



The European Molecular Genetics Quality Network

SUMMARY REPORT

2010 EQA SCHEMES

EMQN
Genetic Medicine, 6th Floor
St Mary's Hospital, Oxford Road,
Manchester M13 9WL,
United Kingdom
Tel: +44 161 276 6741
Fax: +44 161 276 6606
Web: www.emqn.org
Email: support@emqn.org

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SUMMARY REPORT

On behalf of EMQN, I would like to thank all the laboratories that participated in this years exercise. EQA is primarily an educational activity and our aim is to improve laboratory performance. We offered 28 EQA scheme in 2010 and the number of participations was broadly similar to the previous year. Our scheme assessment teams marked more than 4000 clinical reports and evidence from this process suggests that the overall standards of testing and reporting are gradually improving. The 2010 schemes are now complete and a summary of the final scores can be found in Appendix 2 and 3 of this document.

We look forward to your participation in 2011.

Yours sincerely,

Dr Simon Patton
EMQN Executive Administrator

2010 EQA PARTICIPATION

EMQN received 1523 registrations for the disease specific EQA schemes in 2010 and 199 registrations for the technique specific scheme from 422 laboratories world wide (Table 1). Although the participation levels were slightly down (4%) compared to 2009, partly due to the withdrawal of mutation scanning EQA scheme from our catalogue, some schemes had more participants than previously. A new scheme for familial adenomatous polyposis was offered in collaboration with Human Genetics Society of Australasia. The participating countries also increased with two new countries becoming members (Iceland and Russian Federation). The bar chart in figure 1 shows the countries with largest number of participants (more than 5 laboratories); the map displays all the countries which participated in 2010 schemes.

Table 1: 2010 EQA scheme participations

Scheme	2010	Difference to 2009
AZF	142	+9
BRCA	109	+2
CAH	46	+5
CMT	73	+11
DFNB1	57	-14
DM	60	-1
DMD	62	-12
FAP	35	-
FRAX	131	+15
FRDA	57	+13
HD	96	+6
HFE	82	+7
HNPCC	91	-18
HRF	36	-14
MEN2	60	+5
MonoDiab	29	0
MSCAN	-	-38
PKU	20	0
POR	19	-1
PWAS	90	-12
RB	14	0
SCA	64	+3
SMA	76	-2
SEQ	199	-23
VHL	41	+2
WIL	33	+6
TOTAL	1722	-63

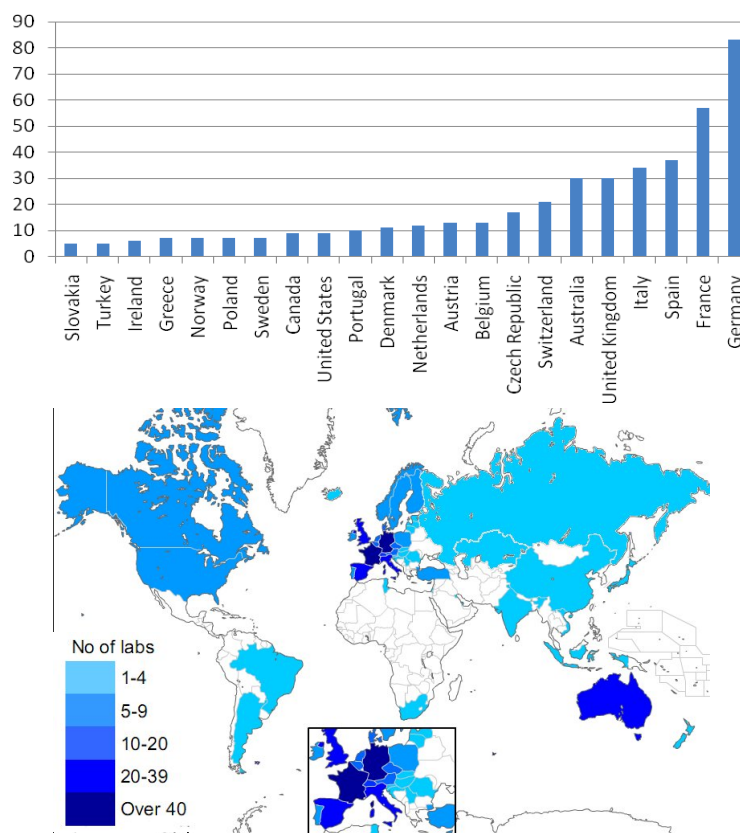


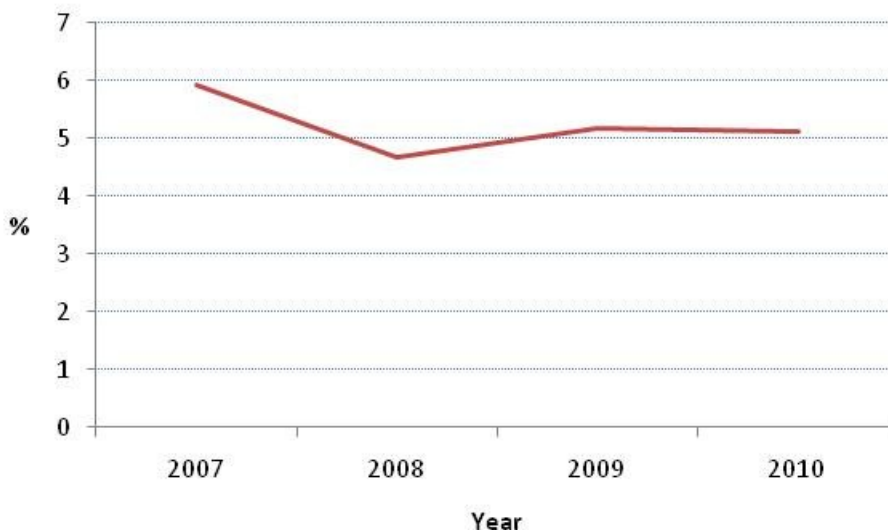
Figure 1: 2010 EQA scheme participation by country

POOR PERFORMANCE

Poor performance criteria address genotyping only and are defined as “An overall mean genotyping score of less than 1.60 for a disease”. (e.g., a single serious genotyping error will classify a lab as a poor performer). This year there were 84 (5.13%) poor performers from 1639 assessed EQA scheme participations (see Appendix 1). The number of poor performers appears to be reaching a steady state of around 5% per annum (see Figure 2 below). If the exceptional results from the HNPCC scheme (discussed below) were excluded, there would have been a marked improvement to 4.1% in 2010.

All poor performers will be contacted by the EMQN Administrator. Please work diligently to find out the source of the error, as it may indicate a flaw in your assay or quality system that requires immediate attention. EMQN scheme organisers are available to offer confidential advice if required.

Figure 2: % poor performers (2007, 2008, 2009, 2010)



SHARED LEARNING FROM EQA

- We are still concerned by the apparent resistance of some participating laboratories to learn from previous EMQN scheme participations. Our dedicated and hardworking assessment teams have reviewed approximately 4000 EQA scheme reports this year and are still encountering laboratories which make the same errors/mistakes each year. We strongly recommend that these labs take our comments seriously, as they are made in the light of extensive experience and are aimed at making sure that patient receives the best possible care. The EMQN Board is considering additional measures to encourage such labs to improve their reports.
- A small number of schemes reported that laboratories were not including a statement of assay sensitivity / specificity. This is especially important when reporting a negative result and should therefore be on the report. This has been mentioned in the 2010 scheme reports concerned.
- The HNPCC scheme reported 23 genotyping errors as many labs failed to detect a variant (c.994dupA) under one of the MLPA probe binding sites. This resulted in these labs stating that a single exon deletion was present. Whilst this was a difficult case, it does however emphasise the importance of confirming all MLPA test single exon deletions with another technique.
- Regular EQA participation does improve reporting. The scheme assessment teams reported anecdotal evidence of a noticeable improvement in reporting from regular EQA participating labs compared to those who are new to EQA.

Some general points for improving the quality of reports are as follows:

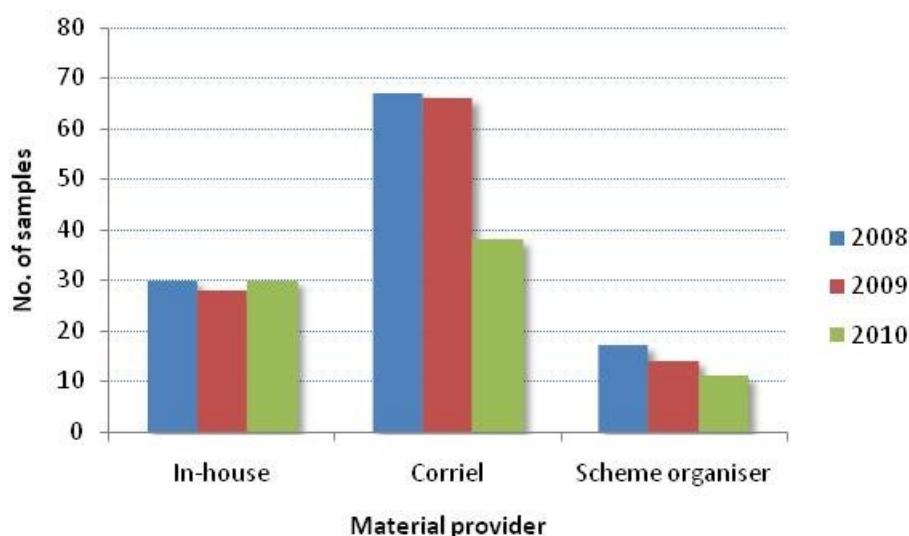
- (1) In order to prevent important information getting lost or separated, reports should be restricted to 1 page only where possible. This year we had quite a few reports of up to 4 pages, virtually constituting clinical reviews of the syndrome in question.
- (2) It is good practice to restate the reason for referral, the gender of the patient and the sample type analysed. Also include the date of referral, date of report and laboratory ID for the sample.
- (3) Any kit number (and its version number) should be stated on reports.

- (4) Presentation of the central results should be highlighted in bold or in a table to improve the clarity of reports.
- (5) The terms 'negative' and 'positive' should not be used when reporting clinical results as they can be misleading.
- (6) Reference sequences (including version number) should be given on the report.

EQA MATERIALS

As in previous years, most of the DNA used for our EQA schemes came from cell lines (see Figure 3). In 2010 half of the DNA was provided by a large commercial provider with the rest of the samples being prepared mainly in Manchester. The DNA was prepared using two different methodologies: salting out (commercial provider) and phenol/chloroform extraction (Manchester). We are pleased to say that we had very few problems this year with the scheme samples and in Appendix 4 you can see a table showing the data on repeat sample request rates, which, we are pleased to say, declined again in 2010. There was no difference in the success rate of DNA prepared using either methodology stated above.

Figure 3: EQA Materials provision (2008 – 2010)



EQA SCHEMES FOR 2011

Registration for the 2011 EQA schemes is OPEN via the website. The closing date for registrations is 22.07.11. Full details of the schemes can be found in the 2011 scheme catalogue (see <http://www.emqn.org/emqn/Schemes> for details). However, for your information, we intend to offer EQA for the following:

- Breast cancer (Familial - BRCA)
- Charcot-Marie-Tooth disease (CMT)
- Congenital Adrenal Hyperplasia (CAH)
- DNA sequencing (DNA-SEQ)
- Duchenne and Becker Muscular Dystrophy (DMD / BMD)
- Hereditary recurrent fevers (HRF)
- Familial Adenomatous Colon Cancer (FAP)
- Fragile-X syndrome (FRAX)
- Friedreich ataxia (FRDA)
- Hereditary Deafness (DFNB1)
- Hereditary Haemochromatosis (HFE)
- Hereditary non-polyposis colon cancer (HNPCC)
- Huntington disease (HD)
- Monogenic diabetes (MonoDiab)

- Multiple Endocrine Neoplasia type 2 (MEN2)
- Myotonic dystrophy (DM)
- Phenylketonuria (PKU)
- Porphyrias (POR)
- Prader-Willi and Angelman syndromes (PW/AS)
- Retinoblastoma (RB)
- Spinal Muscular Atrophy (SMA)
- Spinocerebellar ataxia's (SCA)
- Von Hippel Lindau Syndrome (VHL)
- Wilson disease (WIL)
- Y-Chromosome microdeletions (in conjunction with the EAA - AZF)
- Constitutional Molecular Karyotyping (Microarray / Array CGH - in conjunction with CEQA¹)

NEW PILOT EQA SCHEMES FOR 2011

- Short stature homeobox gene testing (SHOX)
- EGFR mutation testing in NSCLC ((in conjunction with the ESP², ETOP³ and ESMO⁴)

There will be no fee for participation for the pilot schemes if you register during period 1 (see EQA scheme catalogue for details: <http://www.emqn.org/emqn/Schemes>).

2011 TIMETABLE

The timetable ⁵ for the 2011 scheme is as follows:

Activity	Start	Finish
EQA scheme registration	01.05.11	22.07.11
Sample distribution	October 11	October 10
Reporting period ⁶	01.10.11	16.01.12
Genotyping results published	Reporting date + 7 days	
Scheme assessments	20.02.12	21.02.12
Lab results published	01.04.12	01.04.12
Appeals process	01.04.12	14.04.12
Final scheme report published	30.04.12	30.04.12

FEES

The fees for our EQA schemes will remain the same as 2010. The registration fees are €275 (242 GBP) for Period 1 and €300 (264 GBP) Euro per scheme for Period 2. To encourage participation by laboratories in developing countries we are still offering a reduced fees scheme. Full details of the fee structure are available from the EMQN website (<http://www.emqn.org/emqn/Schemes>).

PARTICIPANT FEEDBACK

******* €50 AMAZON VOUCHERS TO BE WON!! *******

This year we are actively looking for feedback from our participants on their opinions of EMQN and its activities. You will receive a separate email from us inviting you to participate in the survey – we would be grateful if you could complete it (approx 10 mins) The closing date for completion of the feedback form is the 31st May 2011. The first 10 forms drawn at random (after the closing date) from all the submissions will win a €50 (45 GBP) Amazon voucher.

CERTIFICATES OF PARTICIPATION

If you registered to receive a certificate of participation, it will be available from the end of third week in May by downloading a copy from the website (see 2010 CERT_PART scheme page).

¹EMQN is also continuing collaboration with Cytogenetics European Quality Assessment (CEQA) (www.ceqa.org) to offer a scheme for constitutional molecular karyotyping using microarray / array CGH analysis. **This scheme runs on a different timeline to other EMQN's schemes and the registration is currently open until 30.06.2011.**

² European Society of Pathology

³ European Thoracic Oncology Platform

⁴ European Society of Medical Oncology

⁵ The dates shown above are provisional – please check the EMQN website for the most up to date information

⁶ There are three different reporting dates depending on the complexity of the scheme. Check the website for information.

APPENDIX 1 – Poor Performance Scores 2010

Scheme	No. Poor Performers (PP)	No assessed	% PP
AZF	2	140	1.43%
BRCA - FULL	4	69	5.80%
BRCA-GENO	2	39	5.13%
CAH	5	44	11.36%
CMT	1	72	1.39%
DFNB1	7	57	12.28%
DM	1	59	1.69%
DMD	1	61	1.64%
FAP	Pilot scheme - criteria do not apply		
FRAX (FULL)	4	83	4.82%
FRAX (PCR)	0	41	0.00%
FRDA	1	53	1.89%
HD	3	95	3.16%
HFE	4	80	5.00%
HNPCC	23	89	25.84%
HRF	2	34	5.88%
MEN2	2	58	3.45%
MODY	2	28	7.14%
PKU	4	20	20.00%
POR	1	17	5.88%
PW/AS	2	87	2.30%
RB	0	13	0.00%
SCA	5	62	8.06%
SEQ-Full	2	161	1.24%
SMA	1	73	1.37%
VHL	2	39	5.13%
WIL	3	32	9.38%
Total	84	1639	5.13%
Mean per scheme	3	62	6.05%

APPENDIX 2 – Mean Genotyping Scores 2010

Scores meeting poor performances criteria are highlighted in pink and red. Note: these criteria do not apply to pilot schemes (FAP)

Lab No.	AZF	BRCA - FULL	BRCA-GENO	CAH	CMT	DFNBI	DM	DMD	FAP	FRAX (FULL)	FRAX (PCR)	FRDA	HD	HFE	HNPCC	HRF	MEN2	MODY	PKU	POR	PW/AS	RB	SCA	SEQ-Full	SMA	VHL	WIL
0001		2.00													2.00									2.00			
0002							2.00																				
0003	2.00													2.00								2.00					
0004					2.00																				2.00		
0006												2.00		2.00										2.00	2.00		
0007		2.00																									
0008	2.00								1.67																2.00		
0012			2.00												1.33										2.00		
0013		2.00																									
0014	2.00				2.00			2.00			2.00		2.00	2.00										2.00	1.50		1.58
0015	2.00	2.00								1.83															2.00		
0018	2.00													2.00											2.00		
0019							2.00			2.00			2.00											2.00		2.00	
0021																							2.00				
0023			1.67														2.00										
0024				1.83		1.83		2.00		1.83						2.00	2.00					2.00		2.00	1.88		1.33
0026								1.67		1.83														1.88	2.00		
0027	2.00				2.00	1.75				1.92						1.92	1.83					2.00	2.00		2.00	2.00	
0030	2.00	1.83			2.00		2.00	2.00		2.00			2.00	2.00	2.00		2.00					2.00		1.33	2.00		
0031													2.00	2.00												2.00	
0032				2.00			2.00			2.00										2.00						2.00	1.83
0033																				2.00	2.00						
0034																									2.00		
0035			1.92																						2.00		
0036			2.00																								
0037															2.00		2.00								2.00		
0038																				2.00					2.00		
0039		2.00		1.83						1.83															2.00		
0042					2.00							2.00	2.00	2.00										2.00	2.00		2.00
0043														2.00													
0044					2.00							2.00		2.00										2.00			
0046					2.00		2.00					2.00	2.00											2.00			
0047			1.92												2.00		1.92										
0049														2.00											2.00		
0050					2.00																						

0052	2.00		2.00			2.00	2.00		2.00				2.00	2.00		2.00				2.00			2.00				
0053					2.00		2.00																				
0054	2.00					2.00	2.00		2.00				2.00													2.00	
0055												1.92														2.00	
0056	2.00					2.00			2.00	2.00	2.00	2.00								2.00			2.00			2.00	
0059	2.00			2.00	1.83	2.00			2.00		2.00	2.00			1.94					2.00			1.83			2.00	
0060													2.00														
0061									1.83					1.33						2.00				2.00		2.00	
0062	2.00																										
0065	1.67				1.83	2.00	2.00		2.00	2.00	2.00									2.00			1.83			2.00	
0068								1.83		2.00																	
0069	2.00													2.00		2.00		2.00								2.00	
0071												2.00															
0073	2.00										2.00		2.00													2.00	
0075	2.00	2.00		2.00									2.00				2.00	1.83			2.00					0.67	
0076	2.00									2.00													2.00	2.00			
0077																						2.00					
0078	2.00								2.00																		
0080	2.00				1.67		2.00																			2.00	
0081		2.00																									
0082				2.00													2.00								2.00	2.00	
0083	2.00			2.00		2.00	2.00			2.00	2.00	2.00	2.00									2.00	2.00	2.00			
0084		2.00								2.00				2.00									2.00				
0085	2.00						2.00			2.00													2.00				
0090				2.00	1.83							2.00	2.00		2.00							1.33			2.00		
0091									2.00																		
0094	2.00				1.17				2.00													2.00			2.00	2.00	1.83
0099				1.00	2.00		2.00			2.00																1.92	2.00
0100	2.00	2.00		1.67						1.75	2.00	1.83	2.00	2.00		2.00	1.33	2.00	2.00					2.00		2.00	2.00
0101																2.00	1.88									2.00	
0102		2.00												1.33													
0103	2.00								2.00																	2.00	
0104												1.92					2.00									1.75	2.00
0105																										2.00	
0106														2.00												2.00	
0107				2.00																							
0109	2.00								1.83			2.00													2.00	1.33	
0111										2.00		2.00															
0112																										2.00	
0114																										2.00	
0117				2.00																						2.00	
0118									2.00																	2.00	
0119													2.00	1.33													
0120		2.00												2.00													
0121				2.00																						2.00	2.00

0122				2.00							2.00	1.83								2.00				
0123																					2.00			
0124	2.00	2.00		2.00	2.00	1.83	2.00	2.00		1.83		2.00	2.00	2.00	2.00	1.75	2.00			2.00	1.50	2.00	2.00	
0125	2.00												2.00									2.00		
0126												2.00												
0127	2.00																							
0128			2.00					2.00	2.00	1.83			2.00							2.00				
0129	2.00				2.00	1.25				2.00			2.00	2.00						2.00				
0130	2.00									2.00										2.00			2.00	
0132											1.83									2.00		2.00		
0133	2.00			1.83						2.00														
0134						1.92				1.92										2.00			2.00	
0135			1.87											2.00		2.00							2.00	
0137			2.00																					
0138							2.00	2.00		2.00												2.00	2.00	
0139	2.00	2.00		2.00					2.00					2.00	2.00					2.00		2.00		
0143		2.00													2.00									
0145	2.00	1.83			2.00	1.83	2.00																2.00	
0146																							2.00	
0148	2.00																							
0150																								
0151				1.50			2.00				2.00									2.00	1.83	1.33		
0152		2.00												2.00	2.00		1.67						2.00	
0153										2.00								2.00		2.00		2.00	2.00	
0154	2.00						2.00			2.00		2.00								2.00	1.67		2.00	
0155	2.00				2.00		2.00	2.00		1.33		2.00	1.83	2.00	0.00					2.00		2.00	2.00	
0160									1.83						2.00		1.92						2.00	
0161							2.00			2.00										2.00			2.00	
0162	2.00	2.00					2.00	2.00		2.00			2.00	2.00	2.00					2.00		2.00	2.00	
0163							2.00					2.00	2.00	2.00						2.00			2.00	
0164	2.00													2.00									2.00	
0168																							2.00	
0169	2.00	1.60		1.83	2.00	2.00		2.00			2.00	2.00	2.00	2.00	2.00	2.00	2.00	2.00	2.00			1.88	1.92	2.00
0171						1.83	2.00		2.00			1.83		2.00		2.00								
0172																					2.00		2.00	
0174	2.00							2.00	1.83					2.00									2.00	
0175	2.00	2.00		1.83	2.00		2.00	2.00	1.83	2.00		2.00	2.00	2.00	2.00	2.00	2.00	2.00	2.00			2.00	2.00	
0177	2.00			1.50						1.83		2.00				1.94	2.00	2.00	1.83	2.00	2.00		2.00	2.00
0178		2.00												2.00									2.00	
0179																							1.75	
0180			2.00			1.83				1.83		2.00		1.33		2.00				2.00	1.83	2.00	2.00	
0181	2.00																							
0182		2.00																						
0183																							2.00	
0184	2.00				2.00																		1.92	

0564	2.00		1.75				2.00					2.00		2.00		1.67		1.92		2.00					
0565												1.83													
0566	2.00	2.00		1.83		1.83		2.00	1.67				2.00	2.00		2.00								2.00	
0567	2.00		1.92		2.00	1.67	2.00	2.00		2.00			2.00	2.00							2.00			2.00	1.83
0568			1.33	1.83				2.00					1.92		1.33										
0569		2.00												2.00											
0570	2.00					1.67				1.75													2.00		
0571											1.83					2.00			1.83						1.92
0572		1.87		2.00	2.00	1.58	2.00					2.00	1.83	1.33							1.92	2.00	1.88	2.00	
0573																						2.00			
0574	2.00	1.00								2.00															
0575																							2.00		
0577				1.67	2.00								1.83									1.50			
0578													2.00												
0579					2.00						2.00		1.67										2.00		
0582			2.00																						
0584	2.00																								
0585																							2.00		
0588																1.94									
0589													2.00												
0590	2.00																								
0592			1.72																						
0593																						2.00			
0594																2.00									
0595	2.00												2.00										2.00		
0600													2.00												
0601														2.00											
0602			2.00						2.00						1.33										
0603																							2.00		
0607				1.83	2.00		2.00							1.58											
0608										1.83	1.33	2.00											2.00		
0609			2.00																						
0610					2.00																		2.00		
0618		0.00		0.00												2.00									1.92
0629																							1.88		
0631	2.00														1.25										
0632	2.00				2.00	1.83		2.00		2.00		2.00	1.83									1.42	2.00	2.00	
0633																							2.00		
0635				2.00																				2.00	
0636																							2.00		
0637																									
0638																2.00							2.00		
0641													2.00												
0642			1.60											2.00									1.25		
0643															1.92										

APPENDIX 3 – Mean Interpretation Scores 2010

Interpretation is not marked for pilot schemes (FAP). Note: Poor performances criteria are not applied to interpretation scores.

Lab ID	AZF	BRCA - FULL	CAH	CMT	DFNBI	DM	DMD	FRAX (FULL)	FRAX (PCR)	FRDA	HD	HFE	HNPCC	HRF	MEN2	MonoDiab	PKU	POR	PW/AS	RB	SCA	SEQ-FULL	SMA	VHL	WIL
0001		2.00											1.42									1.75			
0004				2.00																		1.92			
0006										1.92		1.83										2.00	2.00		
0007		2.00																				2.00			
0008	2.00																								
0012													2.00										1.84		
0013		2.00																							
0014	1.50			1.83			2.00		1.83		1.75	1.50										2.00	1.50		1.75
0015	2.00	2.00						1.67														2.00			
0018	2.00											1.92										1.84			
0019						2.00		2.00			2.00											2.00		2.00	
0021																					2.00				
0023															2.00										
0024			2.00		2.00		1.92	2.00						1.92	2.00				2.00			1.75	1.62		2.00
0026							1.25		2.00													1.00	1.75		
0027	1.83			2.00	2.00			1.67						1.92	2.00				2.00	2.00			2.00	2.00	
0030	1.67	2.00		2.00		1.92	2.00	2.00			1.75	1.58	2.00		2.00				1.92			1.50	1.62		
0031											2.00	1.08												2.00	
0032			1.67			2.00		2.00										1.42					1.75		1.75
0033																		1.75	2.00						
0034																							1.92		
0035																							1.84		
0037													2.00		2.00								2.00		
0038																		1.92					1.59		
0039		1.87	2.00					2.00															2.00		
0042				2.00						1.83	2.00	2.00										2.00	2.00		2.00
0043												1.75													
0044				2.00						1.75		2.00										1.83			
0046				2.00		2.00				2.00	2.00											2.00			
0047													1.67		1.67										
0049												1.67											2.00		
0050				2.00																					
0052	2.00					2.00	1.83	1.83				2.00	1.58		1.83				2.00			1.84			

0121																			2.00		1.92			
0122				1.83						1.92	2.00									1.50				
0123																						1.67		
0124	1.67	2.00	1.83	2.00	2.00	1.75	1.67	1.50		1.58	1.83	1.42	2.00	2.00	2.00				1.42		1.88		1.88	2.00
0125	2.00											1.58										1.84		
0126											2.00													
0127	1.83																							
0128							2.00	2.00			2.00								2.00					
0129	2.00			2.00	2.00			1.83			2.00	1.58							2.00					
0130	1.83							2.00					2.00						2.00					2.00
0132									1.83										2.00			1.83		
0133	2.00		1.83					2.00																
0134					1.83			2.00											2.00				1.75	
0135													1.67		2.00									1.67
0138						1.92	1.92	2.00														2.00	1.75	
0139	2.00	2.00	1.83									2.00	2.00						2.00			2.00		
0143		2.00											2.00											
0145	2.00	2.00		2.00	1.83	2.00																1.84		
0146																						1.59		
0148	2.00																							
0150																								
0151			1.00			1.92			1.83										1.92		1.67	2.00		
0152		2.00										1.83	2.00		1.50									2.00
0153								2.00									1.92	2.00			1.84			1.83
0154	2.00					2.00		2.00			2.00							2.00			1.83		1.75	
0155	1.67			2.00		2.00	2.00	2.00		1.92	1.67	1.67									1.67		2.00	2.00
0160													1.58		1.50									1.67
0161						1.83		2.00											2.00			1.62		
0162	2.00	2.00				2.00	1.92	2.00			1.83	1.83	2.00					2.00			2.00	2.00		
0163						2.00				2.00	2.00		2.00					2.00				2.00		
0164	1.67											1.42										2.00		
0168																						1.84		
0169	2.00	2.00	1.83	2.00	2.00		1.92		2.00		2.00	2.00	2.00	2.00	2.00	2.00	1.83	2.00				1.62	2.00	1.83
0171					2.00	2.00					2.00		1.62		1.83									
0172																			2.00			1.67		
0174	2.00						1.92					1.92											1.75	
0175	2.00	2.00	2.00	2.00		1.92	1.83	2.00			2.00	1.92	2.00	1.88	2.00	1.92	1.92		2.00			1.75	1.88	
0177	1.67		1.83					1.58			1.83			1.88	1.83	1.42	1.33	1.83	1.58				2.00	1.83
0178		2.00											2.00									2.00		
0179																								1.92
0180					2.00			1.75		1.92		0.75			2.00				2.00			1.67	1.84	1.38
0181	2.00																							
0182		2.00																						

0248															0.83	1.83					1.84				
0249	2.00																								
0251		2.00				2.00		1.75			1.67			1.88					2.00			2.00			
0252												2.00													
0253				2.00																			1.75		
0254					2.00	2.00	1.83	2.00			1.83			2.00					2.00			1.67	2.00	1.75	1.83
0257															2.00							2.00		2.00	
0258				2.00	1.83						1.67								2.00						
0259	1.83																				2.00		2.00		
0260						1.83																	1.88		
0261	2.00																					2.00			
0262					2.00								2.00											1.83	
0264														1.92											
0266	2.00																								
0268							1.50																		
0269		2.00			1.83	2.00	2.00				2.00											1.84	2.00		
0270				2.00		1.83					1.58	2.00									1.00	2.00	1.62		
0271	1.50																								
0273								2.00													2.00	1.67			
0274			2.00										1.92	2.00											
0275	2.00																								
0276	2.00	2.00			2.00	2.00	1.92	2.00			2.00	2.00		2.00	2.00	2.00			2.00		1.67		1.88		
0277		2.00											1.25								1.92				
0278																					1.92		1.92		
0279																							2.00		
0280																							1.84		
0281	1.33																								
0284				2.00			2.00	1.83					2.00										1.92		
0288	2.00																						1.88		
0289				2.00										2.00											
0290											1.92											2.00	2.00		
0296	2.00												2.00												
0297														1.67											
0299															1.67										
0301	2.00																						1.92		
0302	2.00								1.67												1.83				
0303																								2.00	
0307		2.00												2.00										0.92	
0311				2.00		1.92					2.00													1.83	
0313																								2.00	
0316							2.00	2.00			2.00											1.67	1.84	1.75	
0317			2.00	2.00						1.92	2.00				2.00							1.67			
0318	1.83																						0.92		

0395		2.00					2.00	2.00												2.00	1.75			
0398				2.00						1.75	2.00									1.92				
0399						2.00				1.92								2.00						
0400														1.67										
0404																					1.84			
0406	2.00		2.00	2.00		1.92	2.00				1.00		0.50		1.25	1.75		1.88	2.00		2.00		0.83	1.67
0408			1.83													2.00								
0413																								
0415							2.00																2.00	
0416	1.50		2.00			2.00		2.00					2.00						2.00					
0418		1.33																				1.83		
0421		2.00																						
0422																						1.92		
0423	1.83																							
0425										1.75										2.00	2.00	2.00		
0426		2.00																						
0428								1.33																
0432										1.42	2.00								2.00		1.67			
0435												1.75												
0436	1.67																							
0438																						2.00		
0439		2.00			2.00														2.00					
0440				1.83							2.00										2.00	2.00		
0441	2.00				2.00		1.92														1.92	1.88		
0445		2.00																						
0446								2.00					1.94	2.00										
0448											1.67											1.83		
0451	1.33																					1.75	2.00	
0452				2.00						1.92											2.00			
0455		1.00											1.33											1.83
0456		1.67																						
0457															2.00									
0458		1.67											2.00	1.83										
0459		2.00											2.00											
0460				2.00	1.67																		2.00	1.67
0461							1.67																1.75	
0462				2.00		1.92				1.67										2.00				
0463							2.00															2.00		
0464	2.00				2.00					2.00	1.50	2.00	1.42						1.67					
0466																			2.00					
0467		2.00		2.00				1.50		2.00	1.92		1.67						2.00			1.75		
0469	2.00						2.00	2.00															1.88	
0474	1.00	2.00	1.83	2.00	2.00		1.92	2.00			1.83	2.00					2.00		2.00				1.75	

0476	2.00						2.00						2.00					2.00			1.92	1.50		
0478																					1.67			
0480	2.00	2.00	2.00	2.00	2.00			2.00		1.83	2.00	2.00	2.00	2.00	1.75		2.00						2.00	2.00
0481					1.92																			
0484									2.00	2.00							2.00			2.00		1.75		
0485					1.83					1.83														
0486	1.67								1.67															
0487	1.83			2.00	1.75		1.75	1.75			1.75						1.67				2.00			
0489							1.83	1.83		1.58	2.00									2.00				
0492	1.83																							
0495																		1.58						
0496			1.83											2.00										
0497								1.83																
0500					2.00																	2.00	2.00	
0502																						1.50		
0503	1.67								1.92														1.75	
0505						2.00																		
0506	1.67																							
0508				2.00					1.92	2.00					1.92					2.00				
0509	2.00						2.00																1.88	
0510	2.00			2.00					1.75	2.00								1.83						
0511		2.00											1.88											
0513																						1.75		
0514	2.00	1.67										2.00												
0518				2.00	2.00		1.83		1.58	2.00	2.00							2.00		2.00				
0520	2.00				1.50																	1.67		
0521	1.75		1.50										1.25			1.67								
0523										2.00														
0524						1.33		2.00										2.00			2.00	1.62		
0525												1.92												
0528														2.00										
0529				2.00														2.00				1.84		
0531			1.67												1.50									
0533	1.83												1.88											
0535													1.88											
0539															1.42								2.00	
0540	1.33	2.00	2.00										2.00								2.00	2.00		
0542	1.83						1.50																	
0543										2.00														
0546		2.00											2.00											
0547										1.83														
0548	1.50																							
0549										2.00														1.83

0552				2.00				2.00					1.83									1.67
0553																						1.83
0554		1.67				2.00			2.00	1.67	1.67		1.38						2.00		1.62	
0555											1.58	1.75										
0560	1.83		2.00				2.00															
0561	2.00								1.75													1.75
0563		1.67								2.00	2.00	2.00							2.00			
0564	0.83						1.92				2.00		2.00		2.00		0.83		2.00			
0565											2.00											
0566	1.83	2.00	2.00		2.00		1.92					1.58	2.00		2.00							2.00
0567	1.83			2.00	2.00	2.00	1.92	2.00				1.83	1.67						1.92		1.88	1.92
0568			2.00				1.92					1.83		2.00								
0569		2.00												1.92								
0570	1.67				2.00			1.83											2.00			
0571									2.00					1.83				2.00				1.58
0572		1.67	2.00	1.83	1.50	1.92				1.42	1.67	0.75							1.67	2.00	1.50	1.67
0573																				2.00		
0574	2.00								2.00													
0575																					2.00	
0577			2.00	2.00							1.83								1.75			
0578											2.00											
0579				1.83					2.00		1.67										1.92	
0584	1.17																					
0585																					1.84	
0588													1.94									
0589												1.92										
0590	2.00																					
0593																			2.00			
0594																1.67						
0595	2.00											1.25									1.75	
0600												1.33										
0601													2.00									
0602													2.00									
0603																					2.00	
0607			2.00	1.83		1.75						1.50										
0608									1.67	1.88	1.83									1.75		
0610				2.00																	1.88	
0618															1.50							0.75
0629																					1.38	
0631	1.83												2.00									
0632	1.50			1.83	2.00		1.92	2.00		1.75	2.00								1.67	2.00	1.62	
0633																					1.59	
0635			2.00																			1.50

APPENDIX 4 – EQA Material Request Rates (2010)

SCHEME	2008	2009	No. EXTRA SAMPLE REQUESTS	No. SAMPLES SENT OUT	EXTRA SAMPLE REQUESTS %
AZF	5	0	5	432	1.57
BRCA	3	22	4	327	1.22
CAH	7	5	2	138	1.45
CMT	4	4	1	219	0.46
DFNB1	0	1	1	171	0.58
DM	7	5	4	180	2.22
DMD	4	0	4	186	2.15
FAP	-	-	0	105	0.00
FRAX (PCR)	-	0	0	129	0.00
FRAX (FULL)	42	10	6	264	2.27
FRDA	4	1	1	168	0.60
HD	3	1	3	288	1.04
HFE	0	1	6	249	2.41
HNPCC	9	1	4	368	1.09
HRF	-	2	1	144	0.69
MEN2	0	0	0	180	0.00
MonoDiab	3	5	1	87	1.15
PKU	3	1	1	60	1.67
POR	0	1	0	57	0.00
PWAS	9	3	3	270	1.11
RB	0	1	0	42	0.00
SCA	0	0	3	192	1.56
SMA	3	0	1	304	0.33
SEQ	11	37	17	800	2.13
VHL	7	2	1	123	0.81
WIL	0	1	2	99	2.02
TOTAL			71	5582	1.27%
2009			113	6055	1.86%
2008			124	4548	3.02%

APPENDIX 5 – Reporting Compliance Rates (2010)

Scheme ⁷	No. participants	By closing date	By genotype results date (7 days later)
AZF	142	97%	99%
BRCA (Full)	70	99%	99%
BRCA (Genotyping)	39	100%	100%
CAH	46	100%	100%
CMT	73	97%	100%
DFNB1	57	96%	100%
DM	60	98%	100%
DMD	62	98%	100%
DNA-SEQ full	168	95%	96%
FAP	35	97%	97%
FRAX (Full)	88	92%	98%
FRAX (PCR only)	43	95%	100%
FRDA	57	96%	96%
HD	96	97%	100%
HRF	36	97%	97%
HFE	82	96%	100%
HNPCC	91	99%	100%
Men2	60	98%	98%
Microarray / array CGH	26 ⁸	92%	92%
MonoDiab	29	100%	100%
PKU	20	95%	100%
POR	19	100%	100%
PWAS	90	96%	99%
RB	14	93%	93%
SCA	64	95%	98%
SMA	76	96%	99%
VHL	41	98%	98%
WIL	33	91%	100%

⁷ Not including DNA SEQ quality only returns

⁸ No of participants registered with EMQN, not including CEQA participants