298	9	509	1138
PT-UK	0	0	0	0	0	0	0	0	3	175	1	69	50	0	19	131	60	84	147	96	875
Conf-Y	10	34	5	12	2	13	4	0	7	0	12	7	14	46	7	31	34	10	170	23	117
Conf-N	14	20	0	0	3	36	128	0	0	1	164	17	2	131	437	32	67	1	181	15	90
Conf-UK	0	0	0	0	0	0	0	0	0	0	0	1	1	0	1	0	0	0	0	1	41

Figure 4.3
Disorders that account for more than 100 annual predictive or confirmatory reports.

There has been a relative increase in cardiac testing

Disease codes are:
ARVC (arrhythmogenic right ventricular cardiomyopathy), HHT (Hereditary Haemorrhagic Telangiectasia) VHL ** neuroendocrine panel (VHL, SDHB, SDHC, SDHD, SDHAF2, PRKAR1A, TMEM127, RET), Marfan syndrome, Tuberous sclerosis (TS) Retinoblastoma (Rb) Friedreich ataxia (FDR) Stickler SDHD/B (succinate dehydrogenase subunit complex B), MUTYH associated polyposis SCA (spinocerebellar ataxia), DM (myotonic dystrophy), FAP (familial adenomatous polyposis coli), Paget's disease, FraX (fragile X) LQT (long QT), HD (Huntington's disease), FH (familial hypercholesterolemia), HCM & DCM (hypertrophic & dilated cardiomyopathies), HNPCC (hereditary non polyposis coli/Lynch syndrome), BRCA1/2 (breast & ovarian cancer),

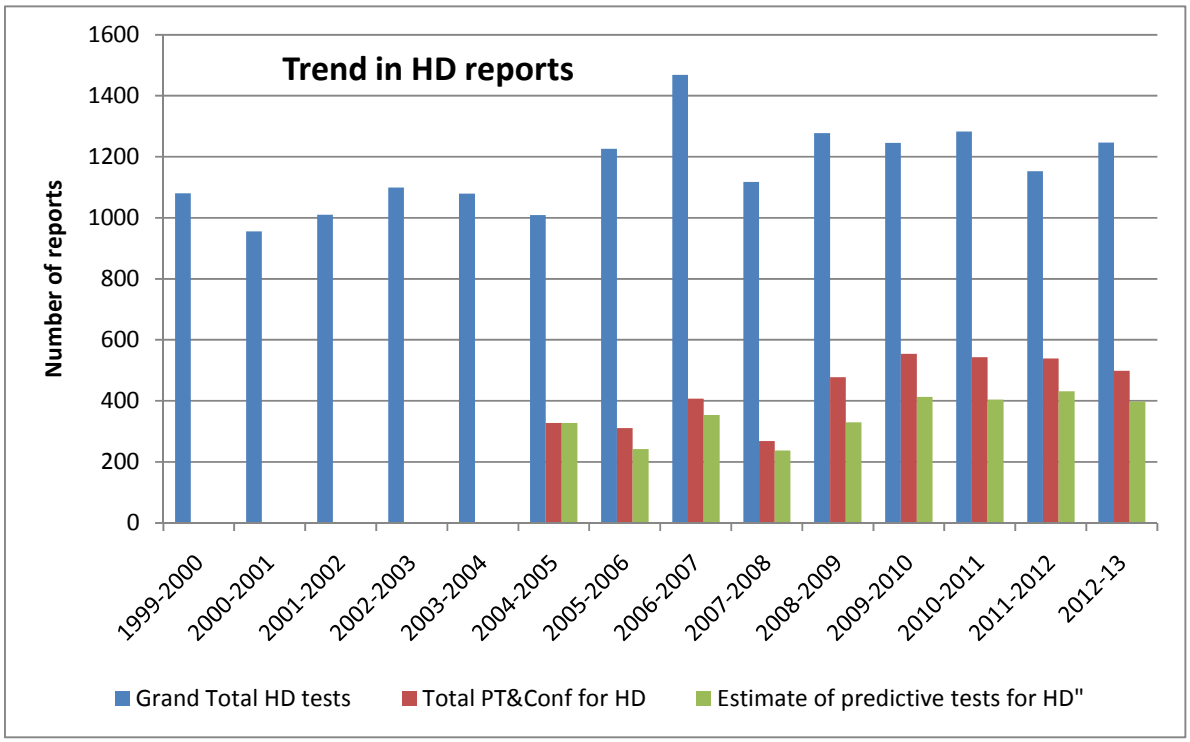
CMGS data	1999 2000	2000 2001	2001 2002	2002 2003	2003 2004	2004 2005	2005 2006	2006 2007	2007 2008	2008 2009	2009 2010	2010 2011	2011 2012	2012 2013
No. HD lab returns					17	15	13	11	13	10	9	13	14	**10**
No. BRCA lab returns					18	18	13	13	14	10	15	14	16	**11**
Grand Total HD tests	1080	956	1010	1099	1079	1009	1226	1469	1118	1278	1245	1283	1153	**1246**
Total PT&Conf for HD						328	311	407	268	478	554	543	539	**499**
Grand Total BRCA tests	1740	1889	2469	3142	4635	4455	5309	6906	9048	7041	7733	7617	8381	**8712**
Total PT&Conf for BRCA						1124	1746	2548	2068	2141	2739	2364	3367	**3145**
Estimate predictive of total HD tests (%)						328 (33%)	242 (20%)	354 (24%)	237 (21%)	330 (26%)	413 (33%)	404 (31%)	431 (37%)	**398 (32%)**
Estimate predictive of total BRCA tests (%)					20%	939 (21%)	1021 (19%)	1393 (20%)	1949 (21%)	1890 (27%)	2564 (33%)	2224 (29%)	3083 (37%)	**2897 (33%)**

4.4 Table showing trend in activity for Huntington disease (HD) and familial breast/ovarian cancer (BRCA)

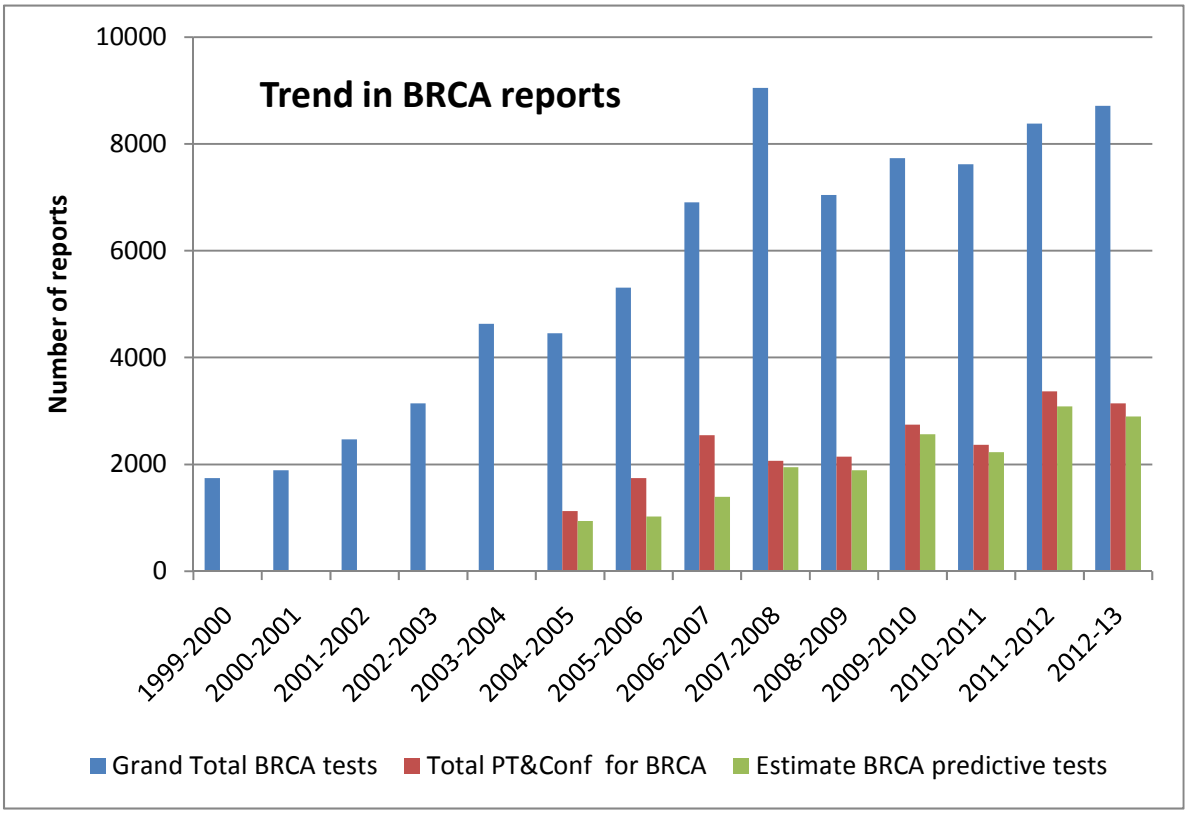
Comments

There is an 8% decrease from last year in the number of reported Huntington disease predictive tests (398/431). The proportion of HD tests recorded as predictive tests against the total number of HD tests (398/1246) is 32% which is also down 5% from last year. The tests that are not predictive or confirmatory are inferred to be diagnostic queries.

There is a 6% decrease from last year in the number of reported familial breast cancer predictive tests (2897/308). The proportion of BRCA tests recorded as predictive against the total number of BRCA tests (2897/8712) is 33% which is down by 4% from last year. The tests that are not predictive or confirmatory are inferred to be diagnostic queries.

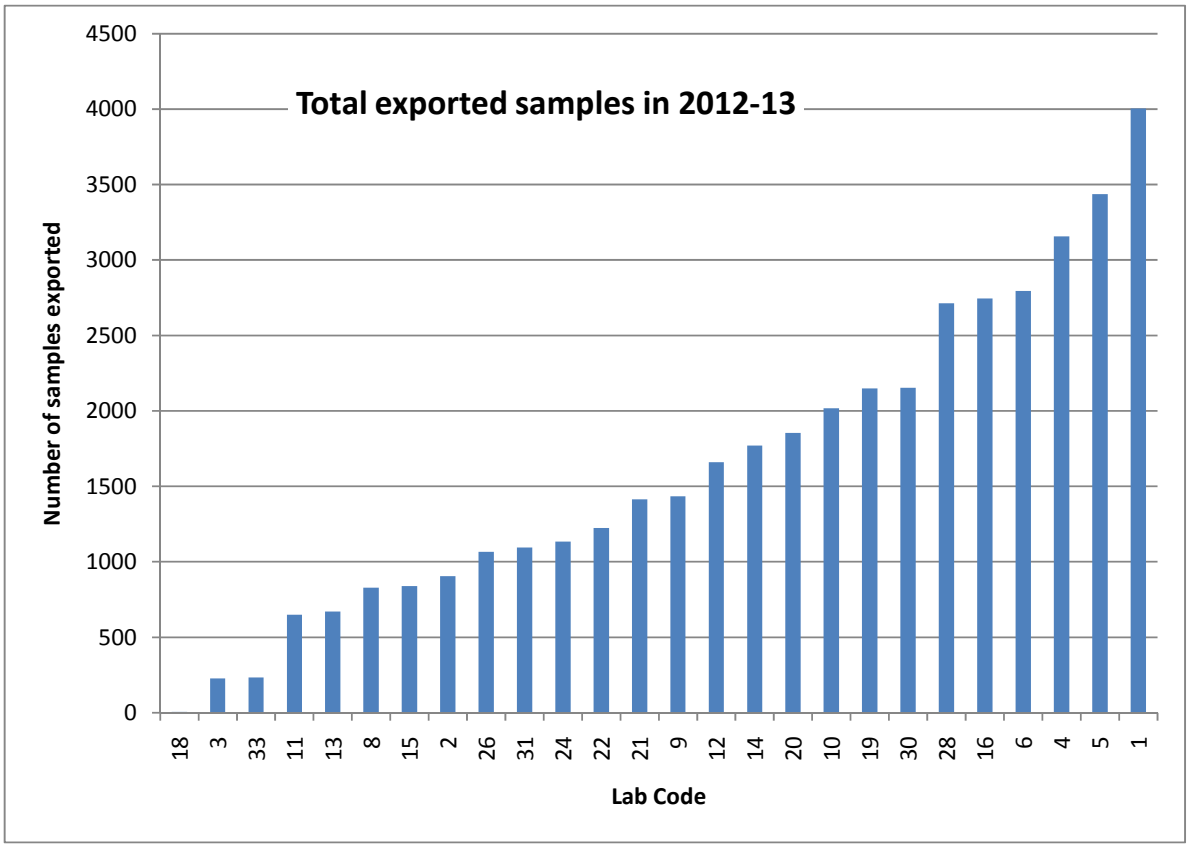


4.5 Trend in test activity for Huntington disease to show total (grand total), number of predictive (PT) and confirmatory (Conf) tests



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

5 EXPORTS

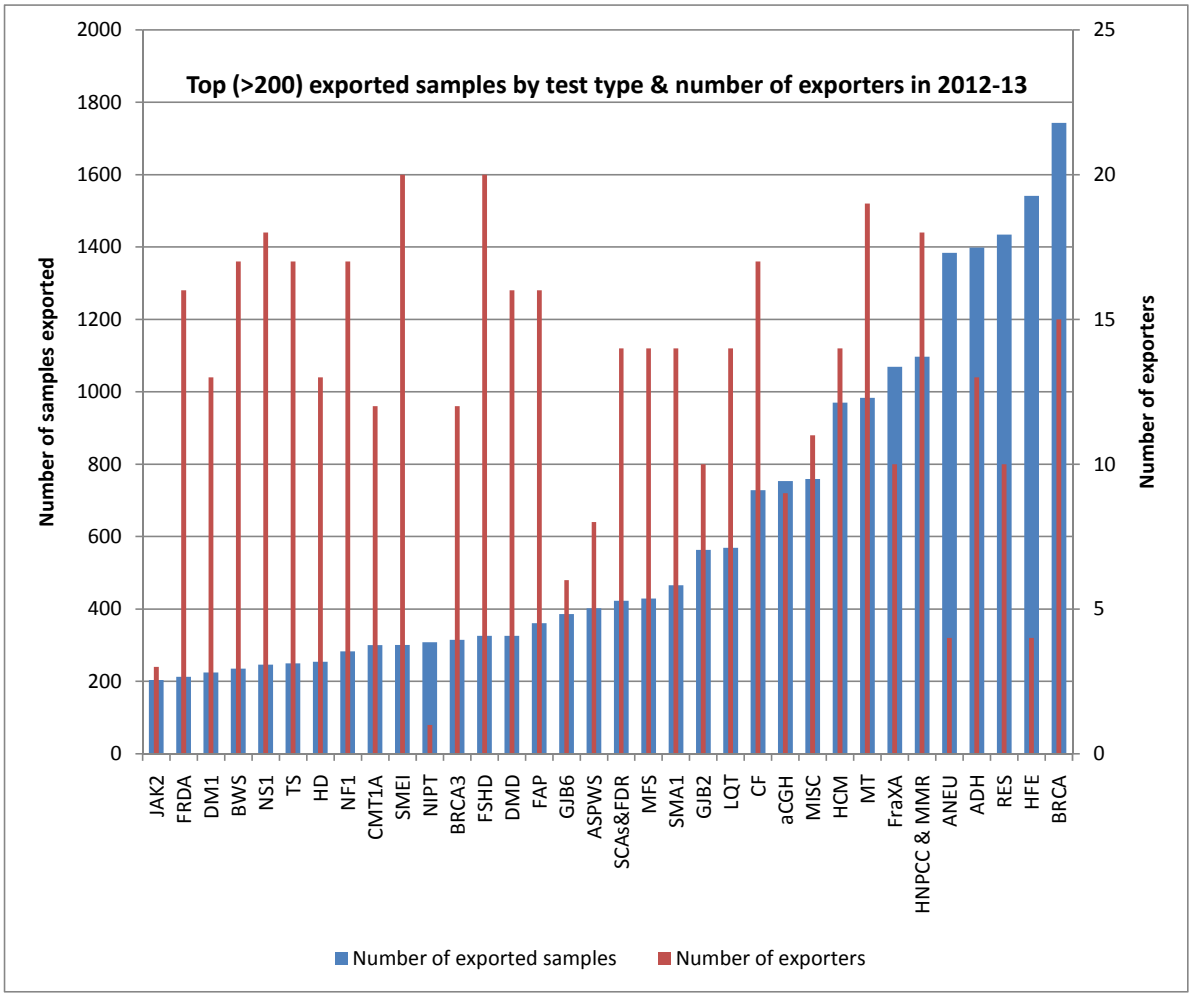


5.1 Chart showing total number of exports by each laboratory

Individual lab activity ranged from 6 to 4003 exports per lab as reported by the 26 respondents. The overall total number of exports was 42175 that was a 5% increase on the previous year.

Note

3.4% of all exports were for research samples and 9 labs reported exporting samples for array CGH that may include interdepartmental transfers but only accounted for 1.8% total exports.



5.2 Exported test activity by disease and number of exporting laboratories where the total number of exported samples was over 200.

BRCA remains the top exported test category.

Note

Most but not all of the laboratories provided a full breakdown by disease of their export activity.

6 REPORTING TIMES

The data is presented for a select range of tests that includes both the larger volume tests and those tests provided by the very specialist labs.
The data includes a summary of the average and range in reporting times and the percentage compliance of reports for each of the three White Paper reporting time targets plus the new 4w routine simple reporting time target.

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

		Reporting time																%											
Gene(s)	Disease	2 month				2 week				4 week				Urgent				2 month			2 week			4 week			Urgent		
		Min	Max	Mean	count	Min	Max	Mean	count	Min	Max	Mean	count	Min	Max	Mean	count	Min	Max	Mean	Min	Max	Mean	Min	Max	Mean	Min	Max	Mean
CFTR	**CYSTIC FIBROSIS**	13	72	43	6	2	14	7	16	5	12	9	7	1	17	3	19	24	100	64	77	100	93	90	100	97	84	100	97
2011-12		14	58	31	7	2	19	8	16	3	13	9	6	2	4	2	20	33	100	81	45	100	90	91	100	96	38	100	92
2010-11		9	45	28	4	3	9	6	14	4	15	9	6	1	3	2	17	31	99	79	50	100	92	84	100	94	80	100	94
2009-10		5	41	24	5	2	13	6	16					1	4	2	17	43	100	78	70	100	93				79	100	96
2008-9		6	38	23	5	3	15	7	17					2	4	3	13	59	100	91	1	100	90				1	100	87
DMPK	**DYSTROPHIA MYOTONICA 1**	16	30	22	6	6	35	15	8	6	16	11	7	1	8	4	12	93	100	99	52	100	79	65	100	91	0	100	79
2011-12		17	50	31	6	4	23	11	9	6	24	13	6	1	12	4	12	55	99	79	19	100	76	61	100	90	1	100	69
2010-11		17	34	22	5	5	9	7	8	7	19	12	6	1	3	2	9	50	100	88	34	100	84	74	100	92	58	100	92
2009-10		8	158	56	4	6	12	9	11					2	3	2	9	8	100	80	61	99	85				1	100	87
2008-9		11	46	26	6	6	16	9	9					2	8	3	9	33	100	78	44	98	77				50	100	83
FMR1	**FRAGILE SITE MENTAL RETARDATION 1**	21	71	34	14	6	17	10	12	9	20	15	6	1	7	3	6	34	100	82	11	100	82	59	98	82	100	100	100
2011-12		21	85	38	15	6	23	10	14	6	22	13	5	2	8	4	7	25	98	72	26	100	84	32	100	86	50	100	88
2010-11		15	77	33	12	6	11	8	12	6	20	15	7	2	7	3	6	53	100	86	33	100	83	57	100	87	33	100	89
2009-10		20	109	39	11	6	16	10	15					2	15	7	6	29	97	76	25	96	80				100	100	100
2008-9		18	42	29	12	6	20	11	13					3	10	5	4	1	100	74	6	20	11				1	100	72
IT15	**HUNTINGTON DISEASE**	36	36	36	1	6	17	9	12	6	15	10	5	1	4	2	11	100	100	100	39	100	88	84	100	94	50	100	94
2011-12		25	25	25	1	6	25	9	11	8	16	12	5	2	23	4	12	100	100	100	15	100	88	90	100	94	33	100	94
2010-11						6	10	7	9	5	15	11	4	1	5	2	9				67	100	93	92	99	95	100	100	100
2009-10		38	38	38	1	7	18	10	10					1	3	2	8	100	100	1	23	100	80				1	100	88
2008-9		0	15	8	2	5	43	11	12					1	14	4	8	98	98	98	1	100	73				0	100	60
Dystrophin	**DUCHENNE/BECKER MUSCULAR DYSTROPHY**	27	66	47	2	7	16	11	6	7	34	19	7	2	4	3	11	36	97	67	14	100	71	75	100	87	33	100	86
2011-12		10	26	18	3	6	14	10	7	6	88	29	5	2	4	3	11	96	100	98	26	98	74	40	99	75	50	100	82
2010-11		11	44	27	3	6	19	10	8	8	19	12	3	1	4	3	8	54	100	84	17	100	67	78	99	92	29	100	75
2009-10		28	55	38	3	6	33	15	10					1	4	3	8	31	94	70	19	100	67				60	100	86
2008-9		12	76	39	4	8	20	12	10					1	5	3	9	6	90	60	27	88	58				0	100	86

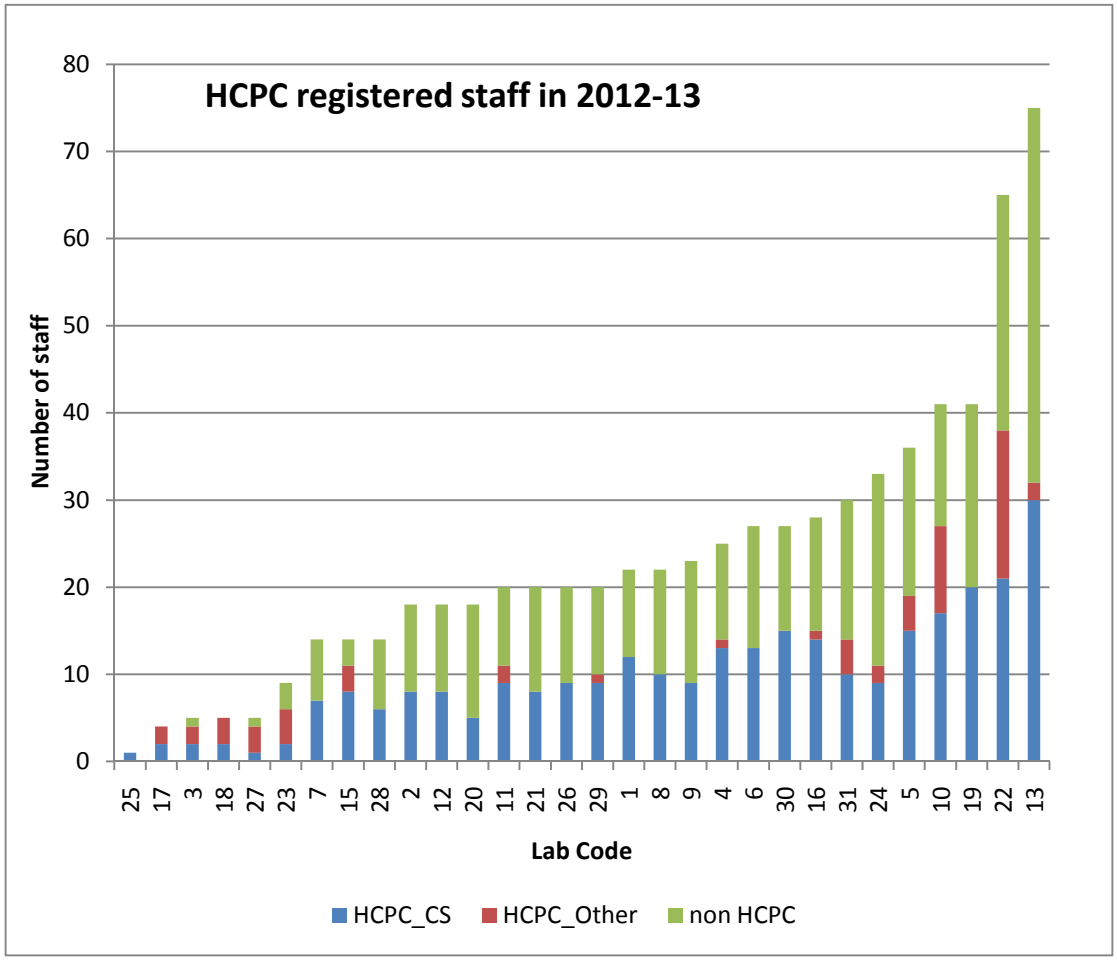
		Reporting time																%											
Gene(s)	Disease	2 month				2 week				4 week				Urgent				2 month			2 week			4 week			Urgent		
		Min	Max	Mean	count	Min	Max	Mean	count	Min	Max	Mean	count	Min	Max	Mean	count	Min	Max	Mean	Min	Max	Mean	Min	Max	Mean	Min	Max	Mean
SNRPN		9	67	34	6	8	16	10	9	9	36	14	12	1	5	3	11	27	100	75	60	100	79	25	100	86	78	100	98
2011-12	**AS&PWS**	27	37	32	2	9	12	10	5	4	74	30	3	2	4	3	2	66	81	74	62	93	80	13	100	69	29	100	64
2010-11		25	32	28	2	2	12	8	6	6	10	8	2	2	6	3	6	78	89	83	53	100	88	95	98	97	0	100	71
2009-10		40	59	49	2	8	35	18	4					2	2	2	1	53	53	53	50	96	72				100	100	100
2008-9		4	33	23	5	6	22	12	9					3	4	3	2	1	97	65	1	95	63				0	25	9
HFE						4	22	10	8	9	18	13	6	3	3	3	1				3	100	83	93	100	97	100	100	100
2011-12						6	16	10	6	6	17	13	7	7	7	7	1				12	99	70	87	100	97	98	98	98
2010-11	**HEMOCHROMATOSIS**					2	11	7	6	9	14	11	6								59	100	85	94	99	97			
2009-10						6	14	11	11												34	97	78						
2008-9						8	20	11	10												36	97	74						
PMP22		21	48	34	2	8	13	11	2	8	13	10	2					79	96	87	67	74	71	100	100	100			
2011-12	**CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 1A**	22	41	32	4	8	100	11	2	9	47	24	3					84	95	90	36	100	68	21	100	74			
2010-11		19	19	19	1	8	12	11	3	12	20	16	2	2	2	2	1	99	100	100	51	51	51	93	99	96			
2009-10		22	39	28	3	10	25	15	5									50	100	81	41	93	65						
2008-9		40	32	4	83	7	36	15	6					14	23	19	2	66	100	83	3	88	47					100	50
SMN		11	30	23	3	5	13	10	7	7	25	14	4	1	4	3	12	100	100	100	42	100	82	71	100	87	50	100	86
2011-12	**SPINAL MUSCULAR ATROPHY, TYPE I**	26	26	26	1	6	97	10	7	6	35	16	5	2	9	4	11	100	100	100	37	100	80	73	100	88	33	100	75
2010-11		8	55	32	3	5	10	8	9	11	17	13	3	2	5	3	9	83	100	92	34	100	83	75	100	91	1	100	77
2009-10		25	25	25	1	7	11	9	7					2	5	3	9	100	100	100	63	100	81				50	100	87
2008-9		61	61	61	1	6	12	8	7					2	5	3	6				56	92	79				14	100	81
BRCA1, BRCA2		26	144	48	14	7	16	10	9	10	25	15	9					13	99	75	66	98	87	78	97	90			
2011-12	**FAMILIAL BREAST & OVARIAN CANCER**	22	142	43	12	6	13	9	11	9	36	21	6	3	3	3	1	5	100	77	74	100	90	47	100	83	100	100	100
2010-11		22	237	55	11	7	10	7	9	7	19	11	5					27	98	70	29	98	74	78	100	89			
2009-10		22	617	110	10	7	26	12	12					2	2	2	1	18	100	69	71	94	66				100	100	100
2008-9		20	356	89	10	6	13	11	10									7	100	61	28	95	63					62	62

		Reporting time																%											
Gene(s)	Disease	2 month				2 week				4 week				Urgent				2 month			2 week			4 week			Urgent		
		Min	Max	Mean	count	Min	Max	Mean	count	Min	Max	Mean	count	Min	Max	Mean	count	Min	Max	Mean	Min	Max	Mean	Min	Max	Mean	Min	Max	Mean
APC	FAMILAL ADENOMATOUS POLYPOSIS	24	47	35	5	6	13	9	7	7	27	15	4	9	9	9	1	30	100	82	62	100	88	81	100	95			
2011-12		27	85	46	8	5	13	10	8	12	58	26	5	2	2	2	1	30	98	71	57	100	82	56	100	79	100	100	100
2010-11		28	51	39	7	6	13	9	6	11	18	15	3	2	3	3	2	23	100	64	29	100	73	67	91	83	100	100	100
2009-10		31	142	53	7	5	55	14	10																				
2008-9		8	238	68	6	7	17	12	7									6	100	57		95	78						
MSH2 MLH1 MSH6 PMS2 MSI	LYNCH SYNDROME	16	104	44	13	7	15	10	11	17	82	31	6	2	2	2	1	21	100	73	64	100	89	19	91	72	100	100	100
2011-12		25	229	58	13	6	15	10	11	15	278	75	5					7	100	69	48	98	79	9	95	62			
2010-11		23	55	39	10	8	21	12	8	17	42	26	3	3	3	3	1	25	25	25	0	96	63	14	47	31			
2009-10		22	180	72	10	3	504	62	11																				
2008-9		37	220	72	8	7	18	10	8									2	100	63	17	97	69						
F5	FACTOR V DEFICIENCY					7	10	8	4	8	20	15	4	2	2	2	1				93	100	98	33	98	76	100	100	100
2011-12						2	14	8	6	9	14	12	3	6	6	6	1				82	100	95	83	100	94	45	45	45
2010-11						1	7	5	3	8	14	11	5	5	5	5	1				90	100	97	77	100	93	56	56	56
2009-10		11	11	11	1	6	14	9	6					5	5	5	1	93	93	93	58	99	83				33	33	33
2008-9						7	20	11	2												51	93	79						
Cx26	GAP JUNCTION PROTEIN, BETA-2	20	44	32	6	9	25	15	4	11	38	21	4	3	3	3	1	46	100	81	22	83	61	26	100	68	100	100	100
2011-12		23	55	33	6	9	39	14	3	24	38	29	3	2	2	2	1	28	100	83	4	76	40	9	43	30	100	100	100
2010-11		18	41	28	7	10	13	11	2	16	60	38	2	2	2	2	1	85	100	95	27	76	53	28	75	52	100	100	100
2009-10		18	35	28	7	8	25	14	5					2	2	2	1	1	99	75	38	100	69				100	100	100
2008-9		20	31	25	4	8	22	17	4					1	1	1	1	1	99	66	0	100	33				1	1	1
Mt	MITOCHONDRIAL MYOPATHY	20	35	26	3	9	9	9	1	13	35	22	4	1	1	1	1	98	98	98	98	98	98	33	94	74	100	100	100
2011-12		20	39	30	4	7	41	19	3	11	16	14	3	2	4	3	2	57	96	78	93	100	97	83	90	88	100	100	100
2010-11		23	31	27	2	8	26	17	2	13	13	13	1	3	3	3	1	90	97	93	17	72	44	89	89	89	100	100	100

		Reporting time																%												
Gene(s)	Disease	2 month				2 week				4 week				Urgent					2 month			2 week			4 week			Urgent		
		Min	Max	Mean	count	Min	Max	Mean	count	Min	Max	Mean	count	Min	Max	Mean	count		Min	Max	Mean	Min	Max	Mean	Min	Max	Mean	Min	Max	Mean
2009-10		27	38	33	2	9	42	22	3					3	3	3	1		92	94	93	21	89	45				100	100	100
2008-9		30	97	63	2	8	15	11	2										25	85	55	25	82	53						
a-globin, b-globin	**ABNORMAL HAEMOGLOBINS**					8	8	8	1	17	17	17	1	4	4	4	1					100	100	100	66	66	66	33	33	33
2011-12		15	22	18	2	6	10	8	2					3	4	4	2		98	100	99	75	99	87				52	93	73
2010-11		17	28	23	2														81	86	84									
2009-10		15	28	22	2														87	100	93									
2008-9		25	25	25	1														87	87	87									
b-globin	**BETA THALASSAEMIA**					7	12	9	2	15	15	15	1	4	4	4	2					91	100	96	80	80	80	45	92	69
2011-12		20	20	20	2	15	15	15	1					4	26	12	3		91	96	94	46	91	69				33	92	50
2010-11		15	27	21	2	13	13	13	1					4	4	4	2		82	94	88	50	93	72				45	91	63
2009-10		17	20	19	2	7	7	7	1					4	5	4	2		88	98	93	93	93	93				37	59	48
2008-9		24	24	24	1	4	30	17	2										80	80	80	53	88	71						
RB1	**RETINOBLASTOMA**	29	36	33	2	7	7	7	1	6	7	6	2	2	3	2	2		76	92	84	96	96	96	100	100	100	100	100	100
2011-12		36	37	37	2	9	9	9	1	8	8	8	1	2	2	2	1		65	71	68	73	73	73	100	100	100	90	90	90
2010-11		46	55	50	2	8	8	8	2					2	3	2	2		40	46	43	73	100	87				100	100	100
2009-10		35	57	46	2	6	8	7	2					2	12	5	3		21	72	47	86	92	89				89	100	94
2008-9		33	33	33	1	9	9	9	1					1	3	2	2		68	68	68	80	80	80				100	100	100
STRs	**Aneuploidy**	45	45	45	1	2	9	4	5	3	15	7	5	0	2	1	10		60	60	60	85	100	97	67	100	93	93	100	98

6.1 Summary table of average reporting times between labs for select tests over past 5 years
Top line in each section is for 2012-13

7. Staff



7.1 Number of Health & Care Professions Council (HCPC) registered staff of total number service staff
Staff on the voluntary register council (VRC) have been included as HCPC_other

No data was provided by Labs 14, 32, 33 & 34

HCPC Clinical Scientists account for around 42% staff.
In total HCPC/VRC registered staff account for around 51% of the workforce.