ACGS Quality Subcommittee

Association for Clinical Genetic Science
Part of the British Society for Genetic Medicine

5 January 2016

Association of Clinical Genetics Science Activity Audit 2014-15

This year's activity report builds on the extension to scope introduced last year to reflect the wider range of services provided by ACGS members. These cover not only constitutional monogenic and cytogenetic services but also acquired cancer tests. Although there are other providers of acquired tests out with the genetic laboratory network, this area of activity is a key part of the genetic laboratory service repertoire because of the shared technology with constitutional analysis.

The data presented includes the number of samples received and processed, the number and type by disorder of constitutional postnatal, prenatal, predictive test reports and acquired tests, the number and type of tests that are sent for analysis at a different network laboratory and reporting time performance against current targets. 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 in support of service development and national benchmarking.

Gail Norbury Lara Cresswell

Gail Norbury and Lara Cresswell
ACGS Quality Sub Committee Audit Co-ordinators

Participants in 2014-15 Audit

Aberdeen Regional Genetics Laboratory
Belfast Regional Genetics Laboratory
Birmingham Children's Hospital
West Midlands Regional Genetic Centre (Birmingham)
Bristol Regional Genetics Laboratory
International Blood Group Reference Laboratory (Bristol)
All Wales Regional Genetics Laboratory (Cardiff)
Cheshire & Merseyside Regional Genetics Laboratory (Liverpool)
Dundee Regional Genetics Laboratory
East Anglian Regional Genetics Laboratory (Cambridge)
South East Scotland Regional Molecular Genetics Laboratory (Edinburgh)
Exeter Regional Molecular Genetics Laboratory
Leeds Genetics Laboratory
Leicester Cytogenetics Laboratory
London North East Thames Regional Genetics Laboratory (Great Ormond Street)
London North West Thames Regional Genetics Laboratory (Kennedy Galton)
Guys & St Thomas' (South East Regional Genetics Laboratory) including Molecular Haemoglobinopathy, Homeostasis, Dermatology and EB Laboratories
London South West Thames Regional Genetics Laboratory (St Georges)
London UCL Hospitals, Clinical Biochemistry, Haemoglobinopathy & Neurogenetics Laboratories
London Kings College Hospital Haemoglobinopathy, Liver, Molecular Pathology, Oncology & LMH,)
London Retinoblastoma Laboratory (Barts)
Royal Marsden Hospital (ICR & Cytogenetics)
Manchester Centre for Genomic Medicine
Christie NHS Foundation Trust Oncology Cytogenetics Laboratory (South Manchester)
Northern Regional Genetics Laboratory (Newcastle)
Norfolk & Norwich Cytogenetics Laboratory
Nottingham Regional Genetics Laboratory
Oxford University Hospitals Regional Genetics & Molecular Haematology & Oncology Laboratories
Sheffield Regional Genetics Laboratory
Wessex Regional Genetics Laboratory (Salisbury)

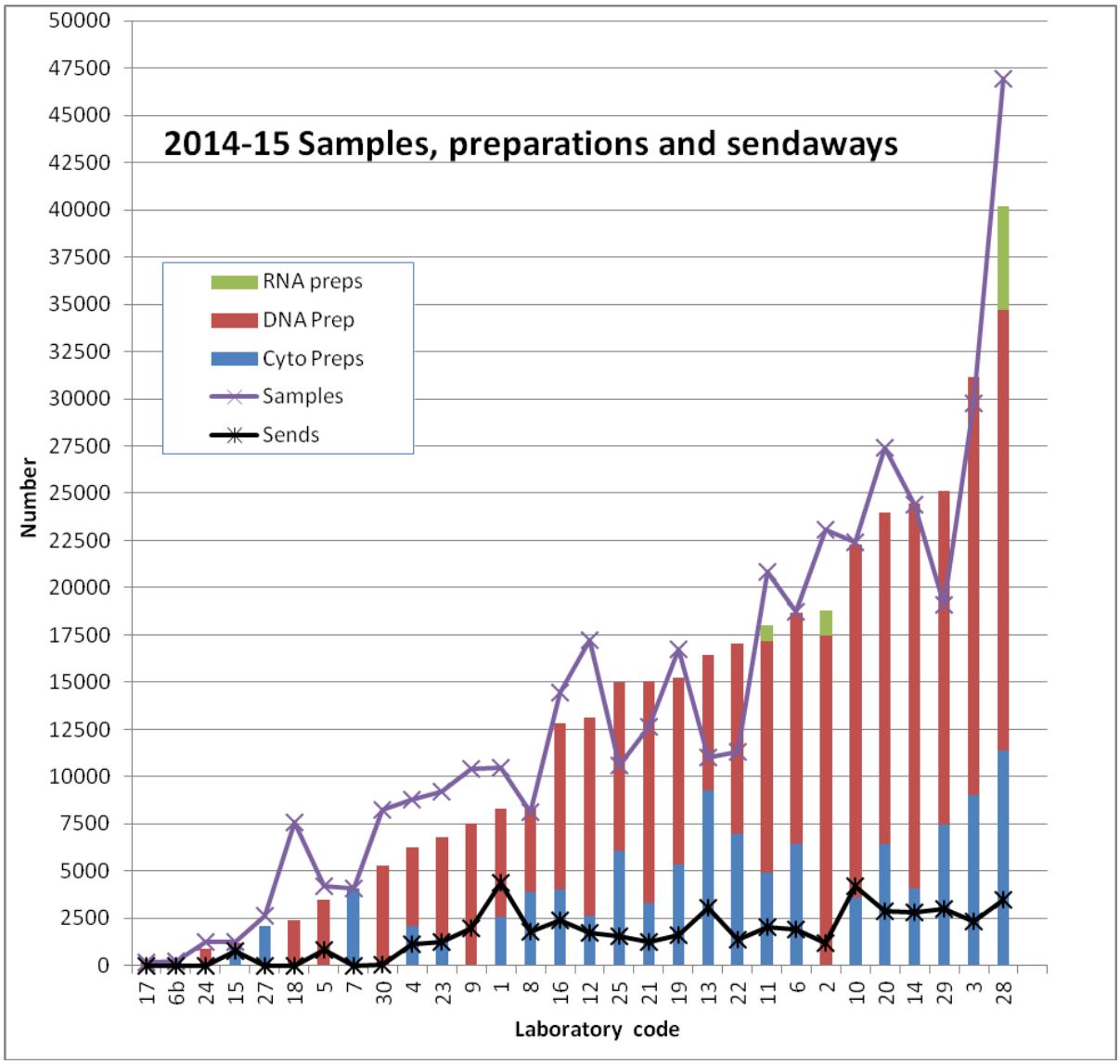
Note the laboratories below are listed alphabetically and the anonymous laboratory codes used on the charts are allocated on a random basis and are changed each year.
Most but not all participants submitted data for all parts of the audit. The code suffixes reflect separate submissions by submissions within the same organisation.

1. SAMPLES RECEIVED AND EXTRACTED 2014-15

	Category	Total	Range		Returns
		Sum	min	max	Count
Samples	Blood	244049	129	27107	29
	Amnio/CVS	17390	3	3552	25
	Buccal	2071	2	504	23
	Archive	19500	2	2953	25
	Bone marrow	39123	15	9252	19
	Solid tissues	11987	12	1720	23
	Other	18873	20	4311	26
	DNA	41688	26	4867	26
DNA Preps	Blood	138414	38	16386	28
	Amnio & CVB	10893	3	3522	24
	Buccal	1911	2	523	23
	Archive	8965	1	2351	20
	Bone marrow	13639	1	5584	12
	Solid tissues	8434	8	1622	20
	Other	8055	3	2135	25
Cyto Preps	Blood	39848	63	5215	21
	Amnio / CVB	11613	38	925	21
	Buccal	42	2	29	6
	Archived samples / Paraffin sections	7593	4	1658	15
	Bone marrow	24839	15	4617	18
	Solid tissues	7308	19	1411	19
	Others (tissues etc)	1904	15	647	12
Totals	Service samples	394681	184	46948	30
	DNA Preps	190311			
	Cyto Preps	93147			
	Research samples	23517	87	17832	18

1.1 Table of Sample and Preparation Summary

Only positive figures were included in the analysis to determine minimum values as there was not a consistent distinction between the use of a zero for no return or nil activity.



1.2 Chart showing total service samples received by each of the *30 laboratories/centres, ranked by the total and broken down by the different sample type.

*The separate returns from individual laboratories within the same organisation been combined for consistency.
Some laboratories included RNA preps in their DNA prep total
Lab 2 sends estimated

Note
Sample number and extraction number represent different measures of activity; sample number received may include aliquots of the same sample being counted multiple times if analysed by 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 ~12% service samples.

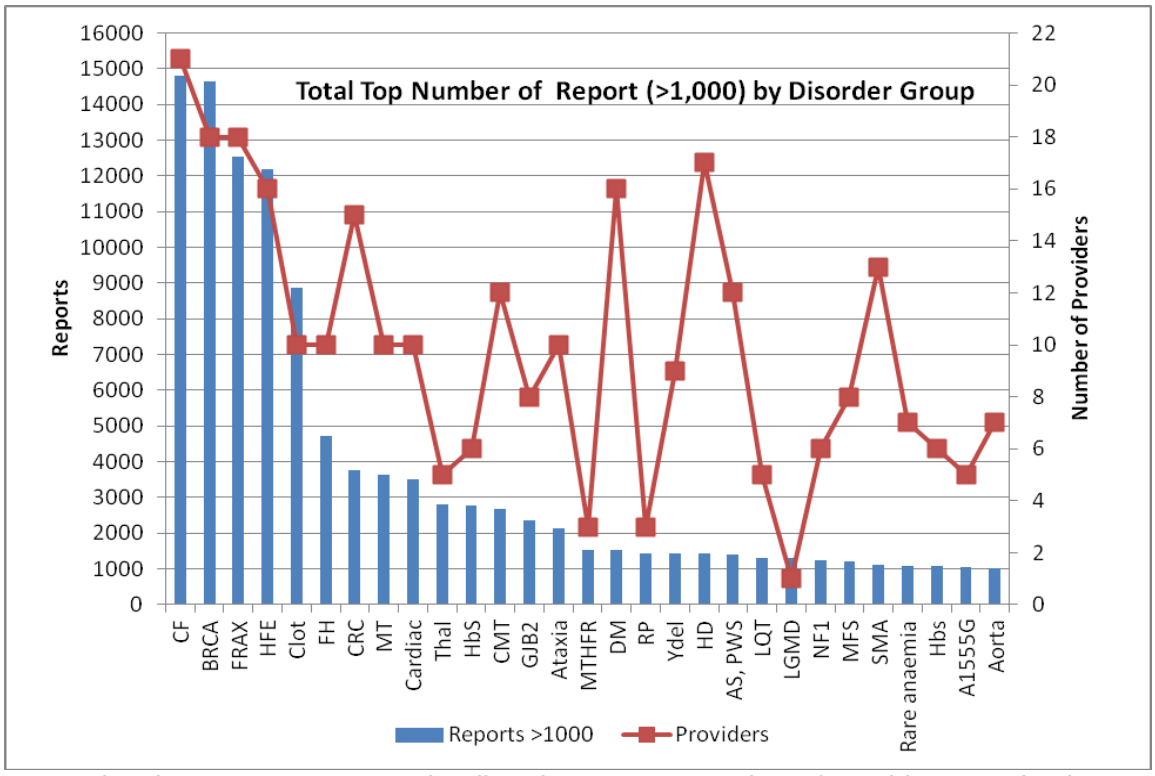
2. REPORTS 2014-15

2014-15	**Total Reports**	**Range Reports**	**Total GenU**	**Range GenU**
Molecular (monogenic)	150,656	173-13,483	1,221,685	740 – 135,312
Cytogenetic (constitutional)	90,818	789-10985	713,898	17,773 – 26,172
Acquired	108,229	87-14,773		
Grand total	**349,703**			

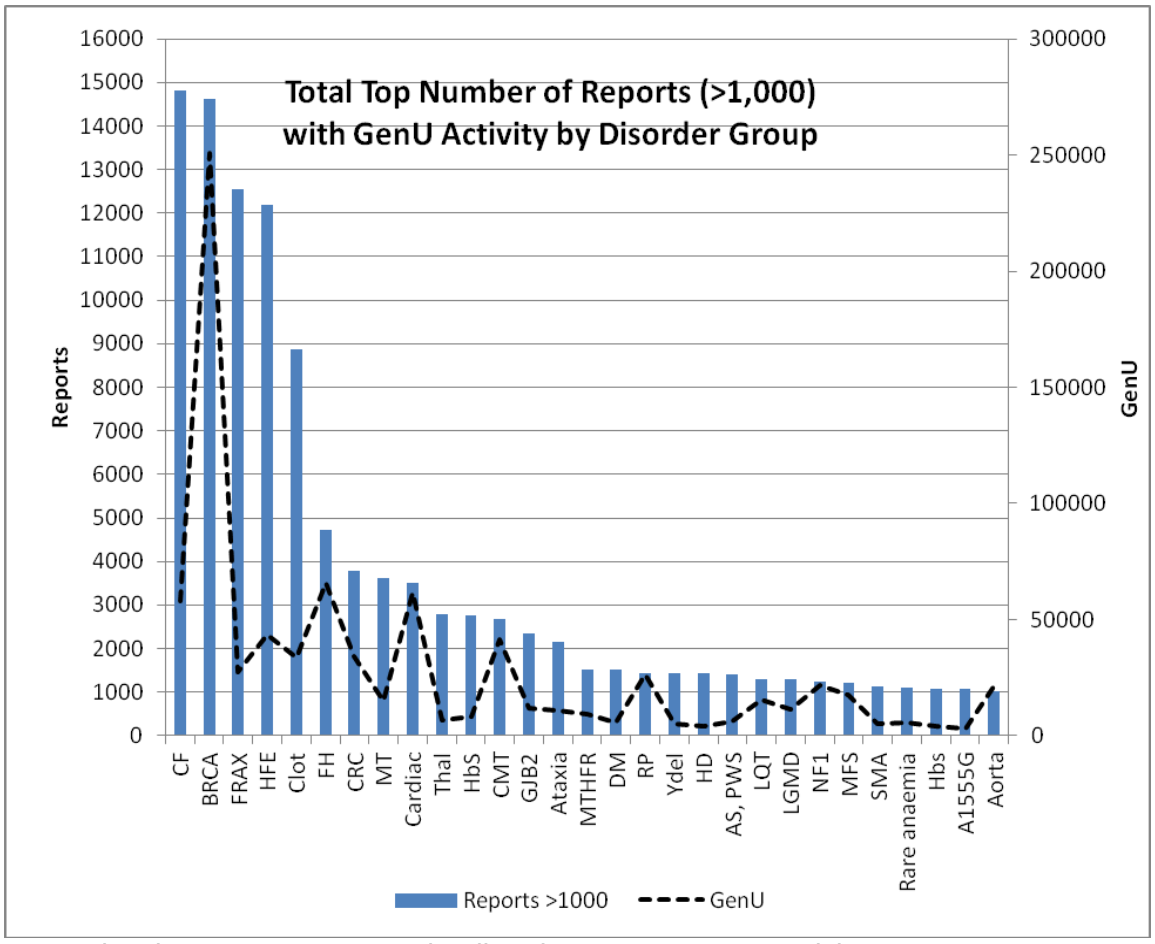
Table 2.1 Summary of Report Activity

2A Molecular (monogenic) Postnatal and Prenatal Test Activity

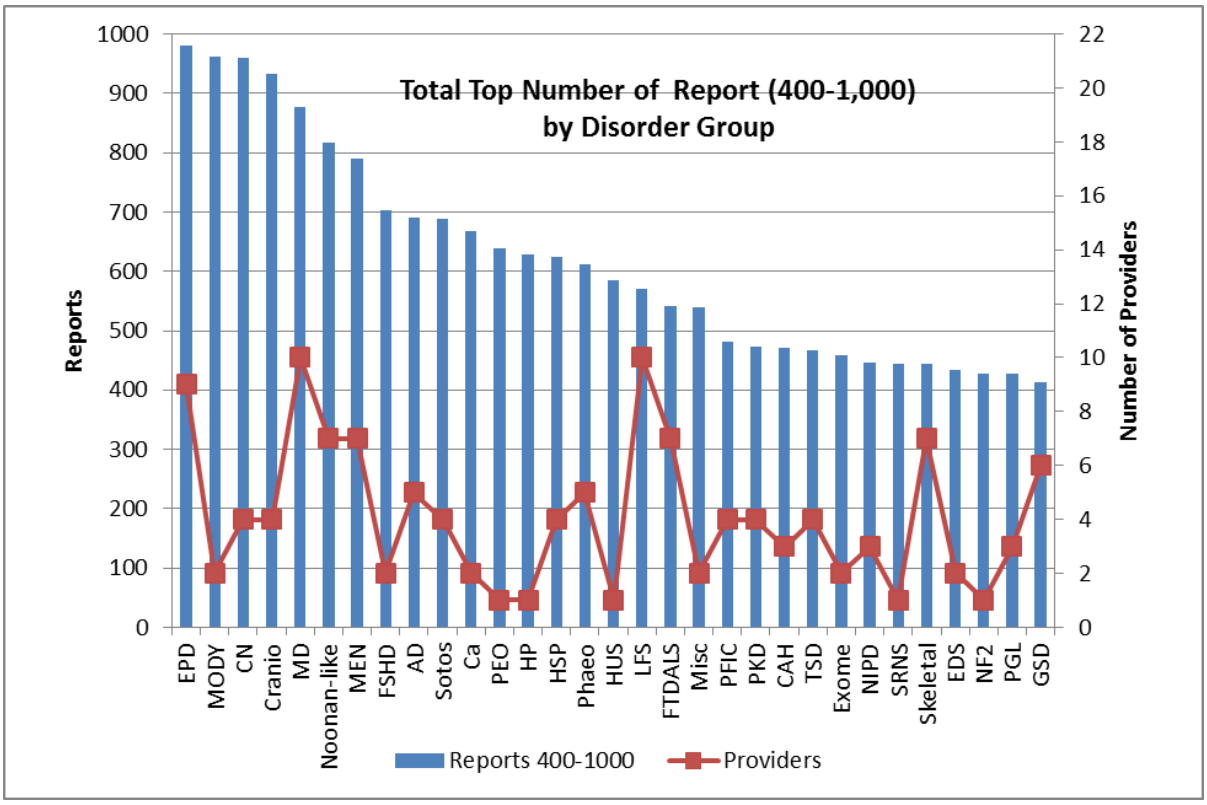
Keys provided at the end of the document (page 33)



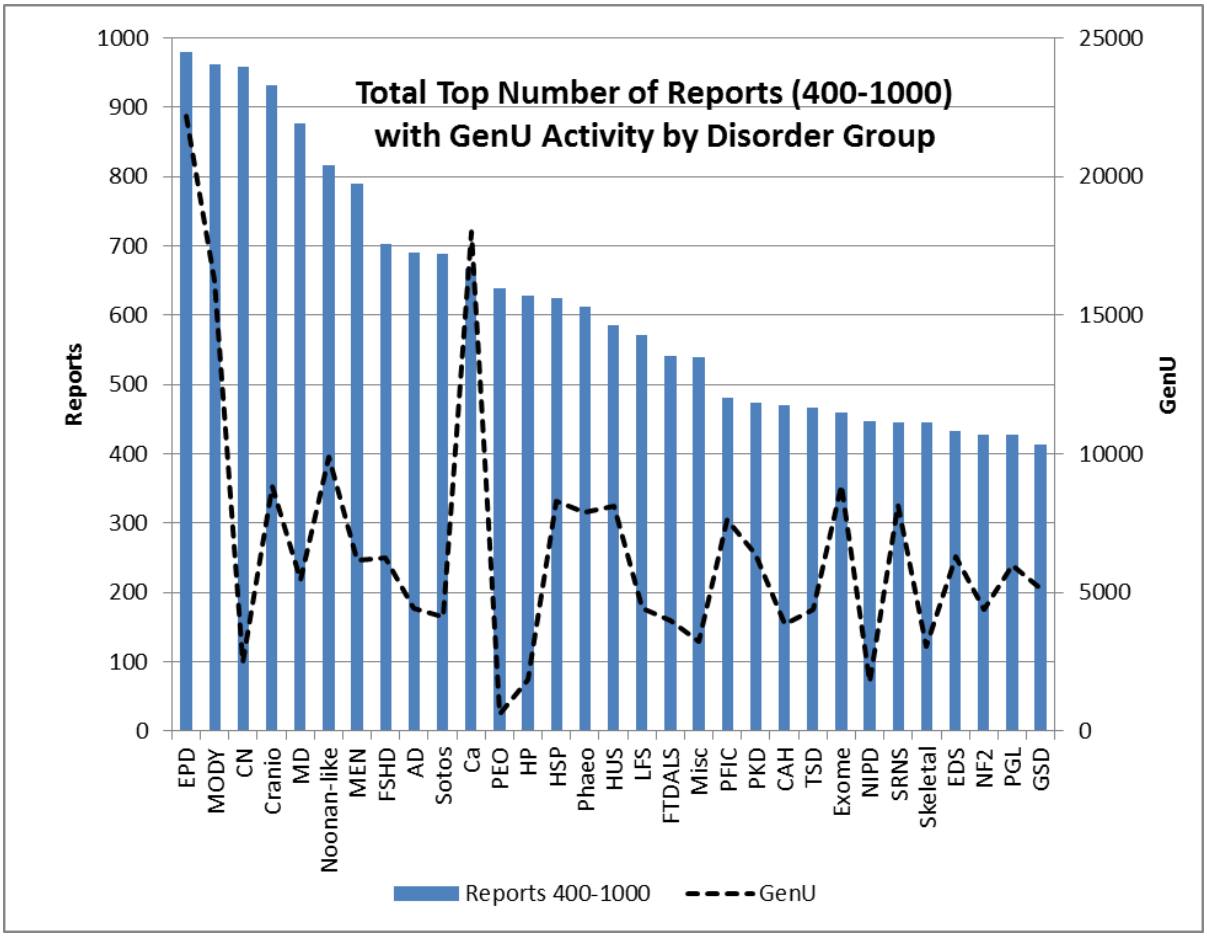
2.2 Molecular reports >1000 pa by disorder group & number of provider organisations



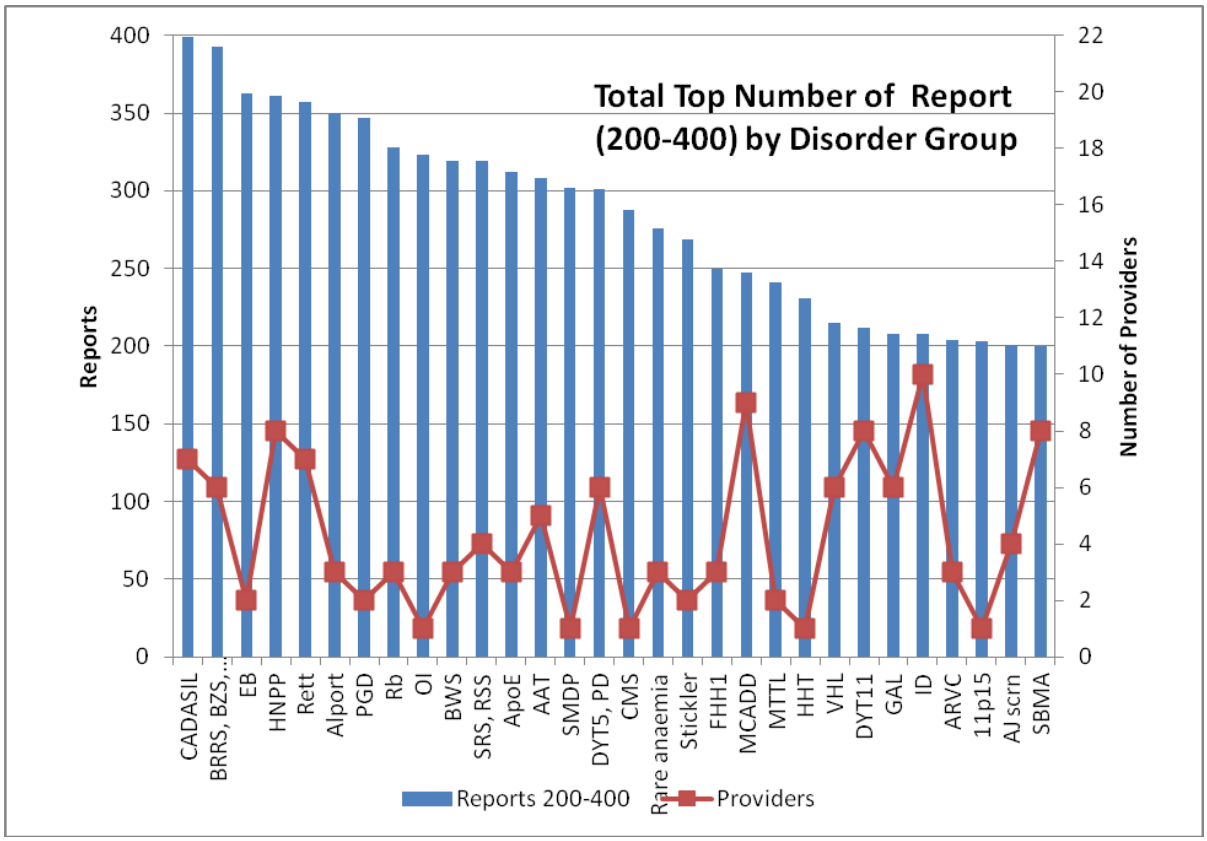
2.3 Molecular reports >1000 pa by disorder group & GenU activity



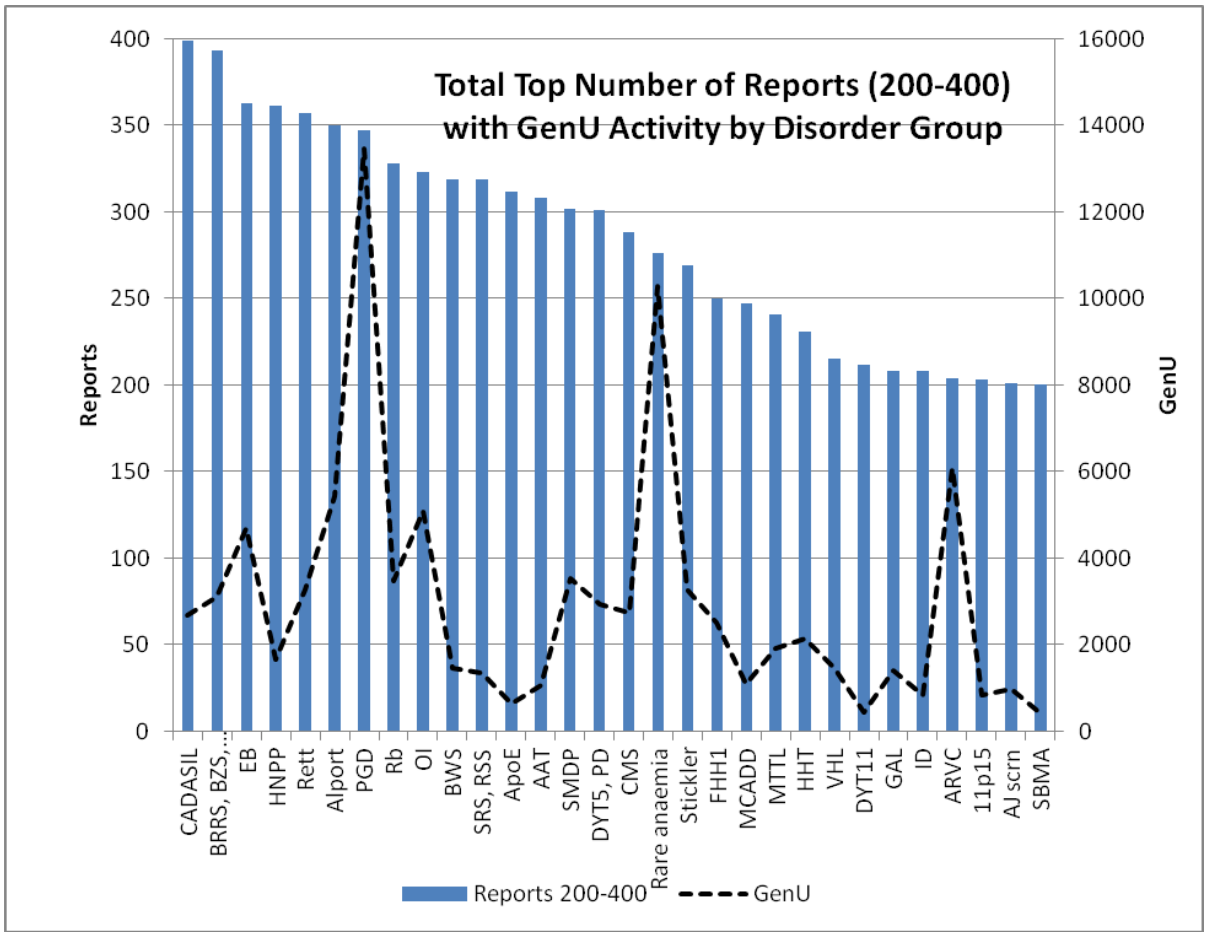
2.4 Molecular reports 400-1000 pa by disorder group & number of provider organisations



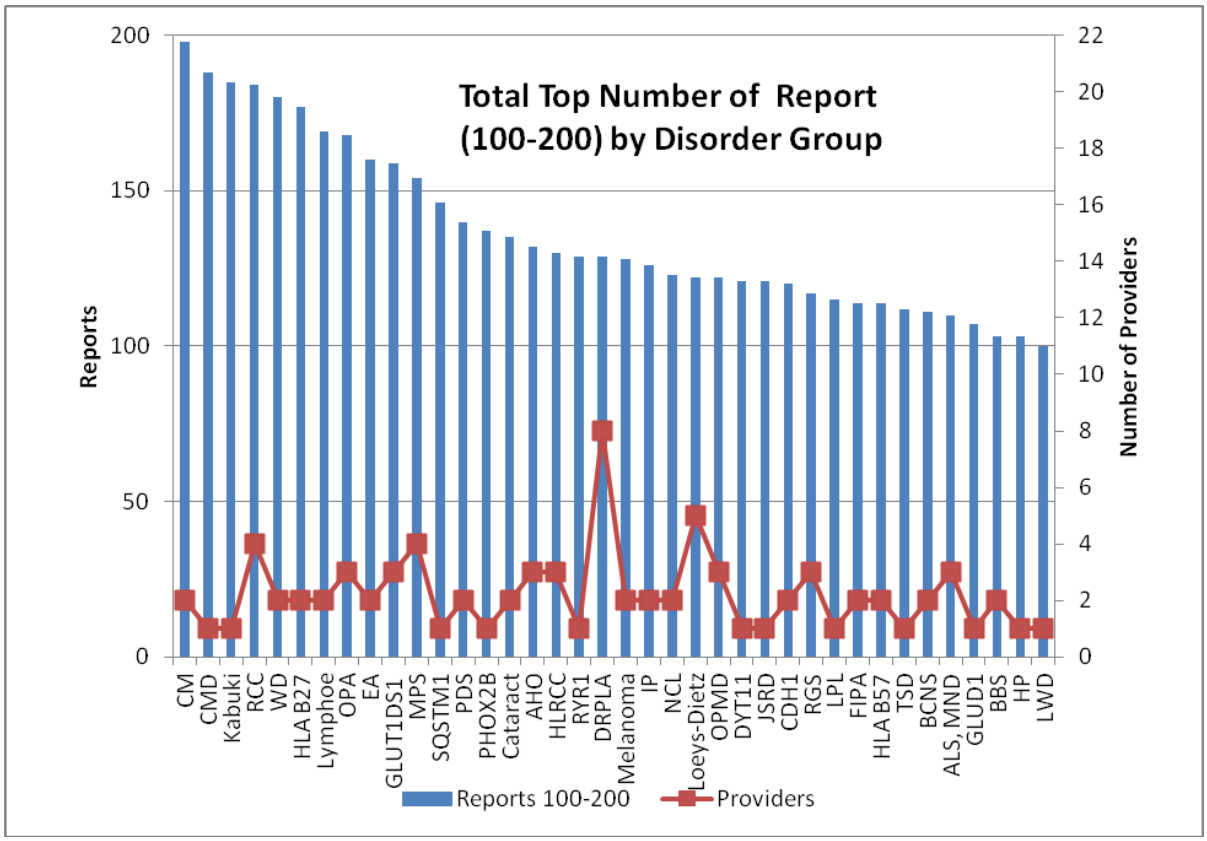
2.5 Molecular reports 400-1000 pa by disorder group & GenU activity



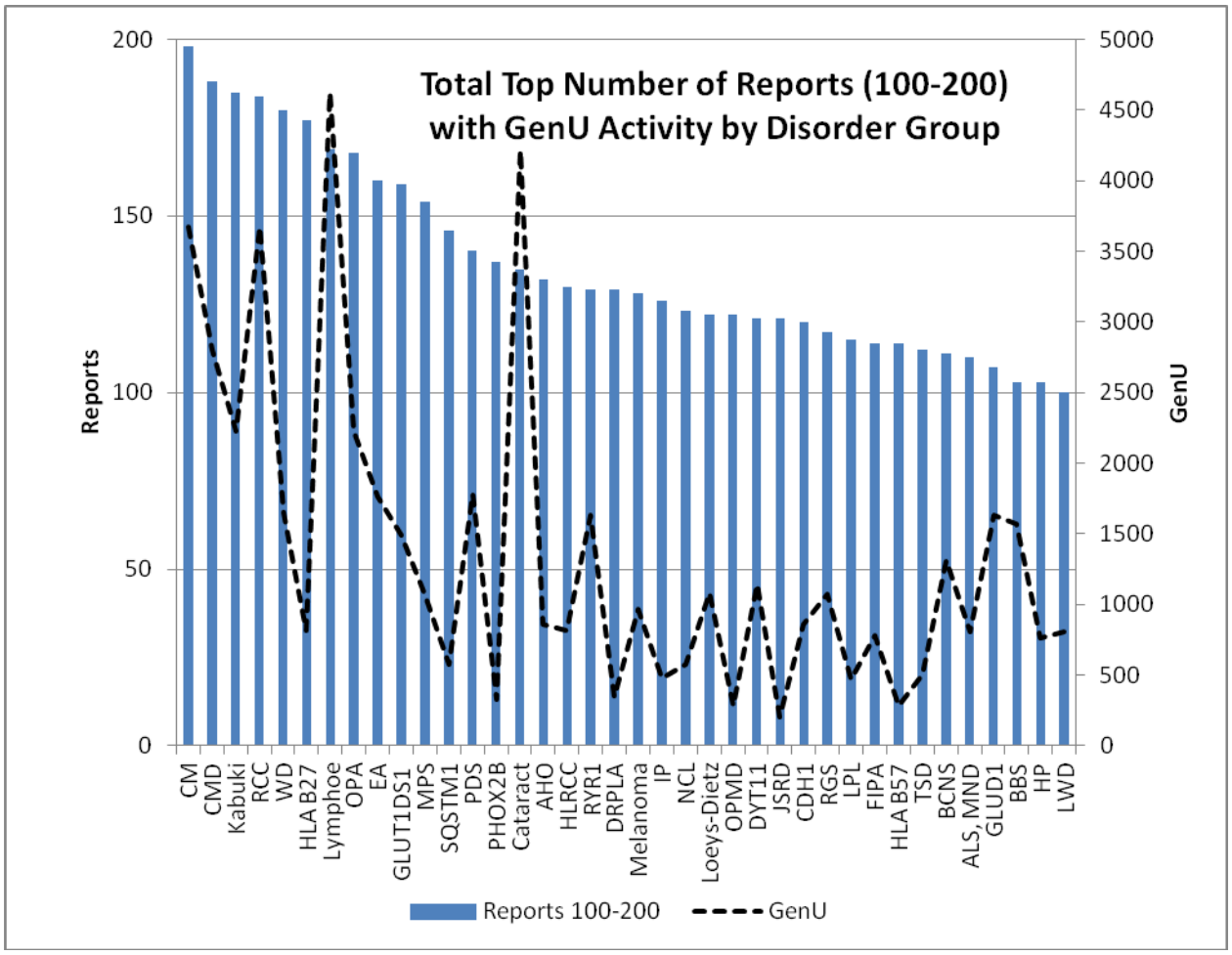
2.6 Molecular reports 200-400 pa by disorder group & number of provider organisations



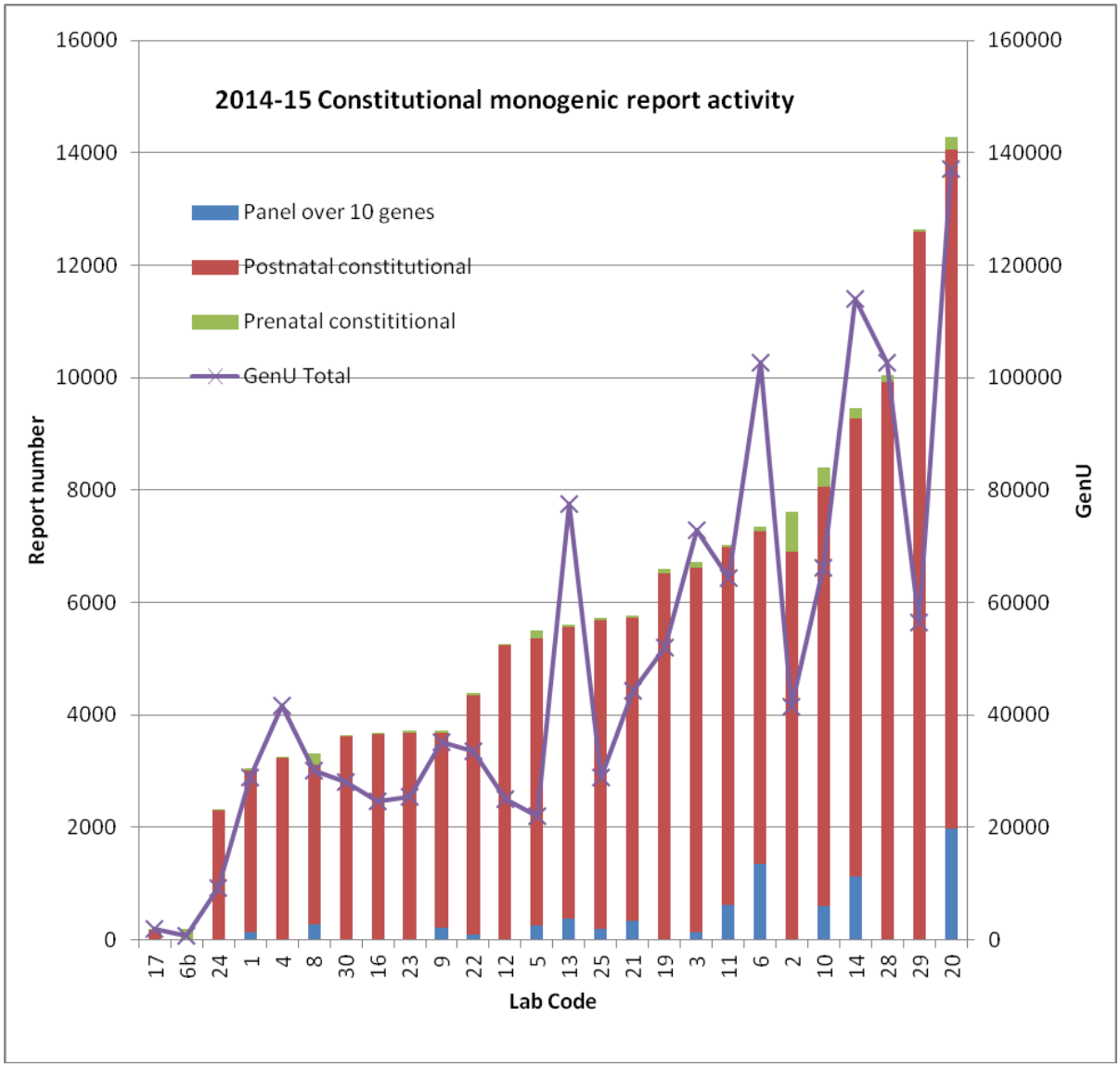
2.7 Molecular reports 200-400 pa by disorder group & GenU activity



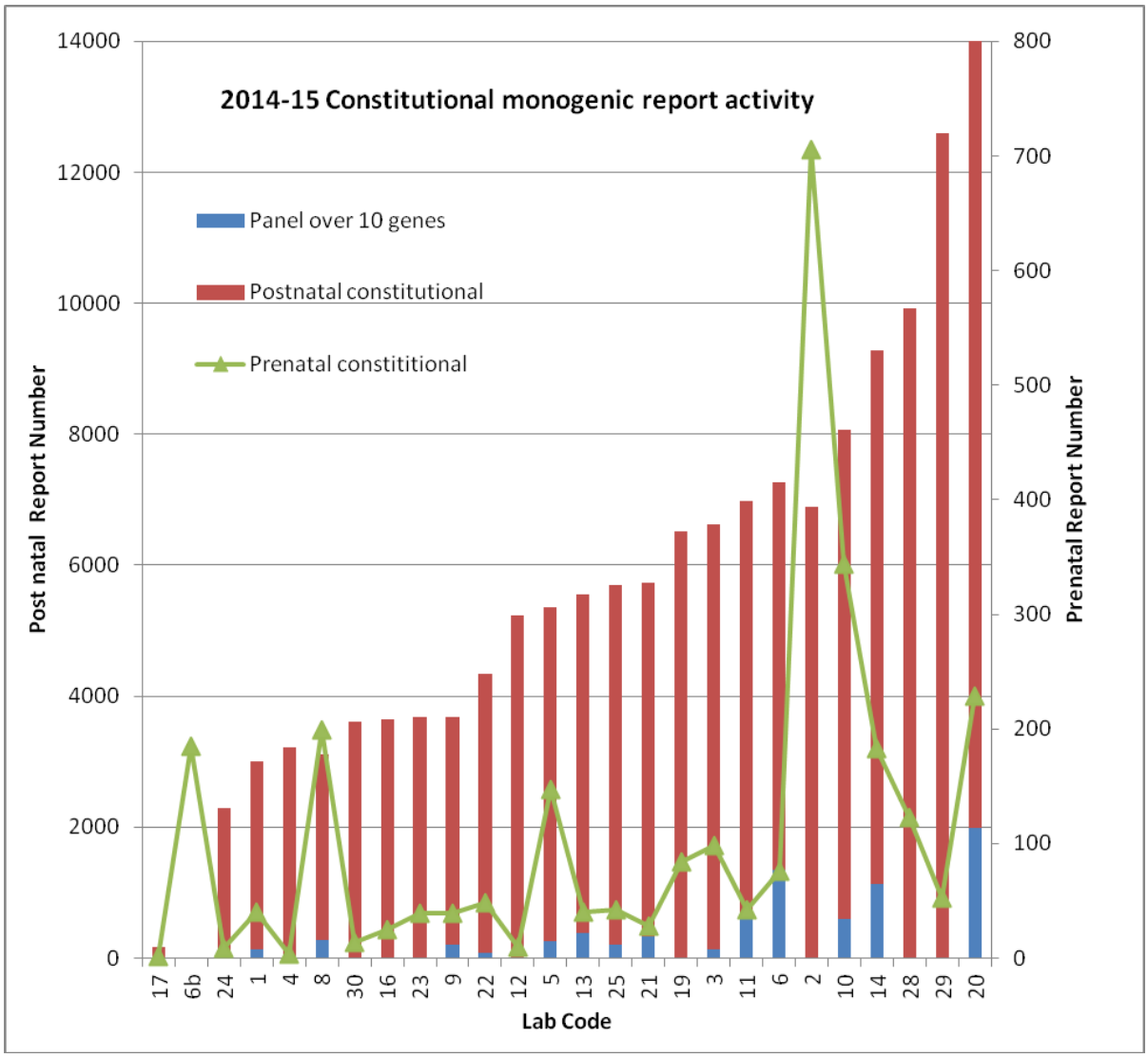
2.8 Molecular reports 100-200 pa by disorder group & number of provider organisations



2.9 Molecular reports 100-200 pa by disorder group & GenU activity

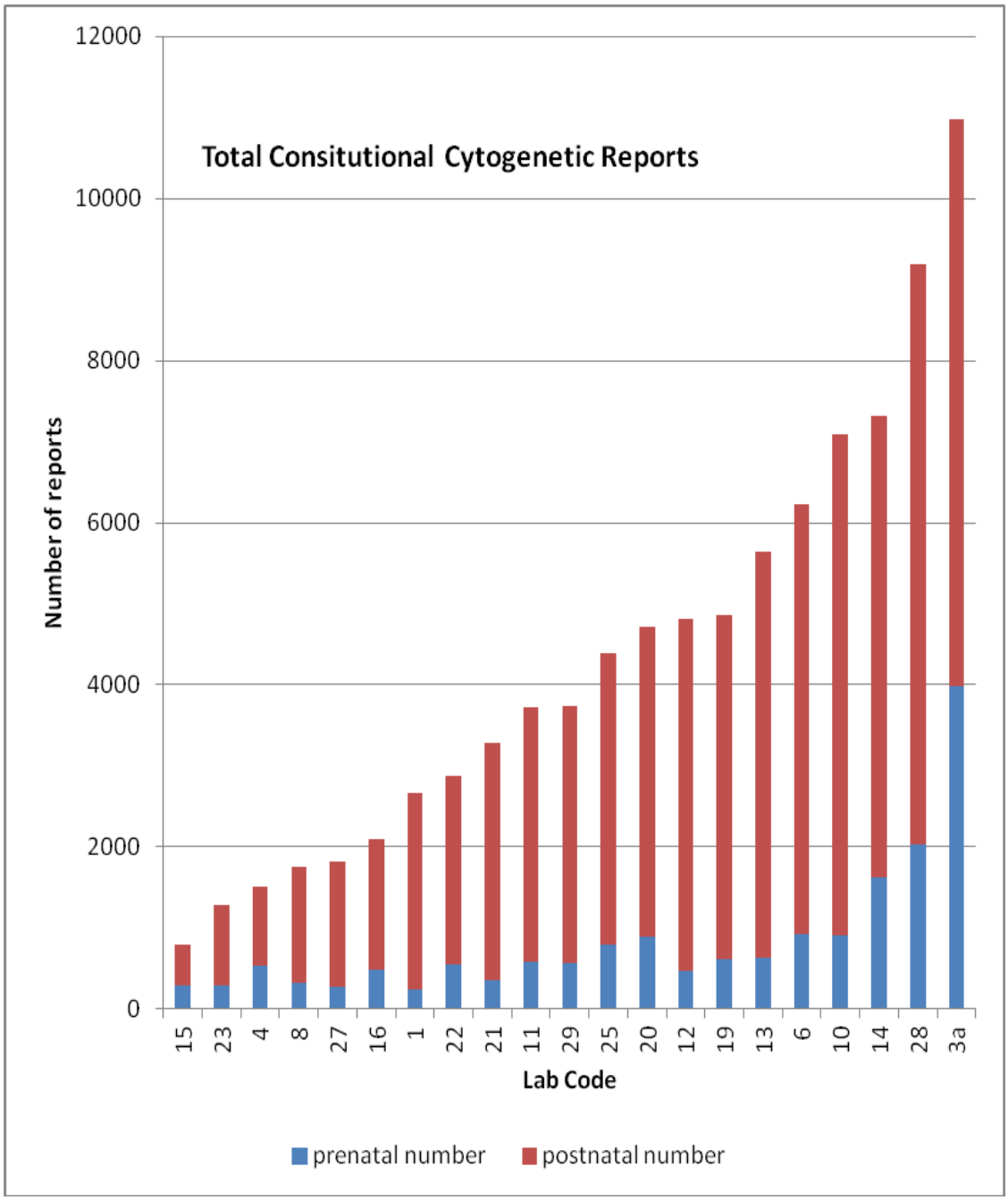


2.10a Total of molecular (monogenic) service reports for each of the 26 responding laboratories, ranked by sum of report numbers & showing GenU.

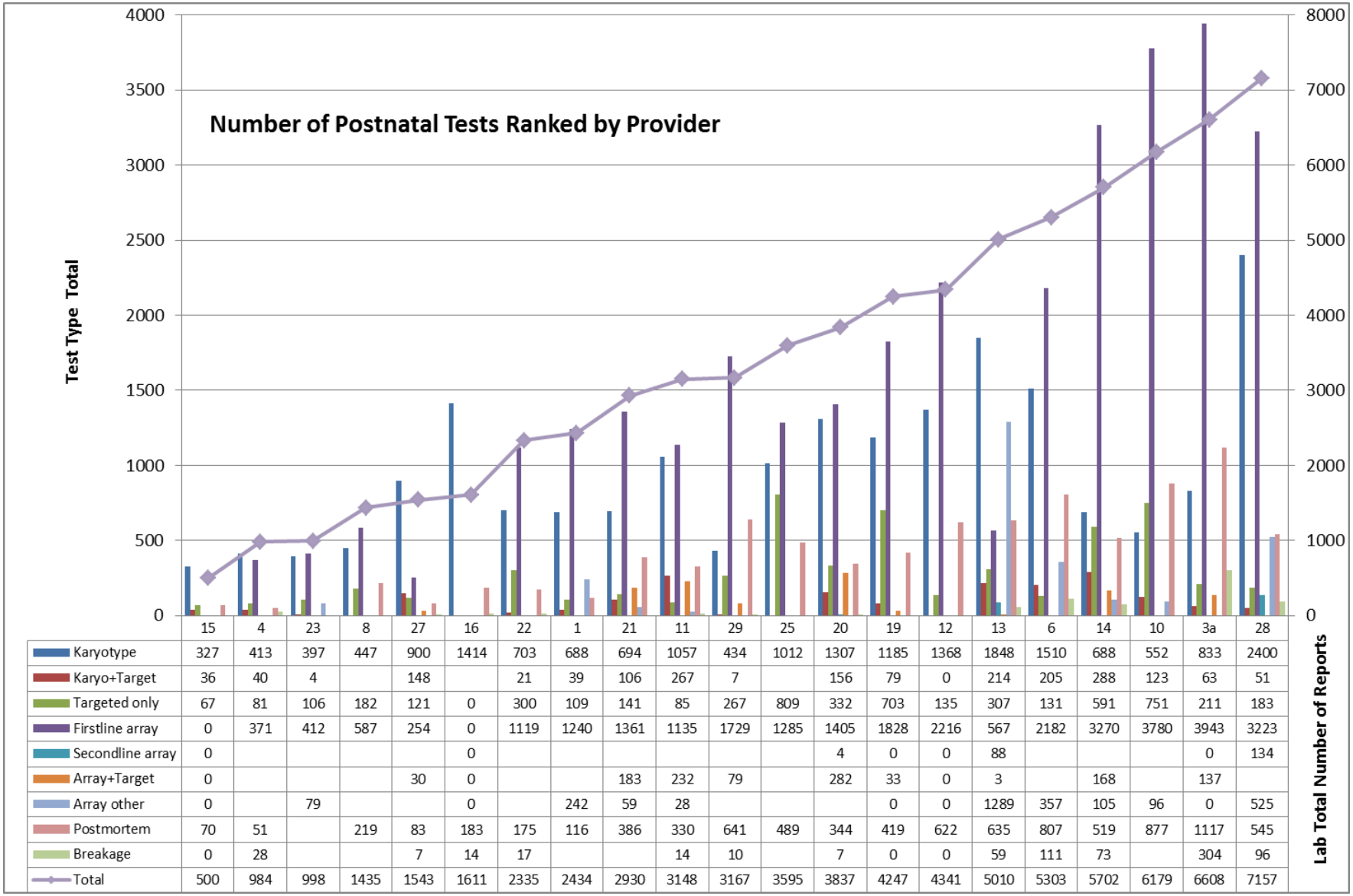


2.10b Total of molecular (monogenic) service reports for each of the 26 responding laboratories, ranked by sum of report numbers and showing prenatal reports separately

2B Cytogenetic Constitutional Disorder Activity

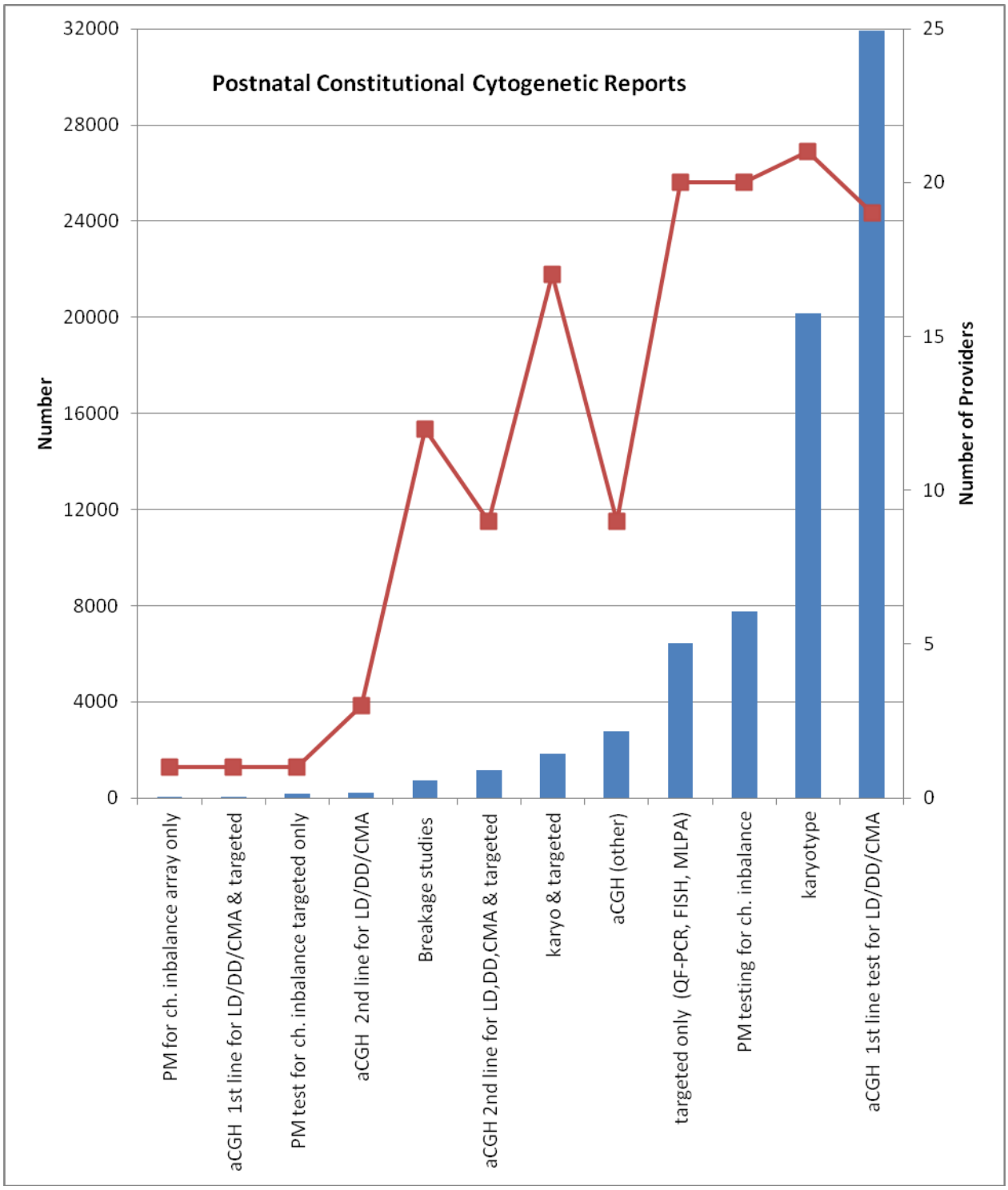


2.11 Total of prenatal and postnatal constitutional cytogenetic service reports for each of the 21 responding laboratories, ranked by sum of report numbers.



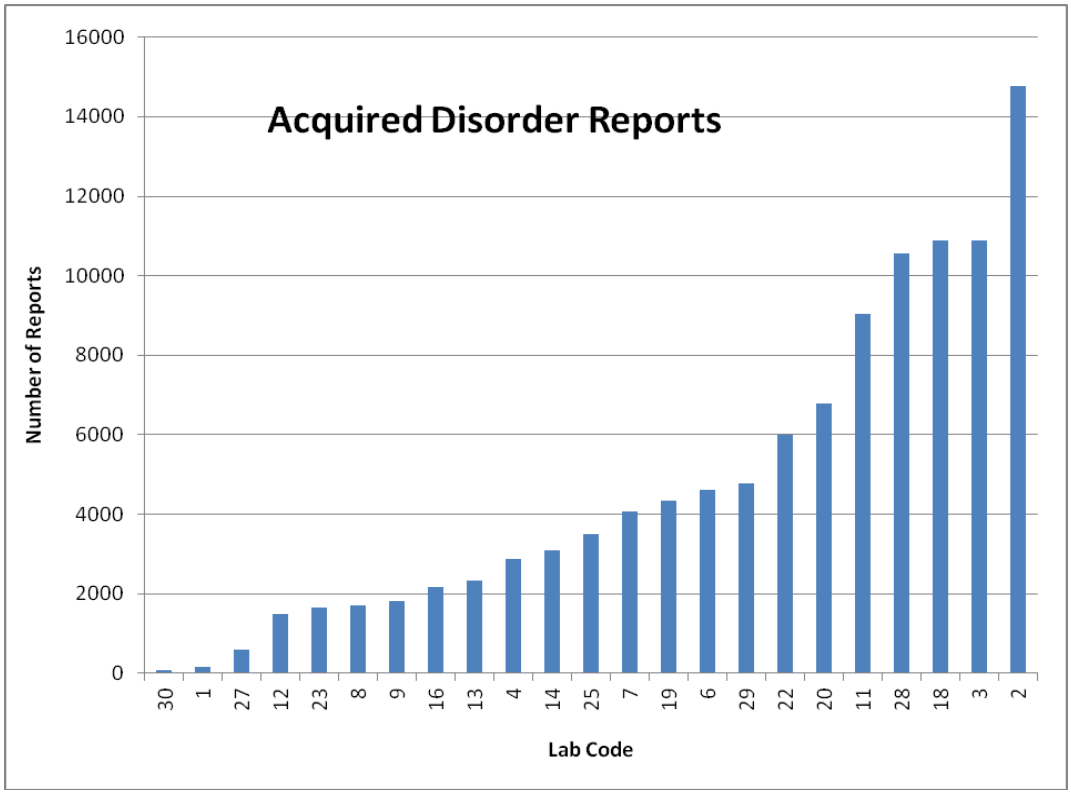
	15	4	23	8	27	16	22	1	21	11	29	25	20	19	12	13	6	14	10	3a	28
Karyotype	327	413	397	447	900	1414	703	688	694	1057	434	1012	1307	1185	1368	1848	1510	688	552	833	2400
Karyo+Target	36	40	4		148		21	39	106	267	7		156	79	0	214	205	288	123	63	51
Targeted only	67	81	106	182	121	0	300	109	141	85	267	809	332	703	135	307	131	591	751	211	183
Firstline array	0	371	412	587	254	0	1119	1240	1361	1135	1729	1285	1405	1828	2216	567	2182	3270	3780	3943	3223
Secondline array	0					0							4	0	0	88				0	134
Array+Target	0				30	0			183	232	79		282	33	0	3		168		137	
Array other	0		79			0		242	59	28				0	0	1289	357	105	96	0	525
Postmortem	70	51		219	83	183	175	116	386	330	641	489	344	419	622	635	807	519	877	1117	545
Breakage	0	28			7	14	17			14	10		7	0	0	59	111	73		304	96
Total	500	984	998	1435	1543	1611	2335	2434	2930	3148	3167	3595	3837	4247	4341	5010	5303	5702	6179	6608	7157

2.12 Chart to show postnatal cytogentic constitutional test activity.

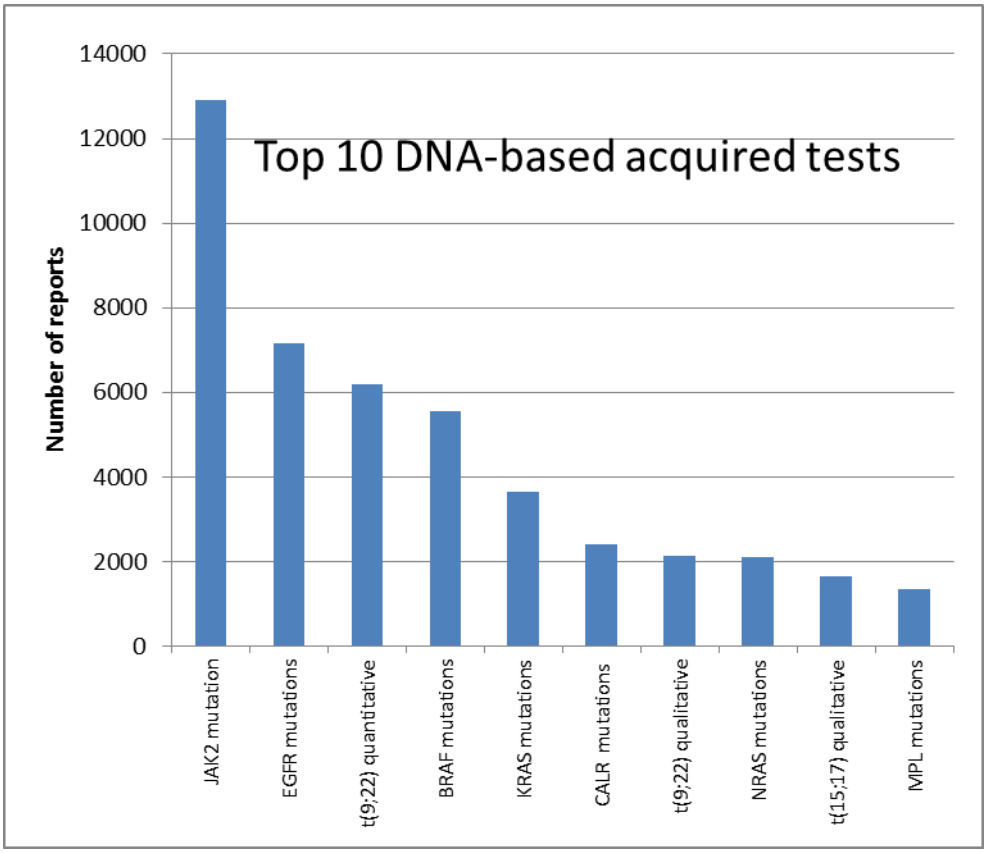


2.13 Chart indicating the top postnatal cytogenetic constitutional type of report. Array first line tests accounted for 44% reports.

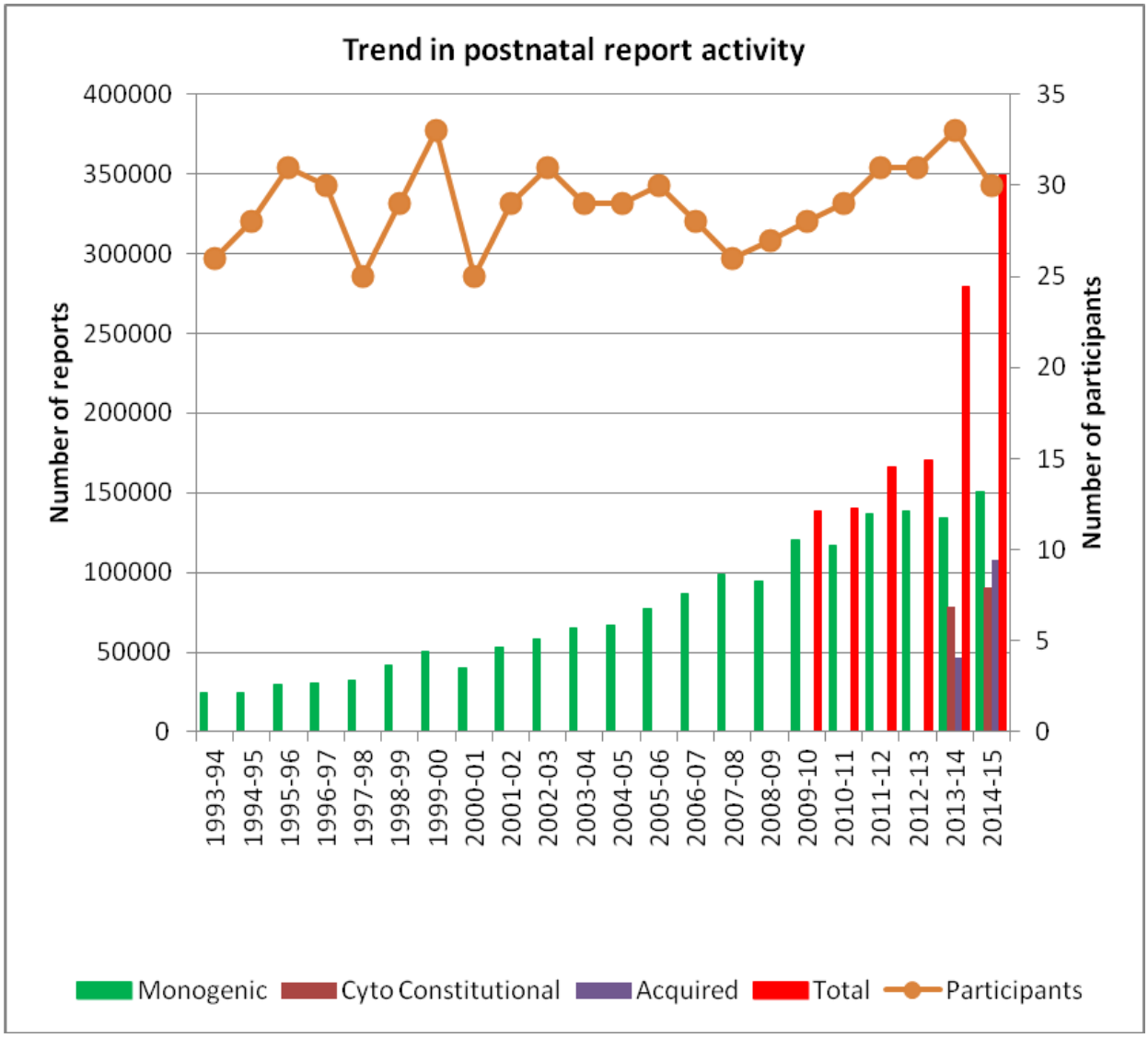
2 C Acquired Disorder Activity



2.14 Reports for acquired disorders were recorded by 23 organisations representing 28 laboratories



2.15 Top ten DNAbased tests for acquired disorders.
Different types of mutations for the same gene have been consolidated.



2.16 Chart showing the trend in postnatal report number with corresponding number of laboratories submitting data over past 22 years

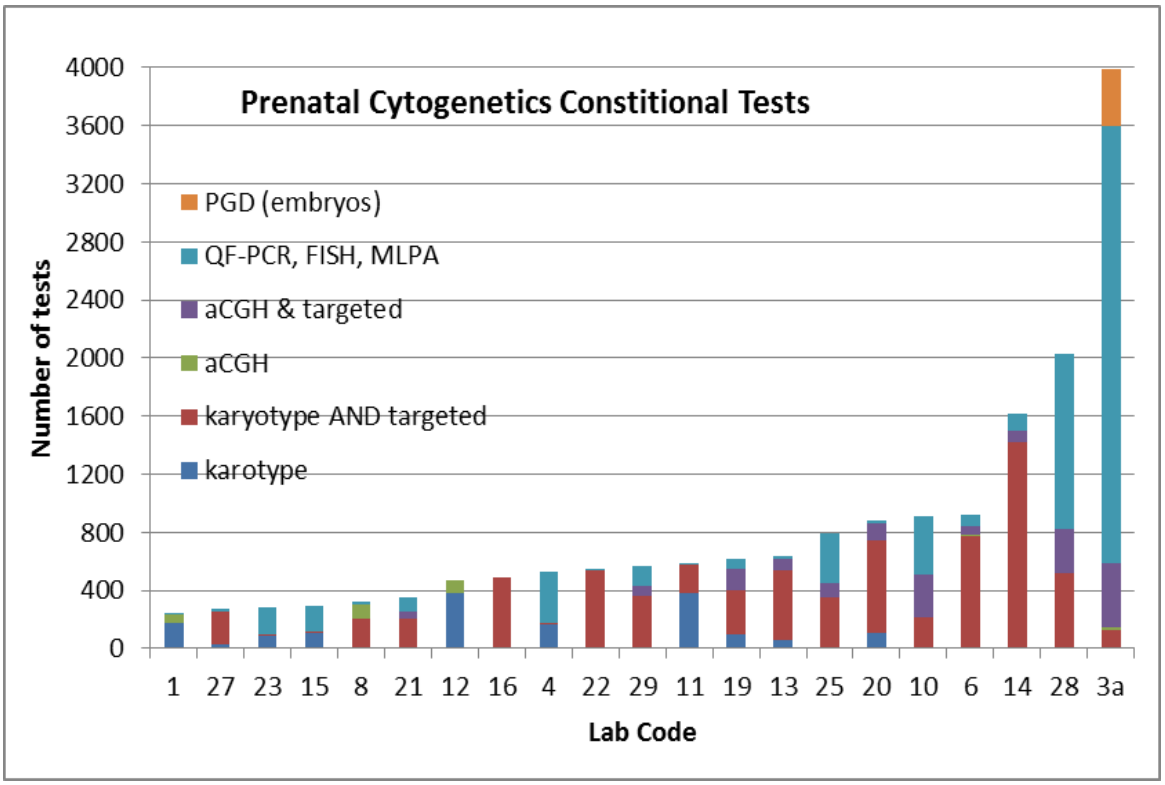
2 D PRENATAL REPORTS 2014-15

Year	Original total	No. of submitting 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
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	38	27	36	16	35	14	8	15	29	12	10	4	3	4	79	22
2013-14	2255	26	470	426	131	78	68	59	33	25	28	36	8	17	14	21	21	40	13	9	5	6	4	0	37
2014-15	2826	27	447	817	152	180*	71	232*	91	29	36	34	13	26	*	18	17	*	17	6	8	8			

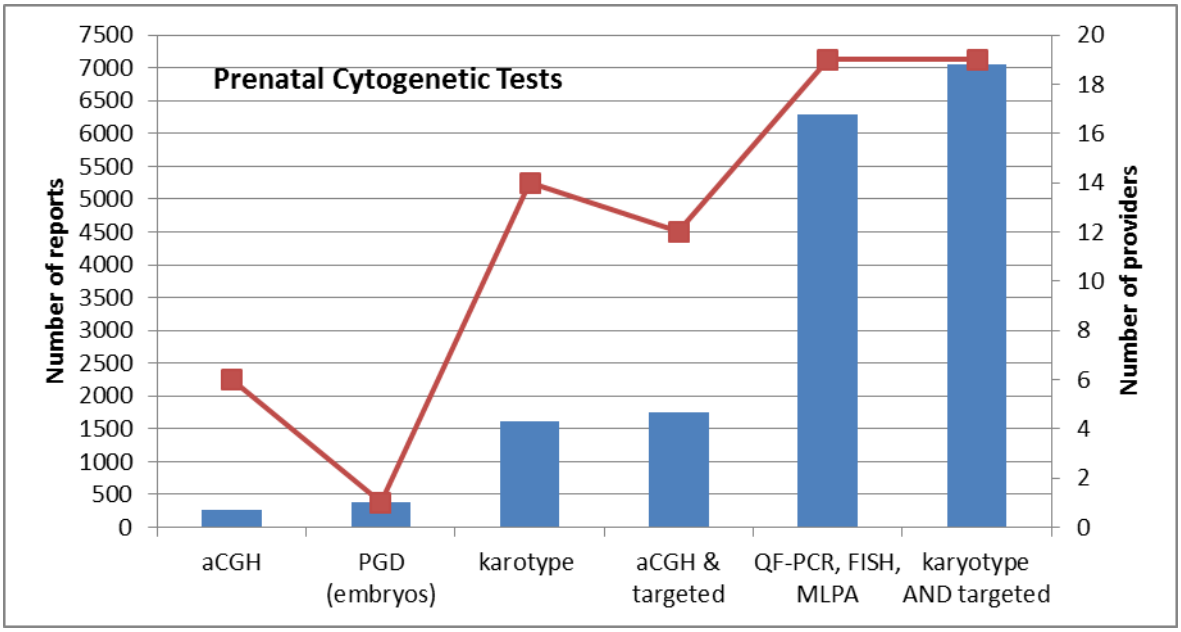
Corrected CF prenatals (1998-99 to 2007-08) included some FEB tests
Totals includes PGD and identity/maternal cell contamination
2014-15 some data collected under wider grouping

2.17 Table summarising prenatal diagnosis activity for single gene disorders

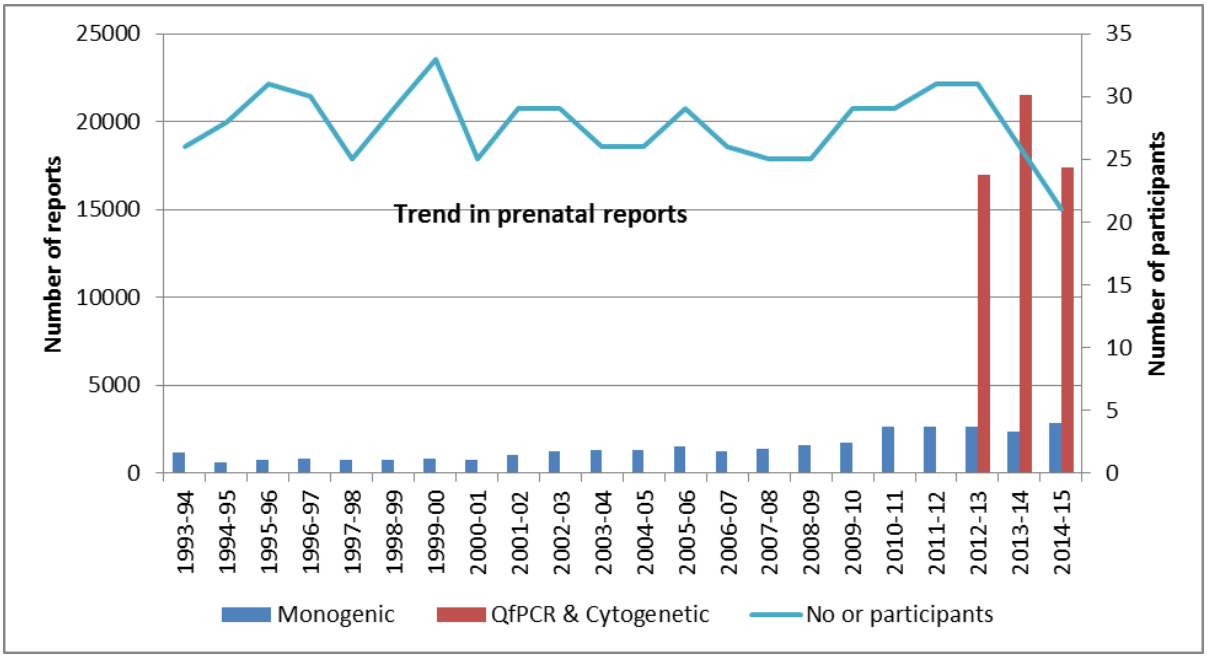
Cytogenetic Prenatal Reports



2.18 Prenatal cytogenetic report activity by provider.
Cytogenetic prenatal reports were recorded by 21 providers.



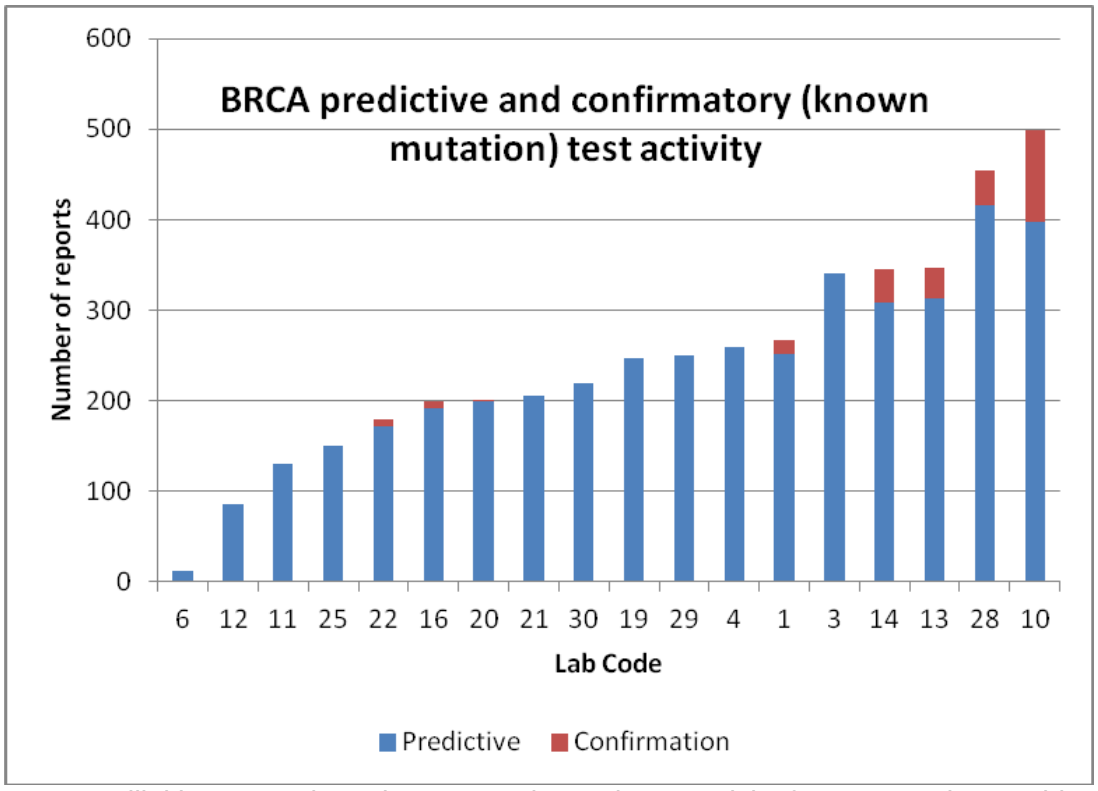
2.19 Prenatal cytogenetic report activity by available test breakdown
Karyotype & targeted testing accounted for reports accounted for 41% reports, QfPCR, FISH, MLPA 36% and array first line plus targeted for 10%.



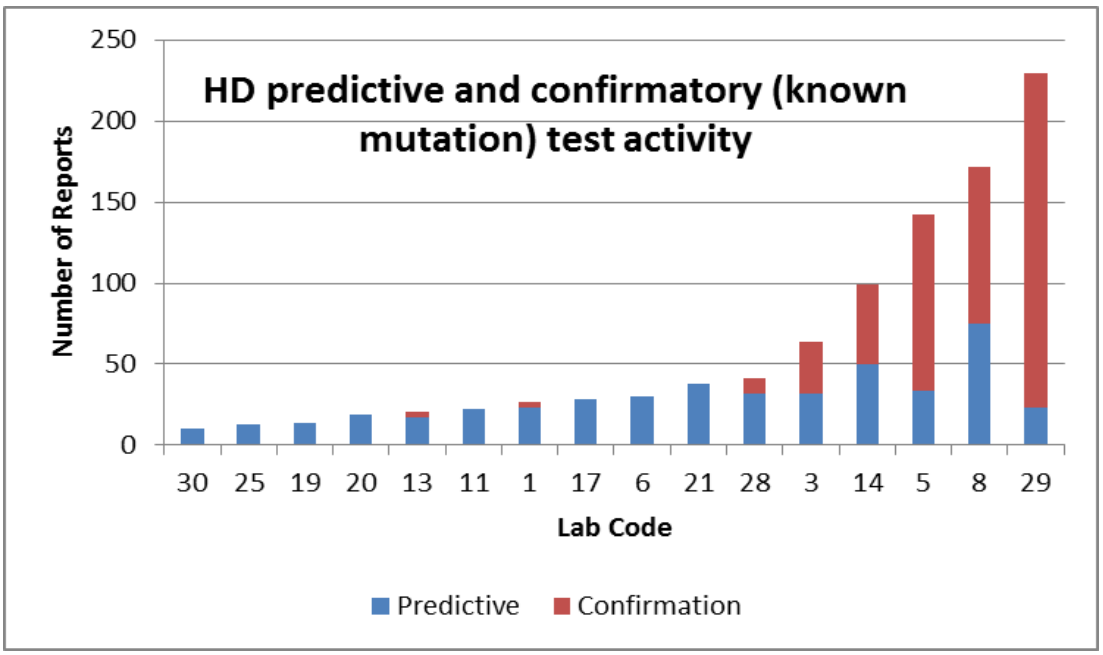
2.20 Chart to show the trend in prenatal constitutional report numbers with corresponding number of laboratories submitting data over past 22 years.

3 PREDICTIVE TESTS 2014-15

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).



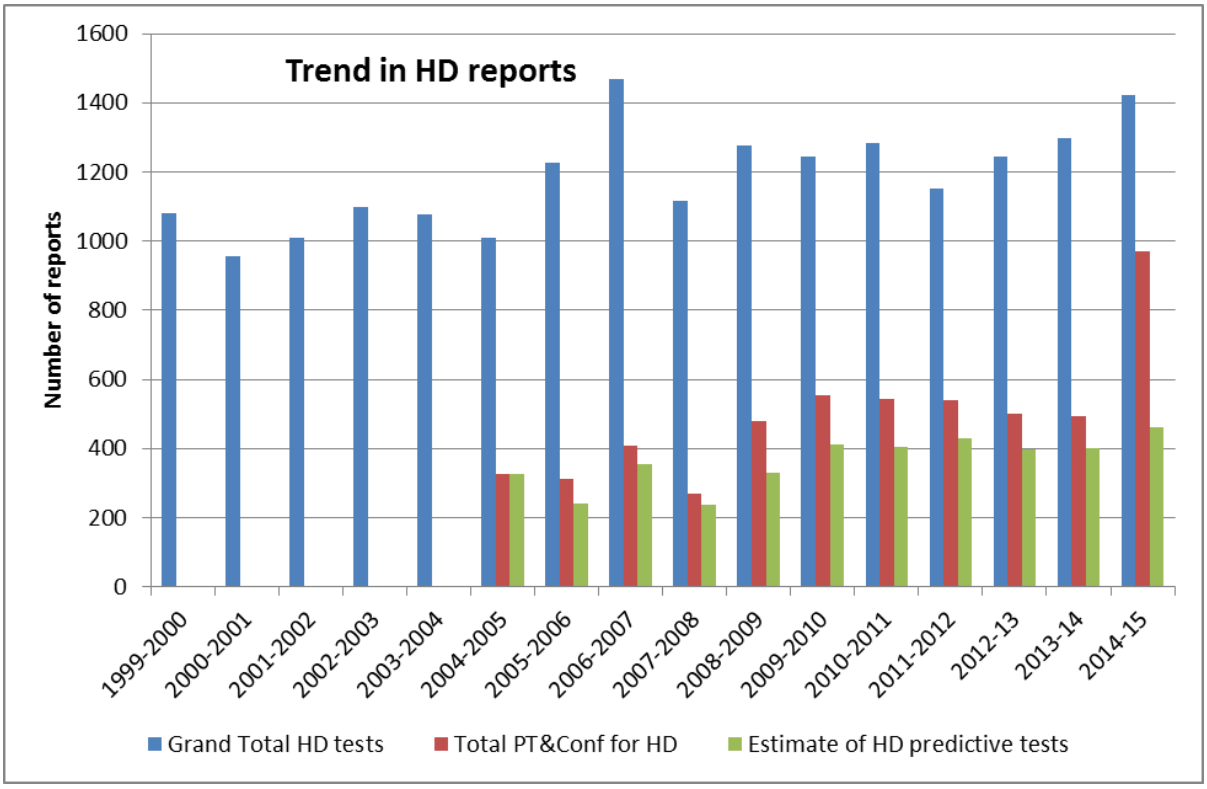
3.1 Familial breast and ovarian cancer (BRCA) test activity for 2014-15 by provider



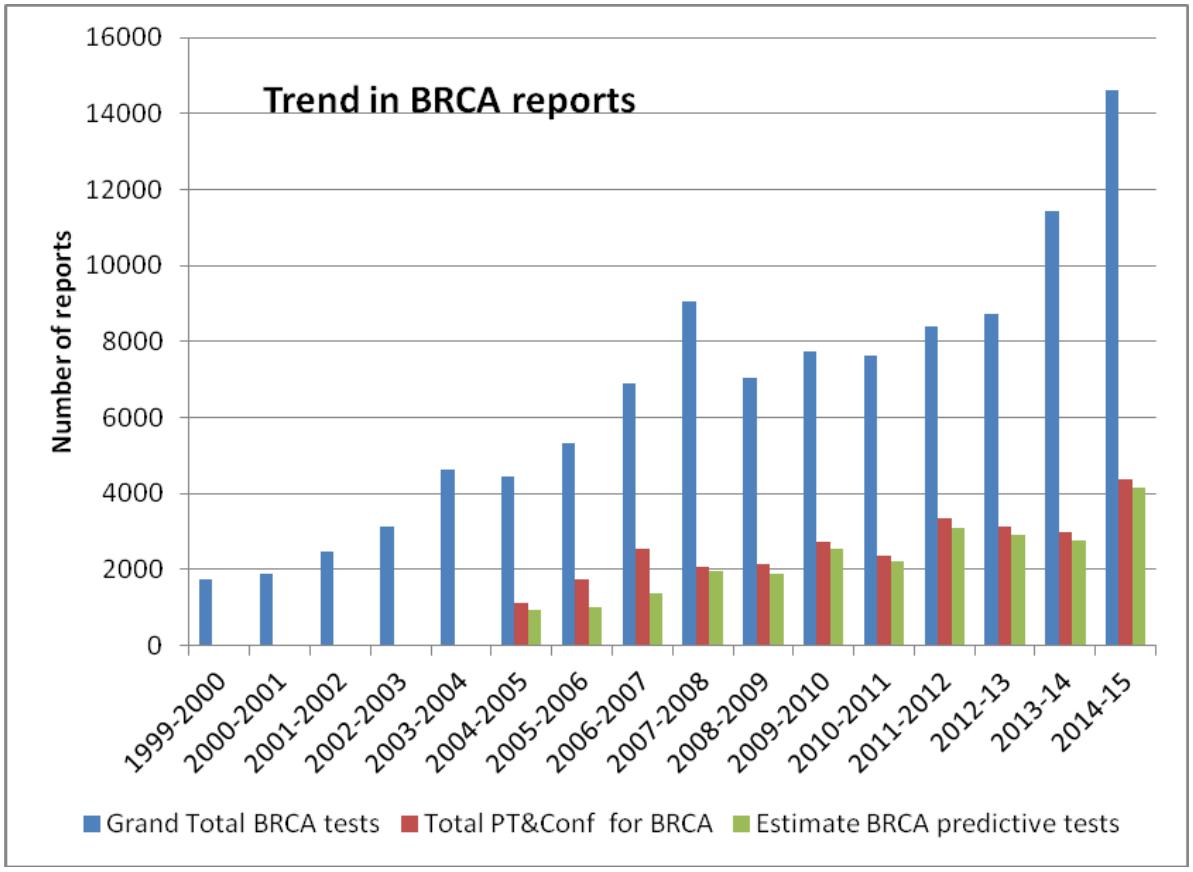
3.2 Huntington disease (HD) test activity for 2014-15 by provider

ACGS 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**	2013 2014	**2014 2015**
No. HD lab returns					17	15	13	11	13	10	9	13	14	10	16	**16**
No. BRCA lab returns					18	18	13	13	14	10	15	14	16	11	18	**18**
Grand Total HD tests	1080	956	1010	1099	1079	1009	1226	1469	1118	1278	1245	1283	1153	1246	1299	**1422**
Total PT&Conf for HD						328	311	407	268	478	554	543	539	499	493	**969**
Grand Total BRCA tests	1740	1889	2469	3142	4635	4455	5309	6906	9048	7041	7733	7617	8381	8712	11434	**14633**
Total PT&Conf for BRCA						1124	1746	2548	2068	2141	2739	2364	3367	3145	2974	**4391**
Estimate predictive of total HD tests (%)						328 (33%)	242 (20%)	354 (24%)	237 (21%)	330 (26%)	413 (33%)	404 (31%)	431 (37%)	398 (32%)	400 (31%)	**460 (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%)	2749 (24%)	**4150 (28%)**

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



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



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

4 REPORTING TIMES

Caveats
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.

% compliance with TAT	3 day	2 week (14 day)	4 week (28 day)	8 week (56 day)	16 week (>10 gene panels)
min	25	25	12	8	2
max	100	100	100	100	100
mean	**92**	**87**	**88**	**79**	**73**
median	100	94	94	86	83
count	273	423	569	557	67

4.1 Summary table of combined monogenic test performance as a percentage against the target reporting time category.

Table 4.2 below is the trend analysis for a select range of molecular 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 reporting time targets.

Note for some categories the scope of tests has been extended. These are marked **.

% compliance with TAT	3 working days	10 calendar days	14 calendar days	28 calendar days
min	72	42	33	8
max	100	100	100	100
mean	**98**	**92**	**80**	**85**
median	99	93	83	83
count	33	47	52	115

4.3 Summary table of combined constitutional cytogenetic test performance as a percentage against the target reporting time category.

Table 4.4 below shows the turnaround performance by constitutional cytogenetic test category.

		Reporting time																%											
Gene(s)	test	56 days				14 days				28 days				Urgent 3 days				56 days			14 days			28 days			Urgent 3 days		
		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	17	383	87	8	1	10	6	14	6	16	11	12	2	40	3	17	17	100	72	76	100	96	88	99	96	50	100	92
2013-14		22	96	43	5	2	15	7	12	4	22	10	10	2	3	3	13	22	100	78	75	98	95	94	100	98	83	100	96
2012-13		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	14	79	43	4	3	12	8	8	8	31	16	11	2	40	3	11	58	100	87	86	100	95	71	100	90	25	100	87
2013-14		21	39	29	3	5	13	7	9	6	20	13	9	1	3	2	7	94	100	98	85	100	94	78	100	90	80	100	97
2012-13		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	11	48	33	10	6	12	9	11	7	27	15	14	3	40	4	10	40	100	82	67	100	91	84	100	94	56	100	93
2013-14		15	47	35	13	6	12	8	10	9	20	16	9	1	4	2	6	33	33	78	75	99	95	77	100	93	80	100	96
2012-13		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

		Reporting time																%											
Gene(s)	test	56 days				14 days				28 days				Urgent 3 days				56 days			14 days			28 days			Urgent 3 days		
		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
IT15	HUNTINGTON DISEASE	30	30	30	1	4	11	8	11	6	23	15	12	1	40	2	12	100	100	100	89	100	96	74	100	89	100	100	100
2013-14		33	33	33	1	5	13	9	10	8	20	13	8	1	4	2	9	100	100	28	80	93	94	90	100	97	75	100	98
2012-13		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	36	36	36	1	8	36	15	6	8	138	35	6	1	40	3	8	88	88	88	75	100	91	20	96	71	94	100	99
2013-14		34	34	34	1	2	11	8	5	6	28	16	5	2	4	3	6	93	93	27	50	98	84	67	96	80	33	100	77
2012-13		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
SNRPN	AS&PWS	28	43	35	2	7	12	9	5	9	22	14	7	2	40	3	5	85	89	87	75	99	92	85	98	92	71	100	94
2013-14		10	33	16	5	6	16	11	2	8	41	15	11	3	3	3	2	100	100	26	63	95	85	66	100	91	100	100	100
2012-13		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		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				100	25	9

		Reporting time																%											
Gene(s)	test	56 days				14 days				28 days				Urgent 3 days				56 days			14 days			28 days			Urgent 3 days		
		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
CMT**	CHARCOT-MARIE-TOOTH DISEASE,S	17	383	87	8	1	10	6	14	6	16	11	12	2	40	3	17	73	100	95	67	100	84	31	100	81	75	100	88
2013-14		32	43	37	2	13	14	14	2	5	26	14	5	2	2	2	1	71	82	77	74	75	75	64	100	89	100	100	100
2012-13		21	48	34	2	8	13	11	2	8	13	10	2					79	96	87	67	74	71	100	100	100			
2011-12		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	SPINAL MUSCULAR ATROPHY	47	47	47	1	8	16	10	8	9	24	15	10	2	40	3	10				70	100	91	75	100	88	43	100	74
2013-14		24	24	24	1	7	9	8	5	7	20	12	6	2	5	3	10	100	100	100	91	100	94	85	100	93	50	100	87
2012-13		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		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	FAMILIAL BREAST & OVARIAN CANCER	21	90	46	16	8	11	10	8	11	97	28	10	3	40	3	1	12	100	72	73	99	88	18	99	77	100	100	100
2013-14		11	148	48	15	8	12	9	7	8	31	17	10					15	100	77	74	98	91	77	100	91			
2012-13		26	144	48	14	7	16	10	9	10	25	15	9					13	99	75	66	98	87	78	97	90			
2011-12		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)	test	56 days				14 days				28 days				Urgent 3 days				56 days			14 days			28 days			Urgent 3 days		
		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
CRC**	LYNCH SYNDROME	22	76	40	14	6	13	9	9	8	130	30	8					48	100	79	52	100	86	45	100	85			
2013-14		28	245	60	11	6	11	8	8	11	38	20	8					25	100	76	76	97	92	39	100	82			
2012-13		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						
Cx26	GAP JUNCTION PROTEIN, b-2	14	39	29	7	6	20	13	4	20	35	25	4	2	40	2	3	33	100	85	67	100	86	30	92	65	100	100	100
2013-14		25	43	33	6	10	15	13	2	15	38	22	4	3	3	3	1	53	99	87	63	90	77	39	91	74	80	80	80
2012-13		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	100	100	33				1	1	1
Mt	MITOCHONDRIAL MYOPATHY	31	198	90	3	4	18	12	3	10	53	21	8	4	40	4	1	50	100	81	75	100	89	63	98	85	100	100	100
2013-14		25	25	25	1	9	10	10	2	18	30	24	2	3	3	3	1	99	99	99	98	100	99	50	91	70	100	100	100
2012-13		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
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						

		Reporting time																%											
Gene(s)	test	56 days				14 days				28 days				Urgent 3 days				56 days			14 days			28 days			Urgent 3 days		
		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
a-globin, b-globin	ABNORMAL HAEMOGLOBINS	40	40	40	1	6	6	6	1	14	29	21	4	5	40	7	2	33	33	33	80	80	80	89	100	93	50	50	50
2013-14										12	12	12	1											83	83	83			
2012-13						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	47	47	47	1	7	9	8	2	13	20	17	3	2	40	3	3	25	25	25	60	86	73	91	100	97	87	100	95
2013-14		28	28	28	1					13	19	16	2	1	4	3	4	92	92	92	100	100	100	85	100	93	68	100	88
2012-13						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	31	33	32	2	7	7	7	2	10	11	10	2	2	40	3	2	86	100	93	93	100	97	100	100	100	100	100	100
2013-14		31	31	31	1	7	7	7	1	7	7	7	1	2	2	2	1	91	100	96	89	89	89	98	100	99	100	100	100
2012-13		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

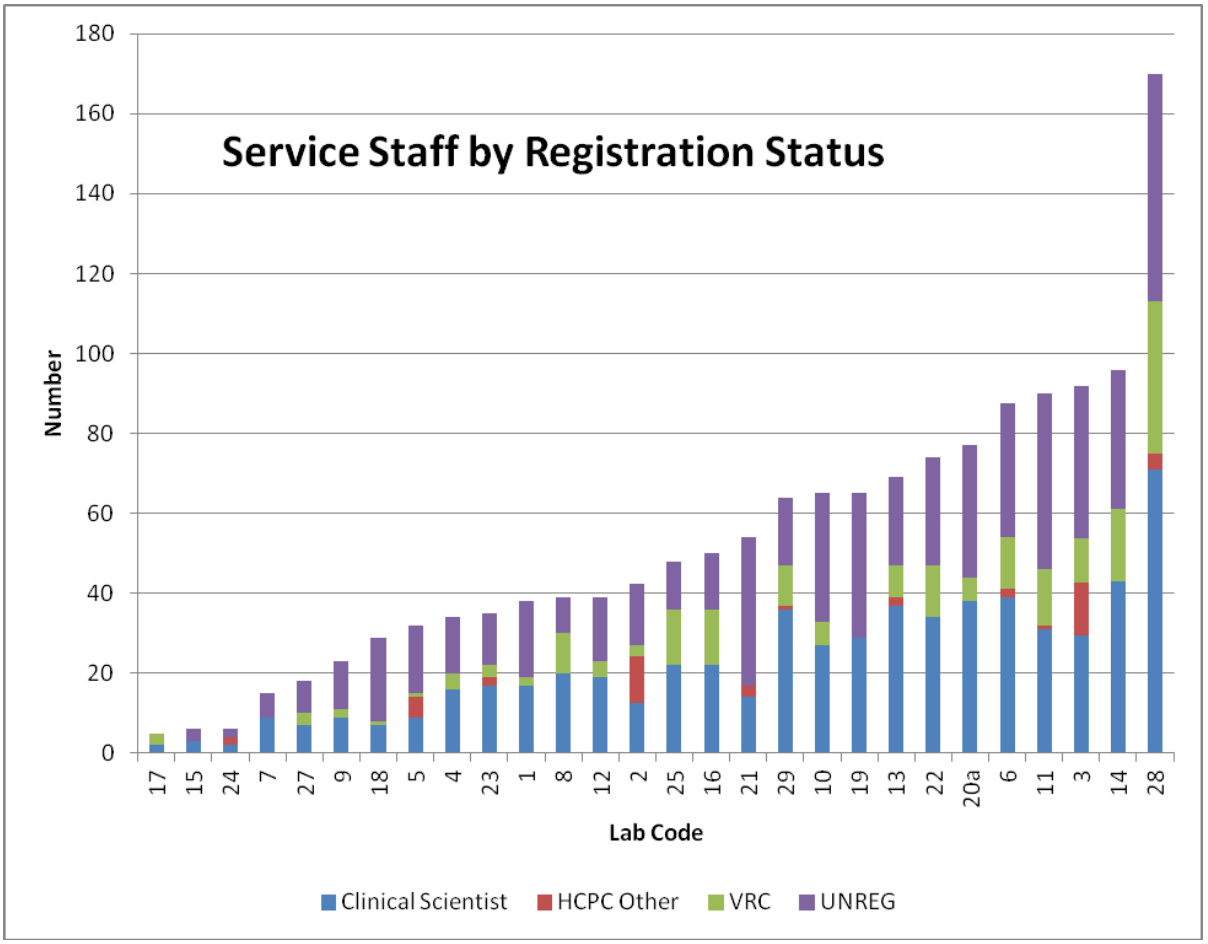
4.2 Summary table of average reporting times between labs for select tests over past 7 years
Top line in each section is for 2014-15

Reporting time	Day																%											
	3 working days				10 calendar days				14 calendar days				28 calendar days/				3 working days			10 calendar days			14 calendar days			28 calendar days		
Test	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
Prenatal Testing by karyotype analysis									10	15	13	12											53	100	85			
Prenatal Testing by karyotype **AND** targeted analysis	1	2	1	8					10	15	12	17					97	100	98				75	98	89			
Prenatal Testing by microarray analysis				1					6	12	9	4	22	22	22	1							91	100	97	100	100	100
Prenatal Testing by microarray **AND** targeted analysis	1	2	2	3					8	15	12	9	14	14	14	1	99	100	100				58	99	81	94	94	94
Prenatal testing by targeted analysis only (QF-PCR, FISH, MLPA)	1	2	2	13					2	20	10	3	21	21	21	1	92	100	97				33	100	75	100	100	100
Postnatal testing by karyotype analysis	2	2	2	1	6	9	7	17					16	93	28	20	100	100	100	84	100	94	93	93	93	30	100	74
Postnatal testing by karyotype **AND** targeted analysis	1	2	1	2	5	9	7	11					15	31	22	12	100	100	100	91	100	98				36	100	80
Postnatal testing by targeted analysis only (QF-PCR, FISH, MLPA)	1	3	2	6	1	10	7	10					11	45	23	17	72	100	92	67	100	90				25	100	80
Microarray analysis as first line test for Learning Disability Or Developmental Delay With Dysmorphism Or Congenital Malformations					8	14	10	4	12	15	13	3	14	109	33	18				42	91	77	59	82	70	8	100	62
Microarray analysis for Learning Disability Or Developmental Delay With Dysmorphism Or Congenital Malformations									16	16	16	1	24	29	27	3							55	55	55	75	99	88
Microarray analysis for Learning Disability Or Developmental Delay With Dysmorphism Or Congenital Malformations **AND** targeted analysis					2	8	6	3	13	13	13	1	9	83	30	8				85	100	93	100	100	100	63	100	81
Postnatal testing by microarray analysis (other)					8	8	8	1	16	16	16	1	19	45	28	6				100	100	100	56	56	56	29	96	74
Post Mortem testing for chromosome imbalance													15	32	23	18										47	100	81
Breakage studies for Blooms, Nijmegen, Ataxia telangiectasia & Fanconi anaemia by chromosome analysis					8	8	8	1					11	28	20	8				92	92	92				63	100	93

Reporting time	Day												%											
	3 working days				10 calendar days				14 calendar days				28 calendar days/				3 working days				10 calendar days				14 calendar days				28 calendar days		
Post Mortem testing for chromosome imbalance array only													20	20	20	1													86	86	86
Post Mortem testing for chromosome imbalance targeted only													8	8	8	1													99	99	99

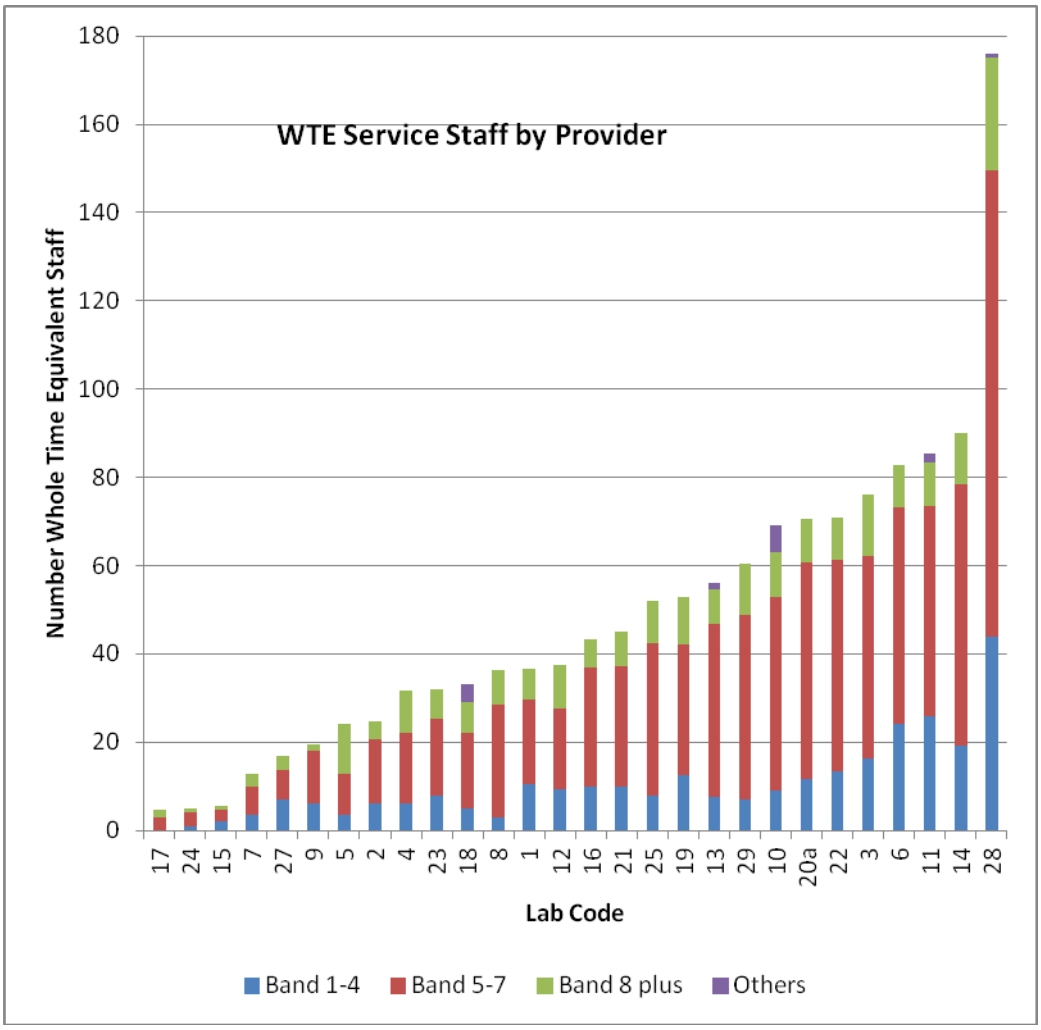
Table 4.4 below shows the turnaround performance by constitutional cytogenetic test category.

5 Service Staff



5.1 Number of laboratory service work staff registered with the Health & Care Professions Council (HCPC) or on the Voluntary Register by individual laboratory
Note: This is number not whole time equivalents

59% total laboratory staff were registered
The percentage of Clinical scientists of total staff was 43% as in last year's audit



5.2 Service staff as whole time equivalents by provider organisation

Key to codes on charts 2.2 to 2.10

Chart	Test (mutation, linkage, instability)	OMIM	Synonyms	Abb	Genes included (not exhaustive)
CF	Cystic Fibrosis including NBS	219700	CYSTIC FIBROSIS	CF	CFTR
BRCA	Familial breast & ovarian cancer (BRCA1&2)	113705, 600185	FAMILIAL BREAST & OVARIAN CANCER	BRCA	BRCA1, 2
FRAX	Fragile X syndrome (A & E)	309550, 309548	FRAGILE SITE MENTAL RETARDATION 1 GENE; FMR1	FRAX	FMR1, AFF2, FMR2
HFE	Haemachromatosis (all types)	235200	HEMOCHROMATOSIS, HEREDITARY; HH, HEMOCHROMATOSIS GENE; HFE, HLAH	HFE	BMPs, HFE
Clot	Clotting disorders (all types)	176930, 227400, 176930, 188055, 188050	COAGULATION FACTOR II; F2,	PTT, FVL, FV, F2	F2, FVL, PTT, F5, F2
FH	Familial hypercholesterolemia & Familial Defective ApoB100	143890	HYPERCHOLESTEROLEMIA, AUTOSOMAL DOMINANT & FDB	FH	LDLR, PCSK9, APOB
CRC	Colorectal cancer, Familial adenomatous polyposis, Mixed Polyposis, Lynch, hereditary mixed polyposis, juvenile polyposis, MAP	114500, 603054, 601228, 174900, 608456	COLON CANCER, FAMILIAL NONPOLYPOSIS, COLORECTAL CANCER; CRC, LYNCH CANCER FAMILY SYNDROME I, Muir Torre, POLYPOSIS, JUVENILE INTESTINAL, COLORECTAL ADENOMATOUS POLYPOSIS, AUTOSOMAL RECESSIVE MUTYH, E. COLI, HOMOLOG OF; MUTYH	HNPCC, MTS, GREM1, JPS, MAP	APC, MUTYH, APC, BMPR1A, CDH1, CHEK2, KIT, MLH1, MSH2, MSH6, MutYH, PALB2, PMS1, POLD1, POLE, SMAD4, STK11, GREM1
MT	Mitochondrial disorders	551500, 174763, 606075, 601465, 137960, 604712, 103220, 188250, 603921, 611224, 604983, 545000, 500001, 174763, 606075, 601465, 137960, 604712, 103220, 188250, 603921, 611224, 604983, #256000, 535000, *185620, 530000	NARP MERRF, LEIGH SYNDROME DUE TO MITOCHONDRIAL COMPLEX I DEFICIENCY, INCLUDED, LEBER HEREDITARY OPTIC NEUROPATHY; LHON,	MT	MT-TL1, MT-ATP6, MT-TK, AGK, C10orf2 (PEO1), DGUOK, DNA2, FBXL4, MFN2, MGME1, MPV17, OPA1, POLG, POLG2, RRM2B, SLC25A4 (ANT1), SPG7, SUCLA2, SUCLG1, TK2, TYMP, mitochondrial complex I, ND6, ND4, ND1, MTND3, MTND4, MTND1, MTND6, SURF1
Cardiac	Cardiac including Hypertrophic & Dilated cardiomyopathy	115200, 275210		HCM, DCM	SCN5A, TNNI3, MYBPC3, MYH7, TNNT2, LMNA, ZMPSTE24
Thal	Thalassaemia (all)		alpha thalassemia, beta thalassemia	Thal	HBA HBB
HbS	Sickle cell anaemia	603903	SICKLE CELL ANEMIA	HbS	HBB
CMT	Charcot Marie tooth related diseases, HMSN (all type), Focal	118220, 302800, 118200, 613237		CMT	PMP22, MPZ, GJB1, Cx32/GJB1, MFN2, INF2

Chart	Test (mutation, linkage, instability)	OMIM	Synonyms	Abb	Genes included (not exhaustive)
	segmental glomerulosclerosis, HMSN + HNPP dosage				
GJB2	Connexin 26/30 related deafness	121011	GAP JUNCTION PROTEIN, BETA-2	GJB2	Cx26, Cx30
Ataxia	Inherited ataxias (all) includes Friedreich ataxia	604326		SCA, MJD, FRDA	SCA1,2,3,6,7, 17, 12, AARS, ABCB7, ABHD12, ADCK3, AFG3L2, AMPD2, ANO10, AP1S2, APTX, ARSA, ATCAY, ATP1A3, ATP8A2, C10orf2, CACNA1A, CACNB4, CASK, CCDC88C, CHMP1A, CLN6, CLP1, COX20, CYP27A1, CYP2U1, DARS2, DDHD2, DNMT1, ELOVL5, EXOSC3, FGF14, FLVCR1, FOLR1, GBA2, GOSR2, GRID2, GRM1, HEXA, HEXB, ITPR1, KCNA1, KCNC3, KCND3, KCNJ10, KIAA0226, MRE11A, MTPAP, NPC1, NPC2, OPHN1, PCLO, PDYN, PEX16, PIK3R5, PLA2G6, PMPCA, PNKP, PNPLA6, POLG, PRKCG, PRRT2, RARS2, RNF170, SACS, SEPSECS, SETX, SIL1, SLC1A3, SLC2A1, SLC9A6, SNX14, SPG7, SPTBN2, SRD5A3, STUB1, SYNE1, SYT14, TDP1, TGM6, TMEM240, TPP1, TSEN2, TSEN34, TSEN54, TTBK2, TTC19, TUBB4A, UBR4, UCHL1, VAMP1, VLDLR, VPS53, VRK1, WDR81, WFS1, WWOX, ZFYVE26, ZNF592, FXN
Hbs	Haemaglobinopathies	141800, 141900	Abnormal haemaglobins (all types)	Hbs	HBA HBB
MTHFR	Homocystinuria due to deficiency of N(5,10)-methylenetetrahydrofolate reductase activity	236250, *607093	MTHFR		MHTFR
DM	Myotonic dystrophy (all types)	602668	DYSTROPHIA MYOTONICA 1	DM1, DM2, PROMM	DMPK
RP	Retinal dystrophy (all types)	#613731, #613767		RP	RDS/PERIPHERIN, RHO, CNGB1
HD	Huntington disease and Huntington-like	143100	HUNTINGTON DISEASE	HD	IT15
AS, PWS	Angelman & Prader Willi like syndromes	176270, 300243	Prader Willi, Angelman	AS, PWS	SNRPN, SLC9A6
Ydel	Y deletion	400005		Ydel	AZF1
LQT	Long QT (all types)	192500, 152427	LONG QT SYNDROME 1; LQT1, LQT2, LQT3	LQT	KCNQ1, KCNH2, SCN5A, KCNE1, KCNE2
LGMD	Limb girdle muscular dystrophies	#253600	LIMB-GIRDLE MUSCULAR DYSTROPHY	LGMD	CAPN3
NF1	Neurofibromatosis 1	162200		NF1	NF1

Chart	Test (mutation, linkage, instability)	OMIM	Synonyms	Abb	Genes included (not exhaustive)
MFS	Marfan syndrome	154700		MFS	Fibrillin
SMA	Spinal muscular atrophy (all types) including SMARD	253300, 301830		SMA, SMARD	SMN, UBA1,IGHMBP2
Rare anaemia	Rare & research confirmation				
A1555G	Mitochondrial deafness	561000	AMINOGLYCOSIDE ASSOCIATED DEAFNESS	A1555G	
Aorta	Aortopathies, Aortic aneurysm (all types), Supravalvular Aortic Stenosis (SVAS), AORTIC VALVE DISEASE	611788, 132900, 190198	Including TAAD, Loeys Dietz	TAAD	AAT4, AAT6, ACTA2, MYH11
EPD	Epilepsy (all types)	600513, 613720, 266100, 308350		EPD	CHRNA4, KCNQ2
MODY	Maturity onset diabetes of the young (all types)	142410, 138079		MODY	HNF1, HNF4A, GCK
CN	Gilbert syndrome,Crigler-Najjar syndrome	143500, 218800, 606785	Crigler Najjar	CN	UGT1A1
Cranio	Craniosynostosis related disorders	123500, 101200, 101600, 101400, 116860, 603284, 603285, 304110, 168500, 609597, 201750	CROUZON , Apert, Saethre Chotzen, Pfeiffer, Muenke	CCM, EFNB1, PF, POR	FGFR3, TWIST, FGFR2, FGFR1, KRIT1, CCM2, PDCD10, ALX4, MSX2, ABS1
MD	Muscular Dystrophy Duchenne, Becker	310200	MUSCULAR DYSTROPHY, DUCHENNE TYPE; DMD&BMD	DMD, BMD	Dystophin
Noonan-like	Noonan spectrum disorders	611533, 607721			BRAF, CBL, HRAS, KRAS, MAP2K2, NRAS, PTPN11, MAP2K1, RAF1, SHOC2, SOS1, SPRED1, RAF1, SOS1
MEN	MULTIPLE ENDOCRINE NEOPLASIA all types, including FAMILIAL MEDULLARY THYROID CARCINOMA	610755, 171400, 131100, 155240		MTC, MEN I, CDKN1B	CDKN1B, RET MEN1
FSHD	Facioscapulohumeral muscular dystrophy	158900	FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY 1A; FSHMD1A	FSHD	D4Z4
AD	Alzheimer, Dementia disorders	607822, 104760, #606889, *137350, #176500	AMYLOID BETA A4 PRECURSOR PROTEIN; APP		PSEN1, APP
Sotos	Sotos syndrome weaver	117550, 277590	Weaver		NSD1, EZH2
Ca	Cancer panel including TruSight & familial				APC, ATM, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDKN2A, CHEK2, KIT, KLLN, MLH1, MRE11A, MSH2, MSH6, MutYH, PALB2, PMS1, POLD1, POLE,

Chart	Test (mutation, linkage, instability)	OMIM	Synonyms	Abb	Genes included (not exhaustive)
					RAD50, RAD51C, RAD51D, SMAD4, STK11
PEO	POLG disorders	*174763	POLYMERASE, DNA, GAMMA-1	PEO	POLG
HP	Hereditary pancreatitis	27600, 167790		HP	SPINK, PRSS1
HSP	Hereditary spastic paraplegia	#610250, 182601	FAMILIAL SPASTIC PARAPLEGIA	HSP	SPG31(REEP1),ATL1, REEP1, NIPA1, SPG4
Phaeo	Phaeochromocytoma panel	171300	Neuroendocrine tumours panel, Pheochromocytoma		VHL, SDHB, SDHC, SDHD, SDHAF2, PRKAR1A, TMEM127, RET
HUS	Hemolytic-uremic syndrome	235400		HUS	CFH1/3, HF1
LFS	Li Fraumeni	151623		LFS	TP53
FTDALS	Frontotemporal dementia and/or amyotrophic lateral sclerosis	*614260 , 105550, 600274	FALS	FTDALS	C9orf72, Tau
Misc	Miscellaneous…				
PFIC	Cholestasis gene panel (including Alagille)	#211600,605814, 603471, 603859, 613404		PFIC	ABCB11, ABCB4, JAG1, NOTCH2,JAGGED, ATP8B1, NPC1, NPC2, SLC25A13
PKD	Polycystic kidney disease	173900, 263200, 26300, 606702		PKD, ARPKD	PKD1, FYCT, PKHD1
CAH	Congenital adrenal hyperplasia (all types)	#202010	ADRENAL HYPERPLASIA, CONGENITAL, DUE TO 21-HYDROXLASE DEFICIENCY	CAH	CYP21B, CYP11B1
TSD	Tuberous sclerosis	19100		TSD	TSC1, TSC2
Exome	Clinical Exome sequence for various panels		NGS panel		
NIPD	Fetal sex by NIPD			Sex	SRY
SRNS	Steroid resistant nephrotic syndrome NGS panel				
Skeletal	Skeletal dysplasia (all types)				FGFR3
EDS	Ehlers Danlos syndrome	130050		EDS	COL5A1
NF2	Neurofibromatosis 2	101000	BILATERAL ACOUSTIC NEUROFIBROMATOSIS	NF2	NF2
PGL	Familial paragangliomas	185470	SUCCINATE DEHYDROGENASE COMPLEX, SUBUNIT B, IRON SULFUR PROTEIN; SDHB	PGL	SDHB, SDHD SDHC
GSD	Glycogen storage diseases (all types)	#232600		GSD	PYGM, SLC37A4
CADASIL	Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy	125310		CADASIL	NOTCH3
BRRS,	PTEN disorders	153480, 158340, 601728	Bannayan-Riley-Ruvalcaba syndrome, Cowden syndrome	BRRS, BZS, RMSS,	PTEN

Chart	Test (mutation, linkage, instability)	OMIM	Synonyms	Abb	Genes included (not exhaustive)
BZS, RMSS, CD, MMAC1				CD, MMAC1	
EB	Epidermolysis bullosa multiple genes	131760, 131900, 131800, 179850	EBS Dowling Meara, Weber Cockayne, Koebner, Dowling-Degos	EBS	KRT5, KRT14
HNPP	Hereditary neuropathy with liability to pressure palsies	162500	TOMACULOUS NEUROPATHY	HNPP	PMP22
Rett	Rett syndrome	312750		Rett	MECP2
Alport	Alport syndrome (all types)	301050, 120131, 120070			COL4A3, COL4A5
PGD	Preimplantation work			PGD	
Rb	Retinoblastoma	180200		Rb	RB1
OI	Osteogenesis imperfecta (all types)	166200		OI	COL1A1, COL1A2
BWS	Beckwith Wiedemann syndrome	130650		BWS	NSD1,H19,KCNQ1,CDKN1C
SRS, RSS	Silver Russell	180860	Russell Silver syndrome	SRS, RSS	
ApoE	Apolipoprotein E	107741			APOE
AAT	Alpha 1 antitrypsin deficiency	107400	PROTEASE INHIBITOR 1	AAT	PI1
SMDP	Surfactant metabolism dysfunction	*178620, 265120, 610921, 267450		SMDP	HSPC, ABCA3, HSPB
DYT5, PD	Dystonia & Parkinsonism disorders	128230, #128235, #118800, #601042, #600116, 609007		DYT5, PD	GCH1, MR1, PARK2, LRRK2, PINK1
CMS	Congenital Myasthenic Syndromes (All types)	100690, 100710, 100720, 100725, 100730, 603033, 118490, 601592, 610285, 191350, 138292		CMS	CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, COLQ, CHAT, DOK7, DPAGT1, GFPT1, RAPSN,
Rare anaemia	Rare inherited anaemias, Diamond Black fan, THIAMINE RESPONSIVE MEGALOBLASTIC ANAEMIA	249270			RPS19, SLC19A2
Stickler	STICKLER SYNDROME, TYPE II; STL2	184840, #604841			COL11A2
FHH1	Hypocalciuric hypercalcaemia	145980, 145981, 600740		FHH1	CASR,GNA11, AP2S1
MCADD	Medium chain deficiency of acyl coA dehydrogenase deficiency	201450		MCADD	ACADM

Chart	Test (mutation, linkage, instability)	OMIM	Synonyms	Abb	Genes included (not exhaustive)
MTTL	RENAL CYSTS AND DIABETES SYNDROME, MATERNALLY INHERITED DIABETES AND DEAFNESS, Diabetes mellitus, TNDM	137920, 520000, 610199, 520000, 601410			HNF1B, MTTL1, ZFP57, PLAGL1
HHT	TELANGIECTASIA, HEREDITARY HEMORRHAGIC, OF RENDU, OSLER, AND WEBER; HHT	#187300, #600376	OSLER-RENDU-WEBER DISEASE		ENG, ACVRL1, SMAD4
VHL	Von Hippel Lindau	193300		VHL	VHL
DYT11	Torsion dystonia	128100			DYT1
GAL	Fabry disease	301500	ANDERSON-FABRY DISEASE, CERAMIDE TRIHEXOSIDASE DEFICIENCY, ALPHA-GALACTOSIDASE A DEFICIENCY, ANGIOKERATOMA, DIFFUSE, GALACTOSIDASE, ALPHA, INCLUDED; GLA, INCLUDED, GLA DEFICIENCY, HEREDITARY DYSTOPIC LIPIDOSIS	GAL	GLA
ID	Maternal cell contamination & identity testing		Identity testing	ID	
ARVC	Arrhythmogenic right ventricular cardiomyopathy	600996		ARVC	RYR2
11p15	11p15 associated conditions group	600856, 606528, 616186, 103280, 604115			H19, ICR1, KCNQ1OT1, CDKN1C
AJ scrn	Ashkenazi Jewish mutation panel				
SBMA	Spinal and bulbar muscular atrophy	313200	KENNEDY DISEASE	SBMA	AR
CM	Congenital myopathy	161800		CM	ACTA1, BIN1, CFL2, DNM2, KBTBD13, MTM1, MYH2, MYH3, MYH7, MYH8, NEB, RYR1, SEPN1, TNNI2, TNNT1, TNNT3, TPM2, TPM3, ORAI1, STIM1, STIM2
CMD	Congenital muscular dystrophy (all types)	#253800, 254090	FUKUYAMA, ULLRICH	CMD	FUKUTIN, UCMD
Kabuki	Kabuki syndrome (all types)	147920			MLL2
RCC	Renal cell carcinoma	605074		RCC	PRCC, MET
WD	Wilson disease	277900		WD	ATP7B
HLA B27	HLA B27 (ankylosing spondylitis)				MHC
Lymphoe	Milroy disease, lymphoedema	153100	Lymphedema,		FLT4

Chart	Test (mutation, linkage, instability)	OMIM	Synonyms	Abb	Genes included (not exhaustive)
OPA	Optic atrophy	165500			OPA1
EA	Episodic ataxia	108500		EA	CACNA1A
GLUT1DS1	Paroxysmal exercise-induced dyskinesia, SLC2A1	*138140	Paroxysmal exercise-induced dyskinesia, SLC2A1	GLUT1DS1	SLC2A1
MPS	Mucopolysaccharidoses (all types)	253000, 309900, 252900, #252920, #252930	MUCOPOLYSACCHARIDOSIS, Hunter, SANFILIPPO, MAROTEAUX-LAMY	MPS	GALNS, IDUA, SGSH, NAGLU, ARSB, HGSNAT
SQSTM1	PAGET DISEASE OF BONE 3; PDB3	#167250			SQSTM1
PDS	Pendred syndrome	274600		PDS	SLC26A4
PHOX2B	Phox2B disorders	209880	CCHS ONDINE-HIRSCHSPRUNG DISEASE, HADDAD SYNDROME, INCLUDED, ONDINE CURSE, CONGENITAL CENTRAL HYPOVENTILATION SYNDROME, CONGENITAL, OHD		PHOX2B
Cataract	Cataract all types	#116200			GJA8
AHO	Albright's hereditary osteodystrophy	#174800, 603233, *615549	MCCUNE-ALBRIGHT SYNDROME, PSEUDOHYPOPARATHYROIDISM, TYPE IB	AHO	GNAS, GNASAS1, STX16
HLRCC	Fumarase deficiency	#605839	FUMARATE HYDRATASE LRCC HEREDITARY LEIOMYOMATOSIS AND RENAL CELL CANCER; HLRCC	HLRCC	FH
RYR1	Malignant hyperthermia	180901	CENTRAL CORE DISEASE		RYR1
DRPLA	Dentatorubral pallidoluysian atrophy	125370		DRPLA	ATM1
Melanoma	Melanoma-pancreatic carcinoma syndrome	606719			CDKN2A, CDKN2A, CDK4
IP	Incontinentia pigmentosa	308300		IP	IKBKG
NCL	Batten disease (all types)	*611124, 204500, 256730	NEURONAL CEROID LIPOFUSCINOSIS		CLN7, CLN3, CLN2, CLN5, CLN6, CLN8, CLN1
Loeys-Dietz	Loeys-Dietz	607432			TGFBR1, TGFBR2
OPMD	Oculopharyngeal muscular dystrophy	164300		OPMD	PABPN1
DYT11	Myoclonic Dystonia	159900		DYT11	SGCE
JSRD	Joubert syndrome and related disorders	213300, 243910, 216360, 277170, 266900		JSRD	AHI1, ARL13B, B9D1, B9D2, C2CD3, C5orf42, CC2D2A, CEP290, CEP41, CSPP1, INPP5E, KIF7, MKS1, NPHP1, OFD1, PDE6D, POC1B, RPGRIP1L, TALPID3, TCTN1, TCTN2, TCTN3,

Chart	Test (mutation, linkage, instability)	OMIM	Synonyms	Abb	Genes included (not exhaustive)
					TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TTC21B
CDH1	Gastric cancer	137215			CDH1
RGS	Aaxenfeld Rieger	*120130 175780, 611773, 607595,#180500,175780, 611773, 607595	AXENFELD-RIEGER MALFORMATION, COL4A1 COLLAGEN OF BASEMENT MEMBRANE, ALPHA-1 CHAIN ARRESTEN, INCLUDED		COL4A1
LPL	Hyperlipoproteinaemia (type1,III,V)	#238600, 107741	Lipoprotein lipase deficiency, chylomicronaemia		LPL, LMF1, APOC2; APOA5, GPI-HBP1, APOE
FIPA	Familial isolated pituitary adenonma	102200	Pituitary adenoma predisposition	FIPA	AIP
HLA B57	HLA B57 (abacavir sensitivity)				MHC
TSD	Tay Sachs disease	272800		TSD	HEXA
BCNS	Basal cell nevus syndrome	#109400	GORLIN SYNDROME	BCNS	PTCH1, PTCH
ALS, MND	Amyotrophic lateral sclerosis	105400	Motor neurone disease	ALS, MND	SOD1, ALS6, NEFH
GLUD1	Hyperinsulinism	256450, 606762			ABCC8, GLUD1, GDH
BBS	Bardet Biedl syndrome	209900		BBS	BBS1, BBS2, BBS3, BBS4, BBS6 (MKS)
HP	Hyperoxaluria (all types)	613616, 259900 260000	HYPEROXALURIA, PRIMARY, TYPE III;	HP	HOGA1, AGXT, GRHPR
LWD	SHOX	127300, 300582, 249700	LERI-WEILL DYSCHONDROSTEOSIS; SHORT STATURE, IDIOPATHIC, X-LINKED; LANGER MESOMELIC DYSPLASIA	LWD	SHOX