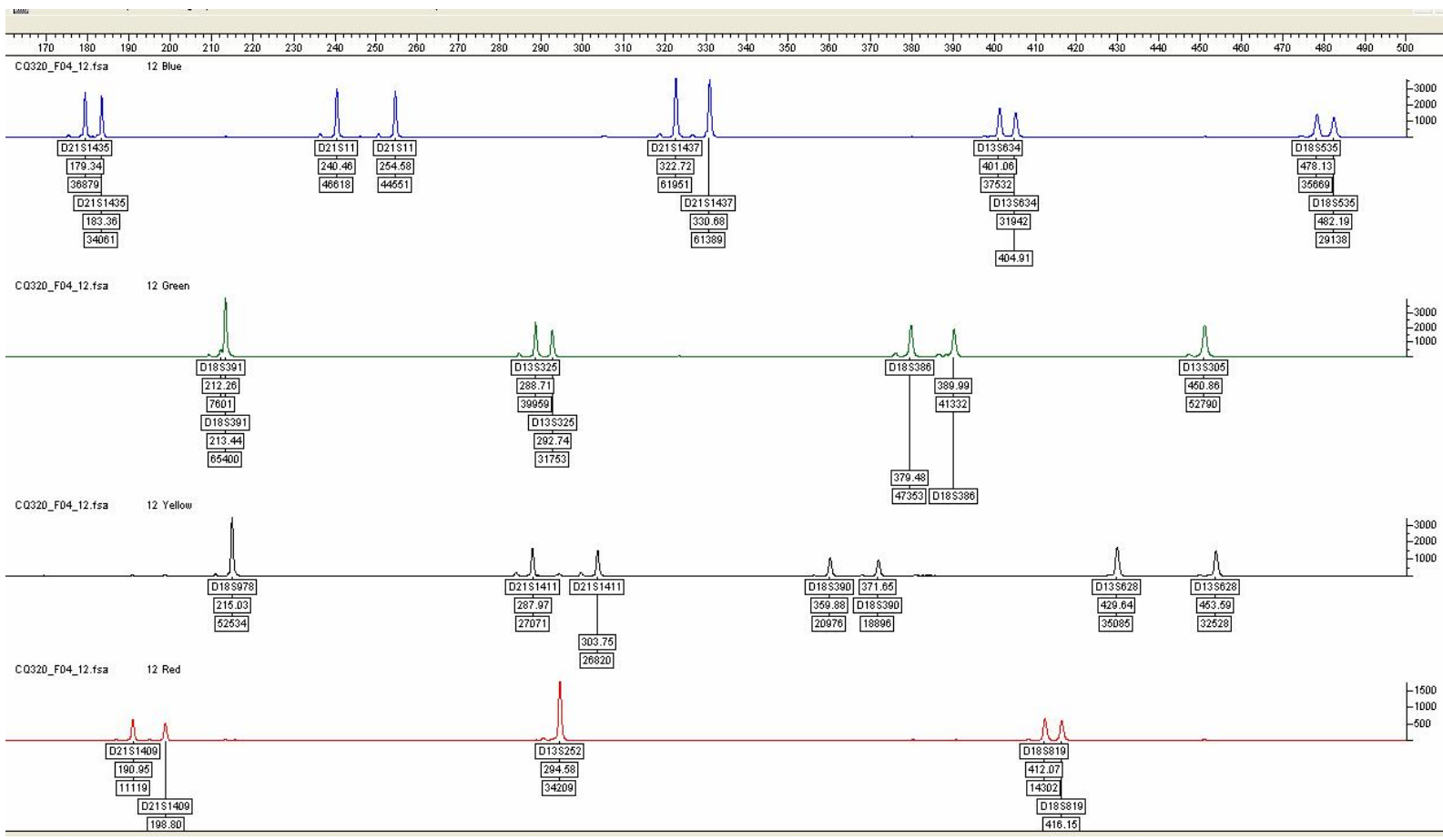




APPENDIX 1

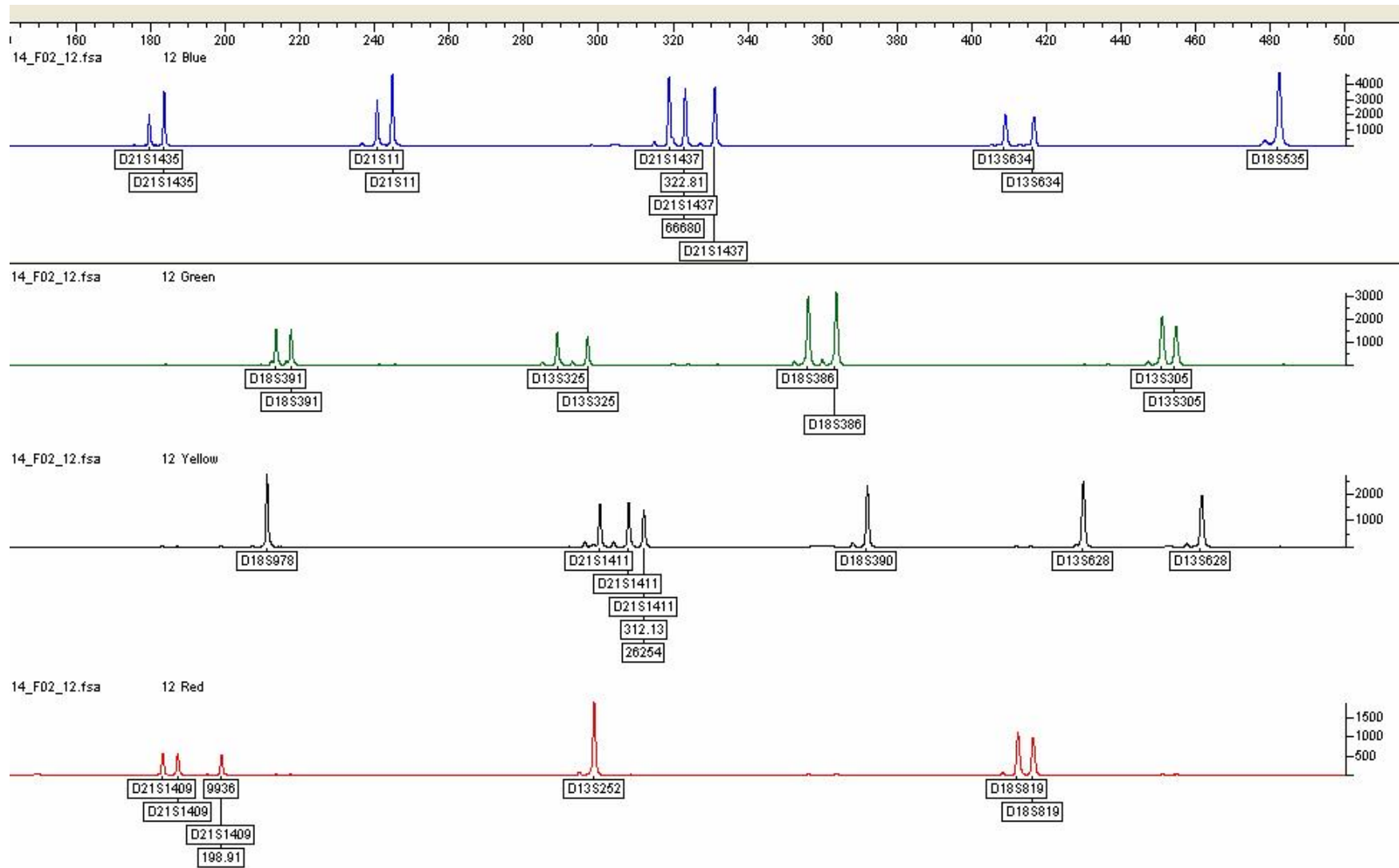
EXAMPLES OF QST*R TRACES FROM RETROSPECTIVELY COLLECTED TISSUE SAMPLES AND AMNIOTIC FLUID SAMPLES FROM NGRL (WESSEX)




CQ320: Example of Normal Karyotype

				TRISOMY		Sample ID		CQ320			
				Run:				Co-ord:		CQ3	
				Filename:		CQ320_F04_12.fsa					
Marker	Area 1	Area 2	Area 3	A1/A2	Warning	1 st Check	2 nd Check	3 rd Check	Comment		
D13S252	34209										
D13S305	52790										
D13S325	39959	31753		1.26							
D13S628	35085	32528		1.08							
D13S634	37532	31942		1.18							
D18S386	47353	41332		1.15							
D18S390	20976	18896		1.11							
D18S391	65400										
D18S535	35669	29138		1.22							
D18S819	14302	13282		1.08							
D18S978	52534										
D21S11	46618	44551		1.05							
D21S1409	11119	9546		1.16							
D21S1411	27071	26820		1.01							
D21S1435	36879	34061		1.08							
D21S1437	61951	61389		1.01							
NOTES:					CONCLUSION:						
					Normal						
										CHECKED:	
					v 1.4						

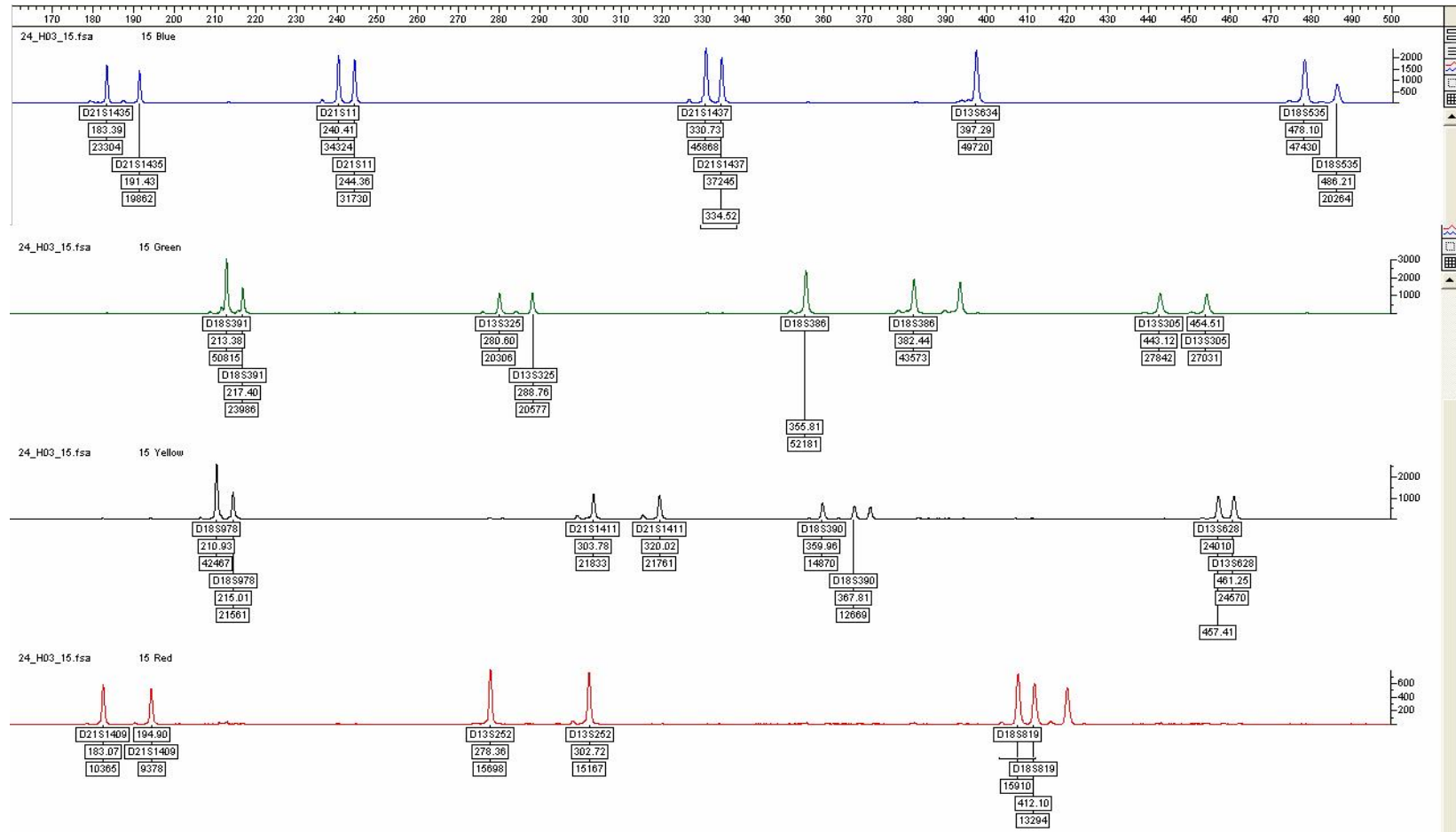
Example of QST*R Trisomy report for Sample CQ320 (Normal)



Ret 14: Example of Trisomy 21

QST*R™				TRISOMY		Sample ID		ret14		
Run:						Co-ord:		14_		
Filename:				14_F02_12.fea						
Marker	Area 1	Area 2	Area 3	A1/A2	Warning	1 st Check	2 nd Check	3 rd Check	Comment	
D13S252	38888									
D13S305	52624	43570		1.21						
D13S325	27415	23528		1.17						
D13S628	54312	45868		1.18						
D13S634	43539	41731		1.04						
D18S386	66147	71229		0.93						
D18S390	48256									
D18S391	27396	27201		1.01						
D18S535	126698									
D18S819	24902	22894		1.09						
D18S978	47013									
D21S11	48620	78922		0.62	Ratio					
D21S1409	10741	10305	9936	1.04	3 Alleles					
D21S1411	29512	30887	26254	0.96	3 Alleles					
D21S1435	29772	50247		0.59	Ratio					
D21S1437	80698	66680	70150	1.21	3 Alleles					
NOTES:					CONCLUSION:					
					T21+ve					
CHECKED:							DATE:			
					v 1.4					


Example of QST*R Trisomy report for Sample ret 14 (Trisomy 21)



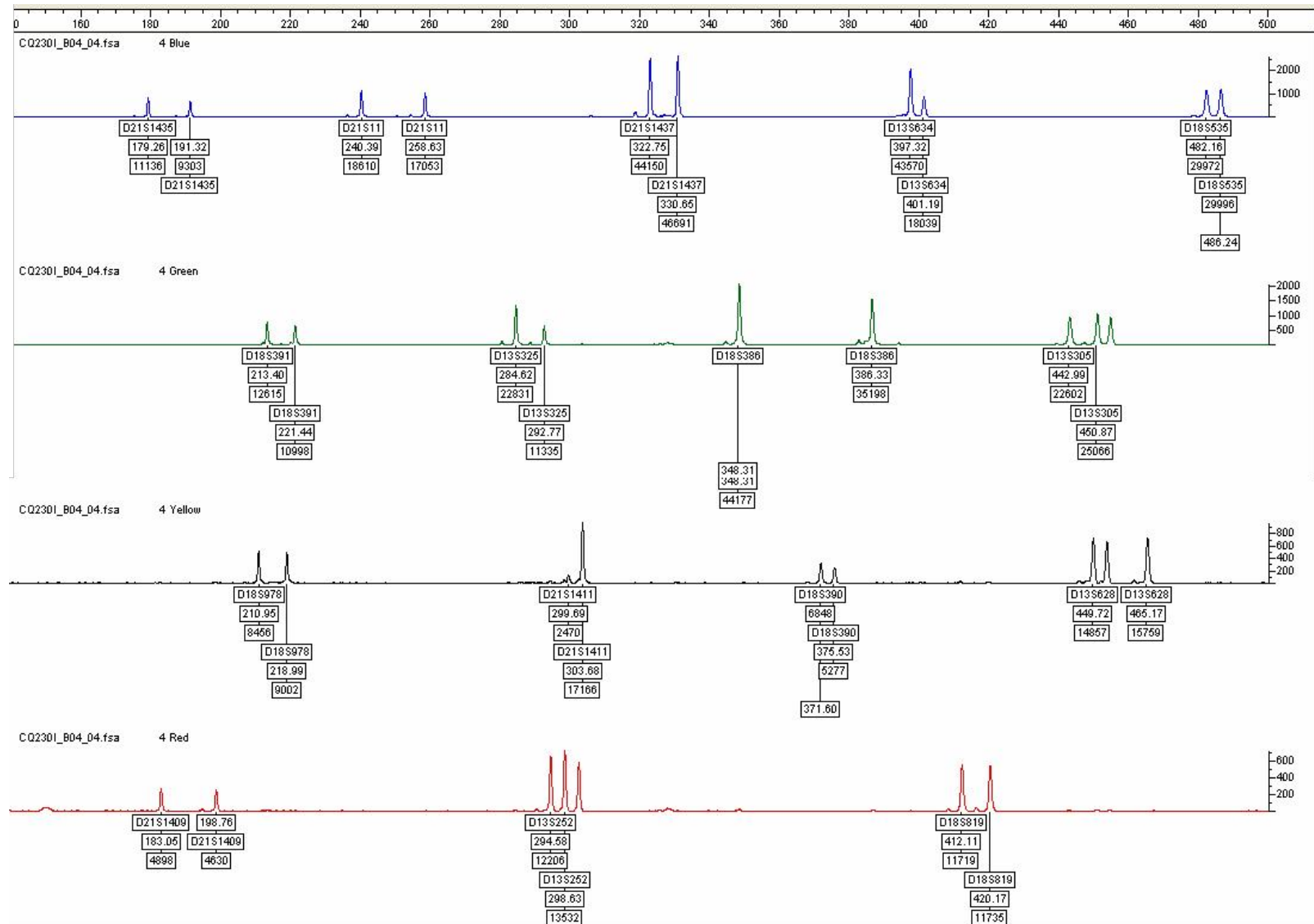
Ret 24: Example of Trisomy 18

QST*R™				TRISOMY		Sample ID		ret24		
Run:						Co-ord:		24_		
Filename:				24_H03_15.fsa						
Marker	Area 1	Area 2	Area 3	A1/A2	Warning	1 st Check	2 nd Check	3 rd Check	Comment	
D13S252	15698	15167		1.04						
D13S305	27842	27031		1.03						
D13S325	20306	20577		0.99						
D13S628	24010	24570		0.98						
D13S634	49720									
D18S386	52181	43573	40011	1.20	3 Alleles					
D18S390	14870	12669	12140	1.17	3 Alleles					
D18S391	50815	23986		2.12	Ratio					
D18S535	47430	20264		2.34	Ratio					
D18S819	15910	13294	11902	1.20	3 Alleles					
D18S978	42467	21561		1.97	Ratio					
D21S11	34324	31730		1.08						
D21S1409	10365	9378		1.11						
D21S1411	21833	21761		1.00						
D21S1435	23304	19862		1.17						
D21S1437	45868	37245		1.23						



NOTES:	CONCLUSION:		
	T18		
CHECKED:		DATE:	

				v1.4			
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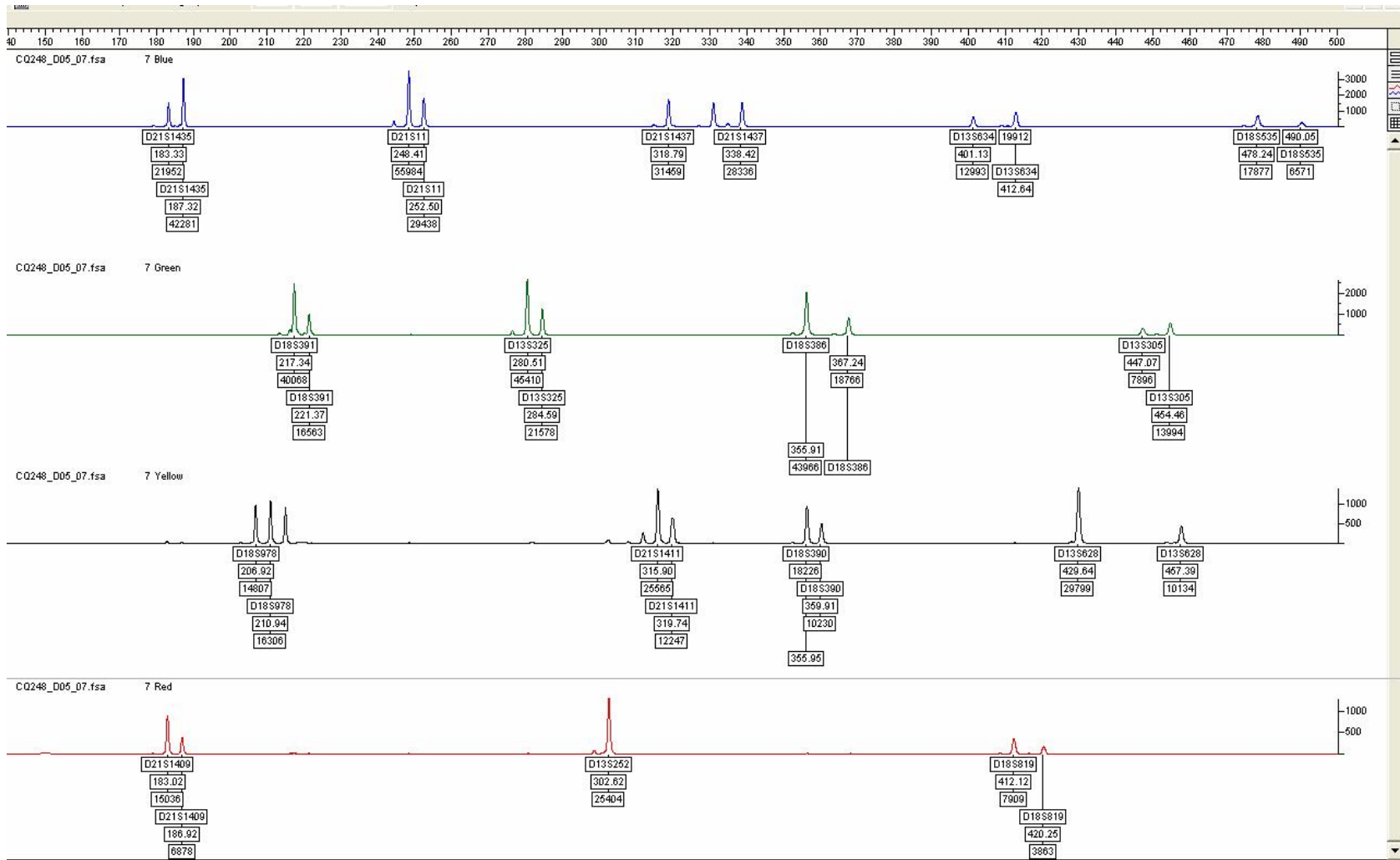
Example of QST*R Trisomy report for ret 24 (Trisomy 18)



CQ230I: Example of Trisomy 13

				TRISOMY		Sample ID		CQ230I			
				Run:				Co-ord:		CQ2	
				Filename:		CQ230I_B04_04.fcs					
Marker	Area 1	Area 2	Area 3	A1/A2	Warning	1 st Check	2 nd Check	3 rd Check	Comment		
D13S252	12206	13532	11035	0.90	3 Alleles						
D13S305	22602	25066	21818	0.90	3 Alleles						
D13S325	22831	11335		2.01	Ratio						
D13S628	14857	13860	15759	1.07	3 Alleles						
D13S634	43570	18039		2.42	Ratio						
D18S386	44177	35198		1.26							
D18S390	6848	5277		1.30							
D18S391	12615	10998		1.15							
D18S535	29972	29996		1.00							
D18S819	11719	11735		1.00							
D18S978	8456	9002		0.94							
D21S11	18610	17053		1.09							
D21S1409	4898	4630		1.06							
D21S1411	17166										
D21S1435	11136	9303		1.20							
D21S1437	44150	46691		0.95							
NOTES:					CONCLUSION:						
					T13						
					CHECKED:				DATE:		
					v1.4						

Example of QST*R Trisomy report for Sample CQ230I (Trisomy 13)



CQ248: Example of Triploid

QST*R™				TRISOMY		Sample ID		CQ248			
Run:								Co-ord:		CQ2	
Filename:				CQ248_D05_07.fsa							
Marker	Area 1	Area 2	Area 3	A1/A2	Warning	1 st Check	2 nd Check	3 rd Check	Comment		
D13S252	25404										
D13S305	7896	13994		0.56	Ratio						
D13S325	45410	21578		2.10	Ratio						
D13S628	29799	10134		2.94	Ratio						
D13S634	12993	19912		0.65	Ratio						
D18S386	43966	18766		2.34	Ratio						
D18S390	18226	10230		1.78	Ratio						
D18S391	40068	16563		2.42	Ratio						
D18S535	17877	6571		2.72	Ratio						
D18S819	7909	3863		2.05	Ratio						
D18S978	14807	16306	13664	0.91	3 Alleles						
D21S11	55984	29438		1.90	Ratio						
D21S1409	15036	6878		2.19	Ratio						
D21S1411	25565	12247		2.09	Ratio						
D21S1435	21952	42281		0.52	Ratio						
D21S1437	31459	27850	28336	1.13	3 Alleles						
NOTES:					CONCLUSION:						
					Triploid						
					CHECKED:				DATE:		



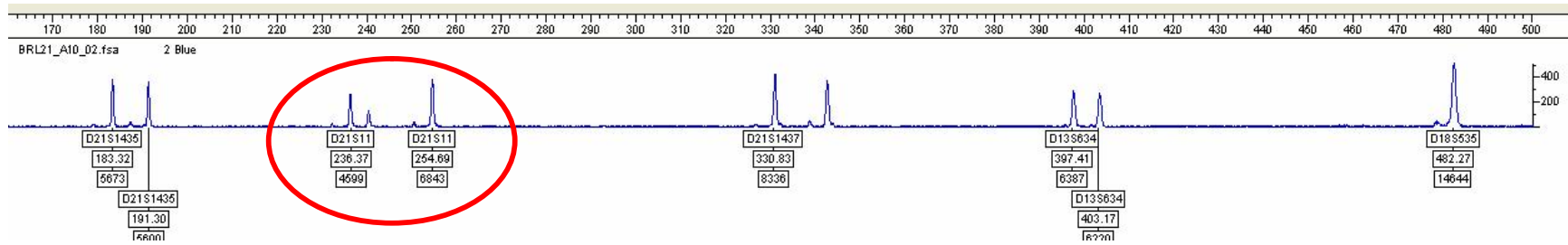
v1.4

Example of QST*R Trisomy report for Sample CQ248 (Triploid)

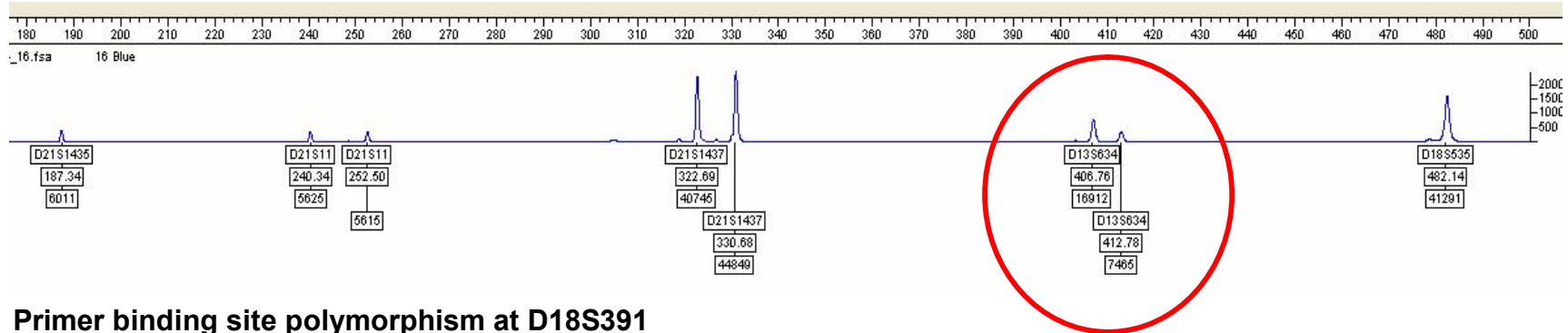
APPENDIX 2

EXAMPLES OF QST*R TRACES FROM UNUSUAL PRENATAL SAMPLES SUBMITTED FROM OTHER UK LABORATORIES

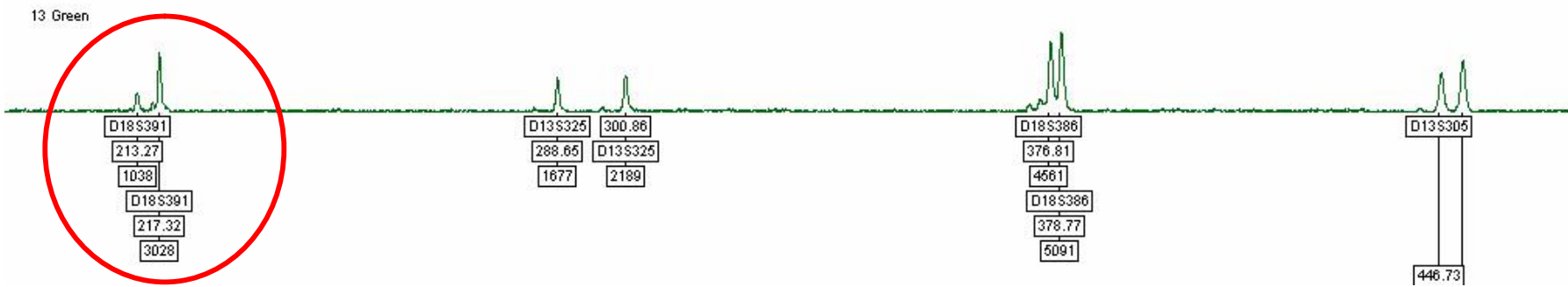
Somatic microsatellite mutation at D21S11

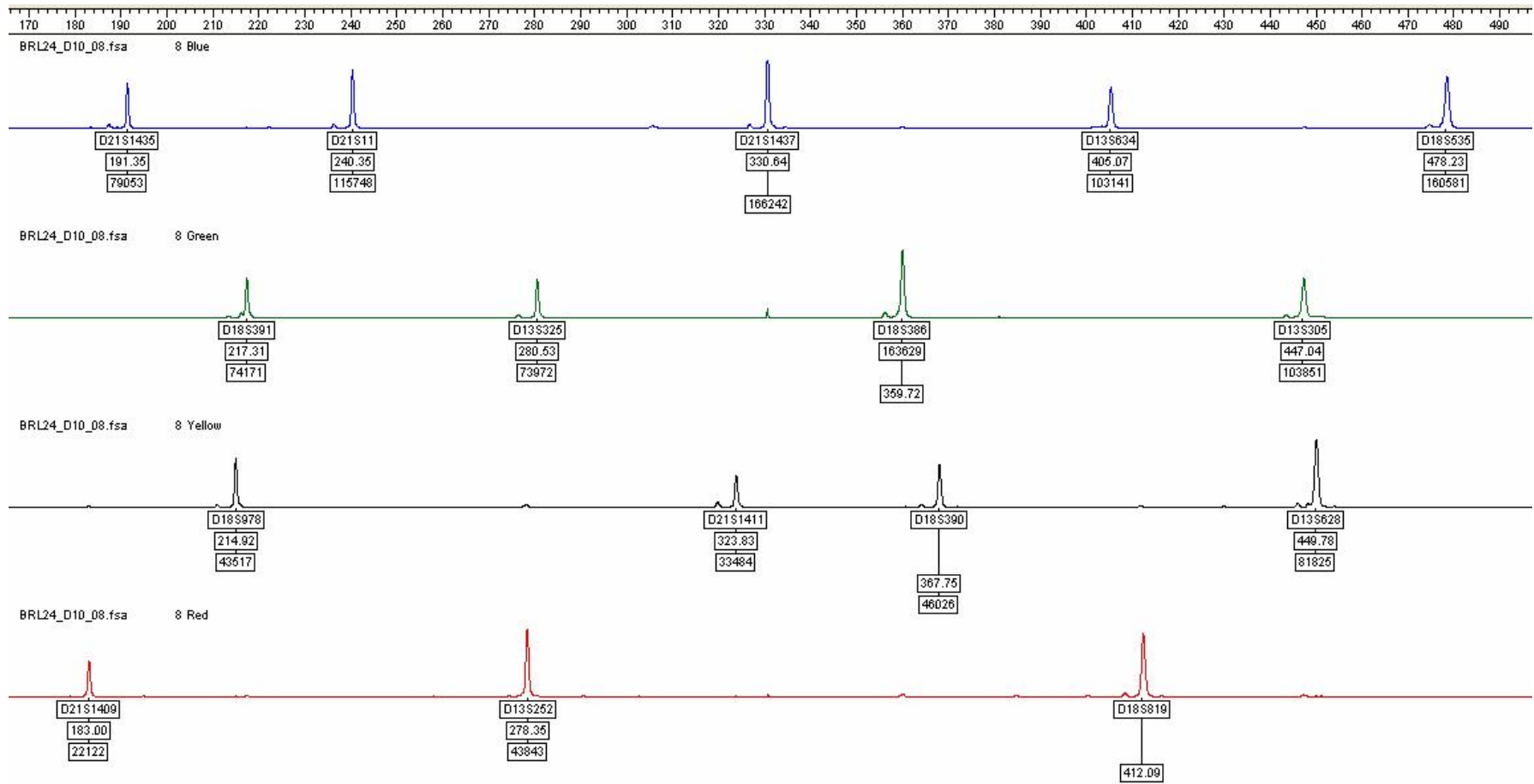


Submicroscopic duplication at D13S634pat

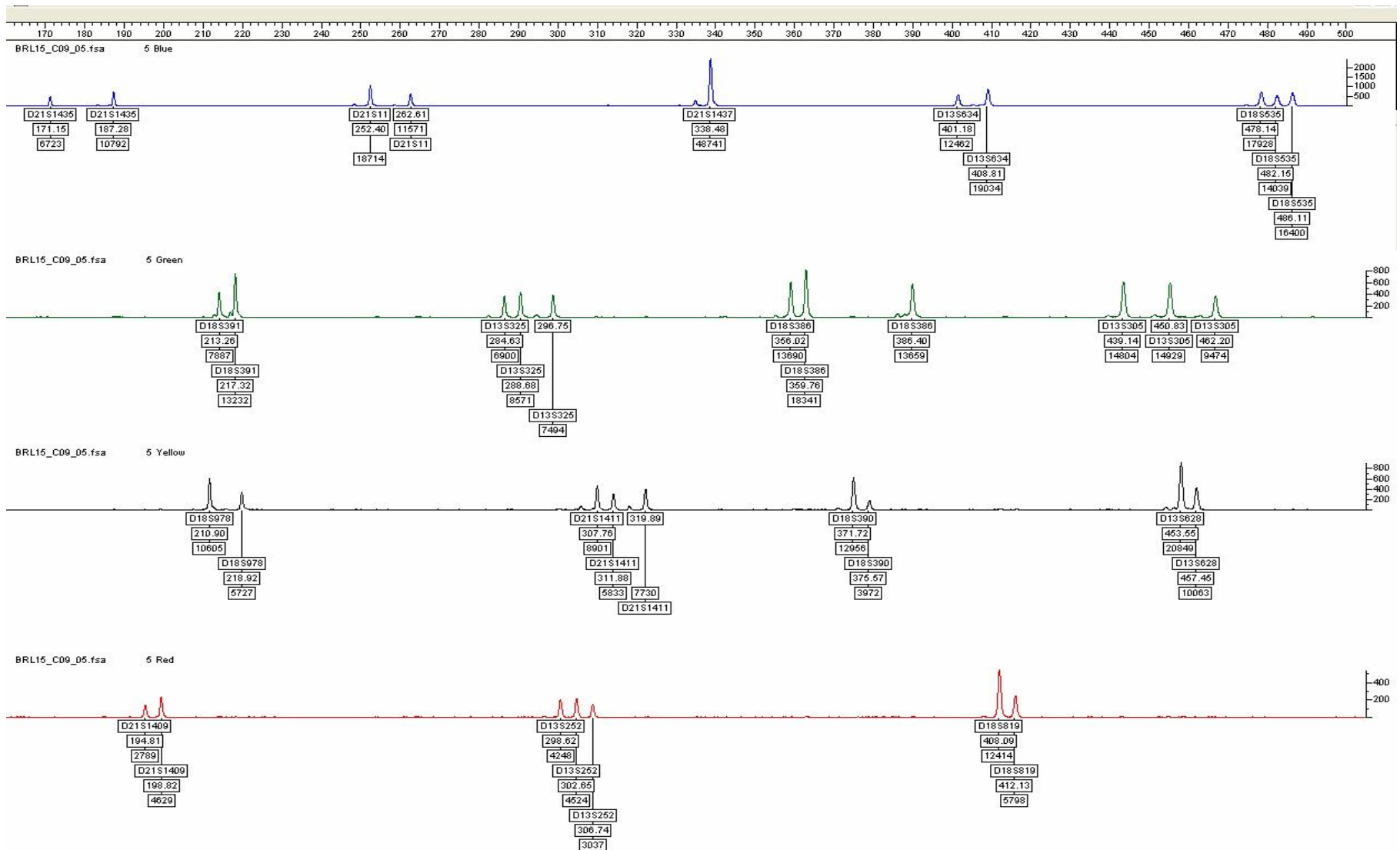


Primer binding site polymorphism at D18S391

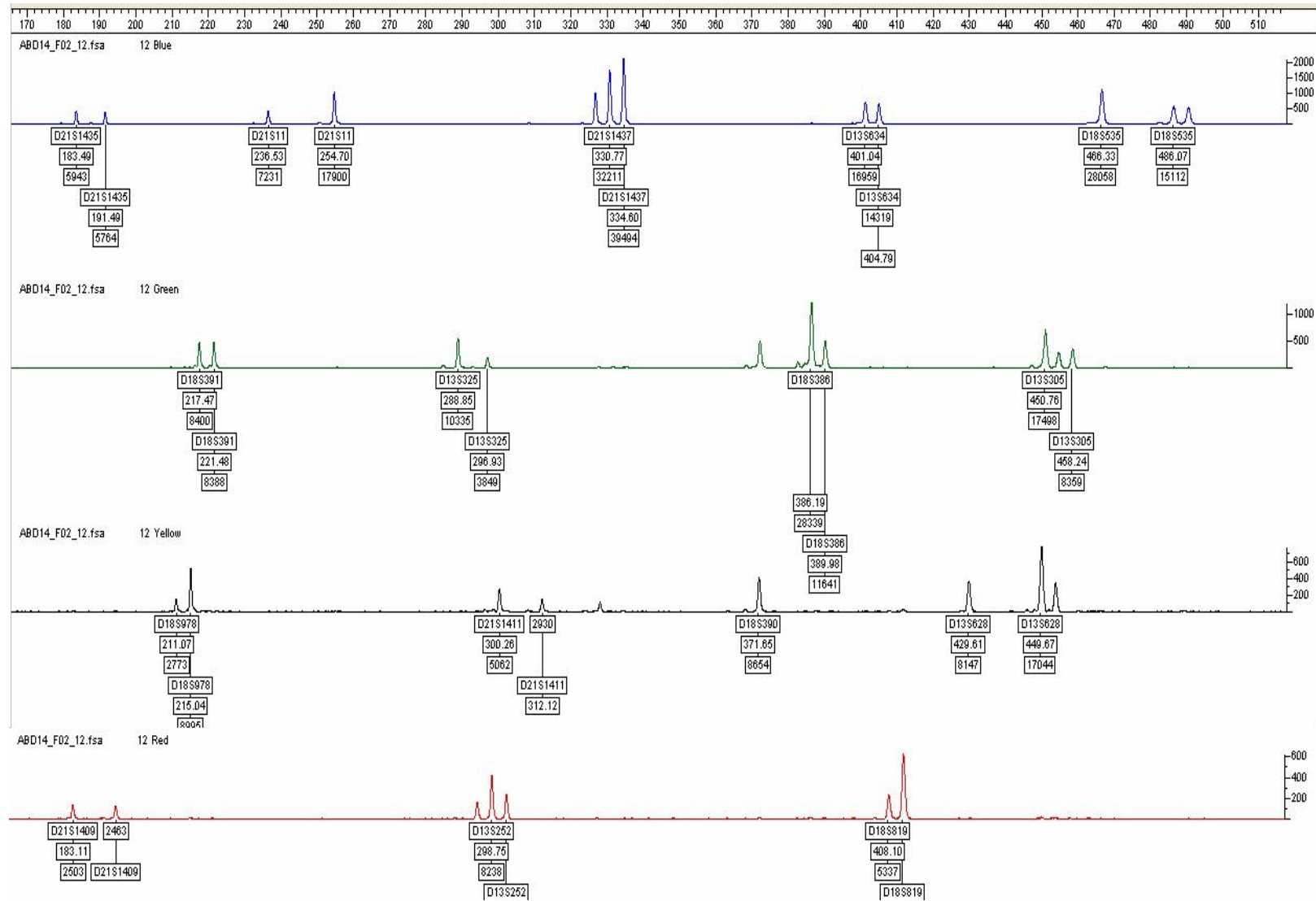




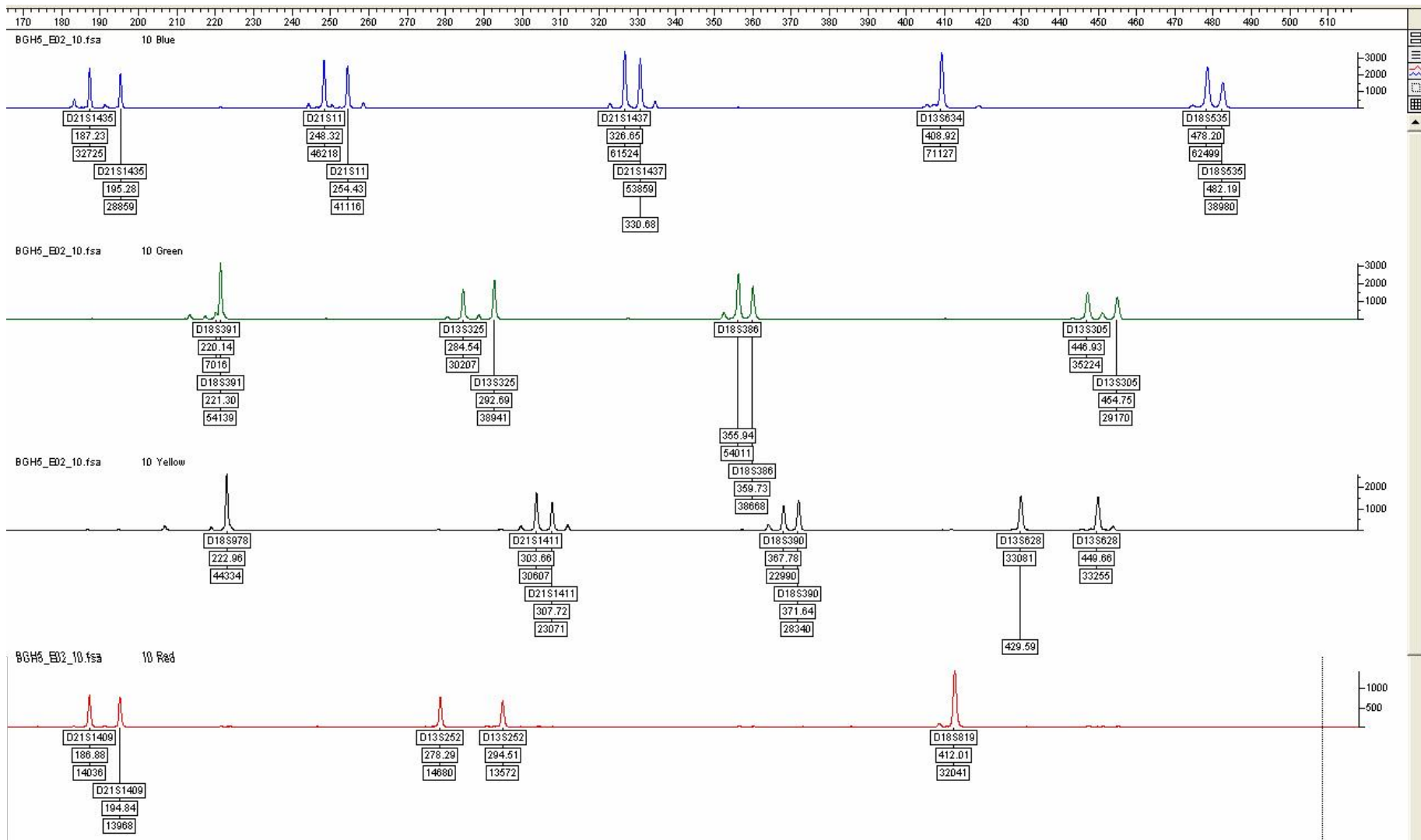
BRL24: Example of female mole



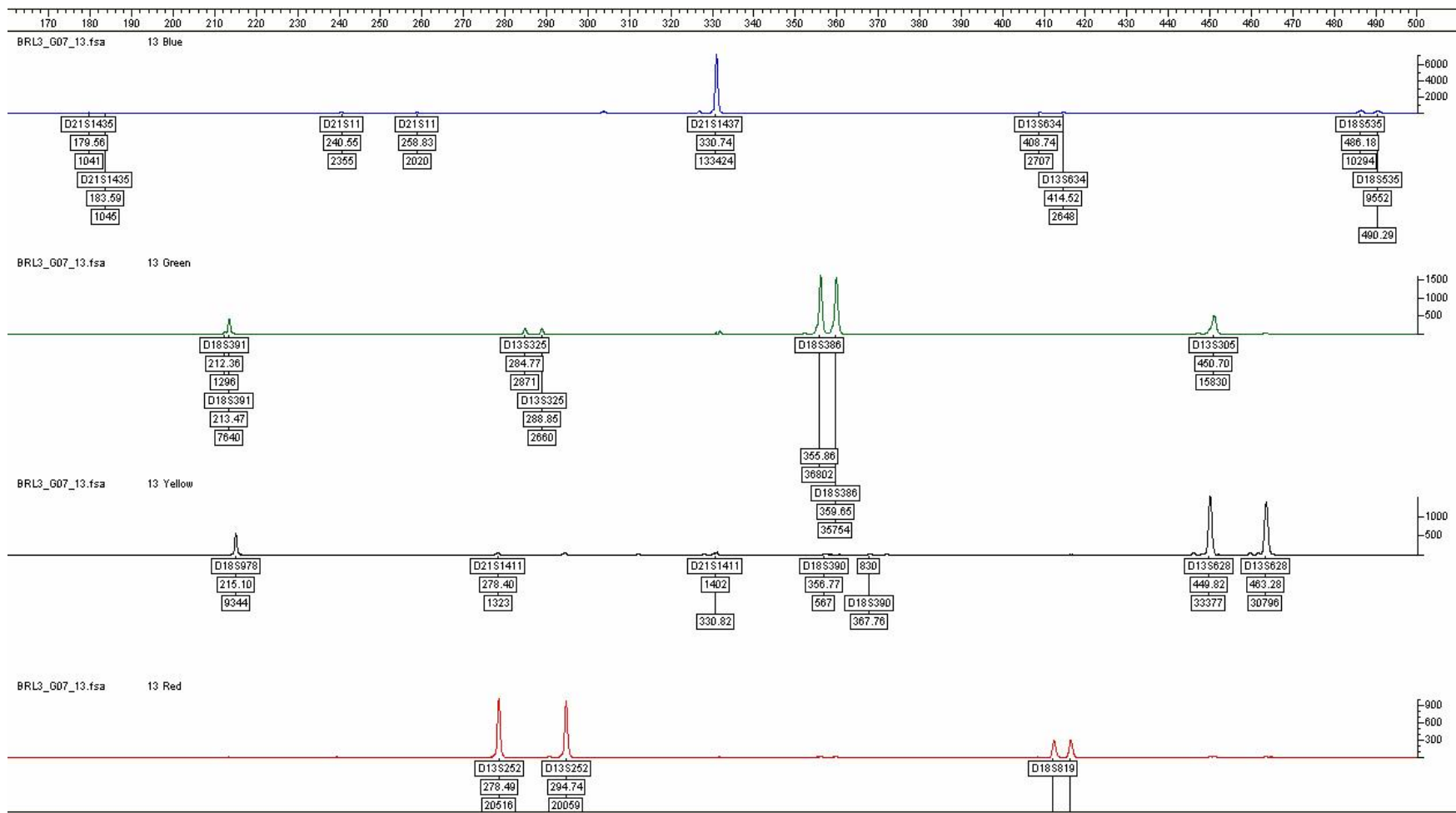
BRL15: Example of a mosaic triploid sample



ABN 14: Example of maternal cell contamination supplied by lab1



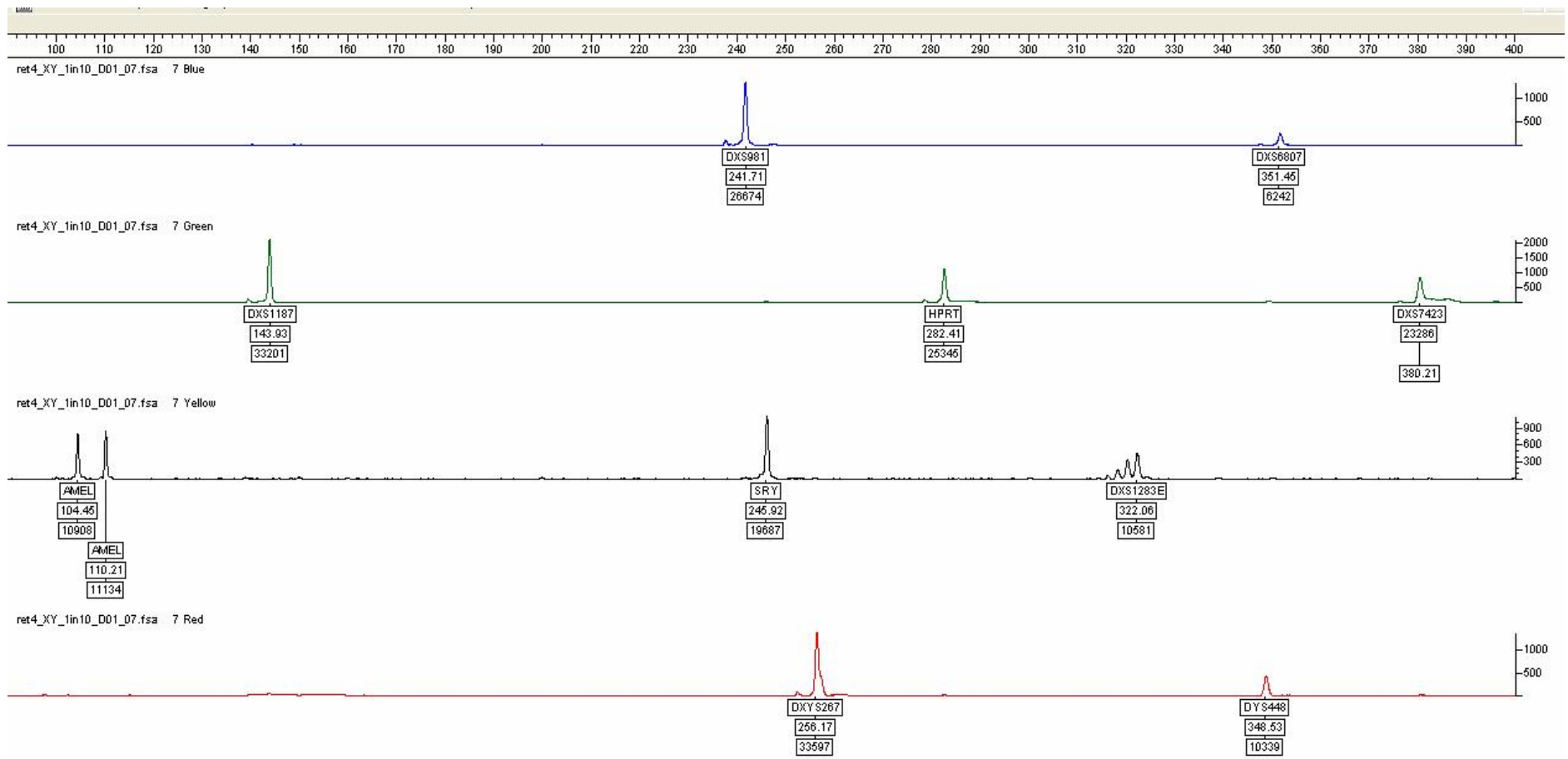
BGH 5: Example of maternal cell contamination supplied by lab2



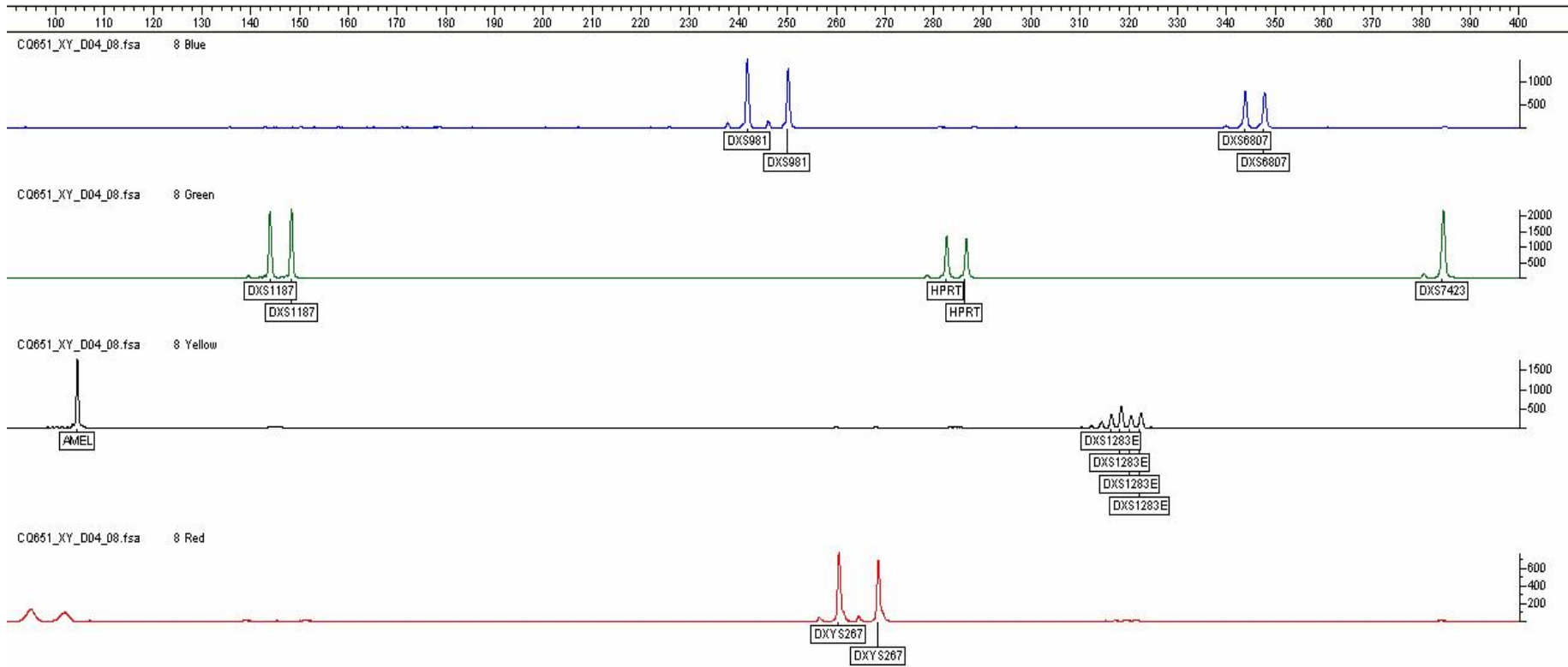
BRL 3: Example of a sample where some markers have amplified more efficiently than others – interestingly this is independent of the molecular weight of the marker products.

APPENDIX 3

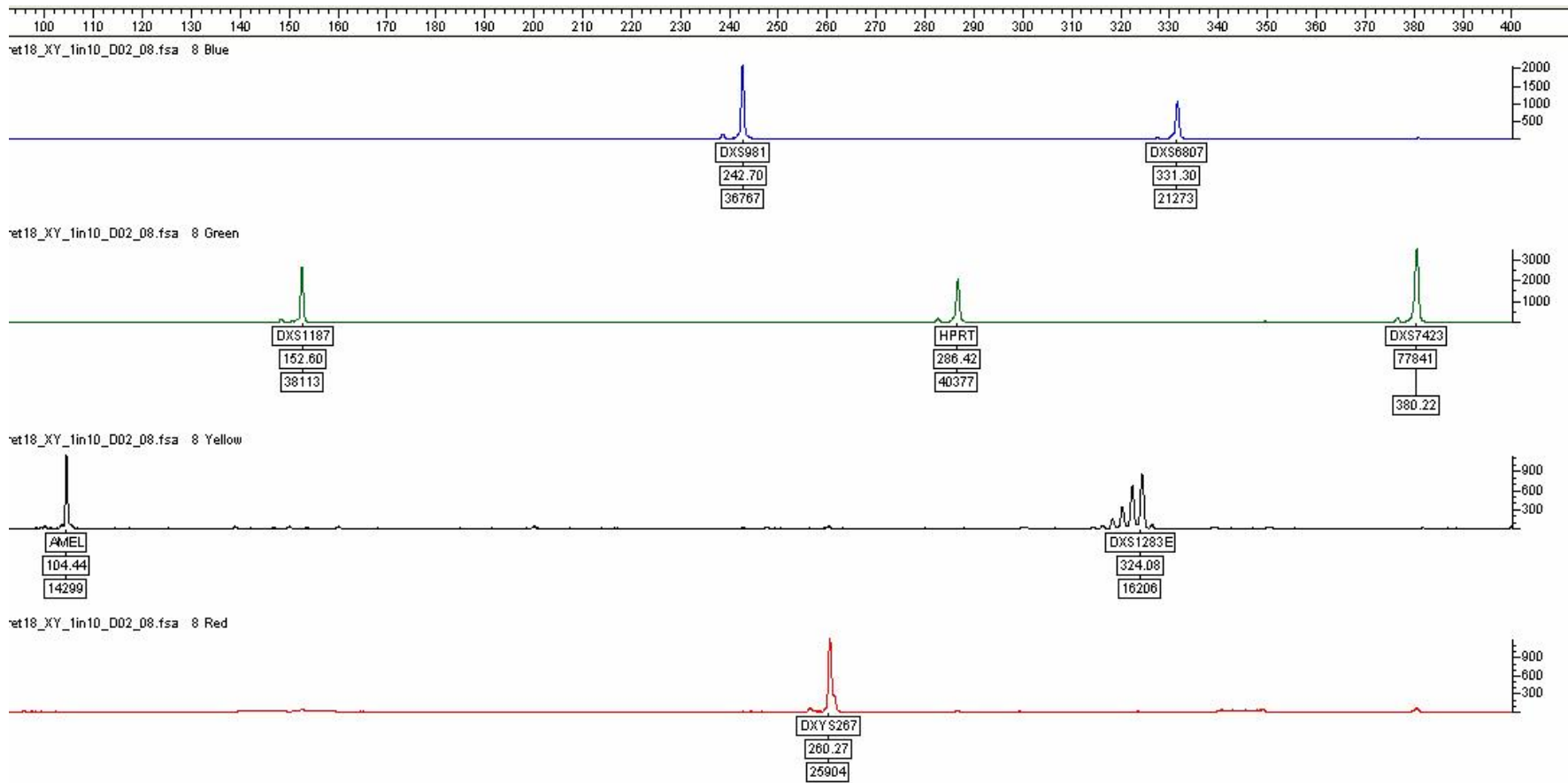
EXAMPLES OF QST*R-XY TRACES FOR A VARIETY OF SEX CHROMOSOME ANEUPLOIDY



Ret 4: Example of a normal male



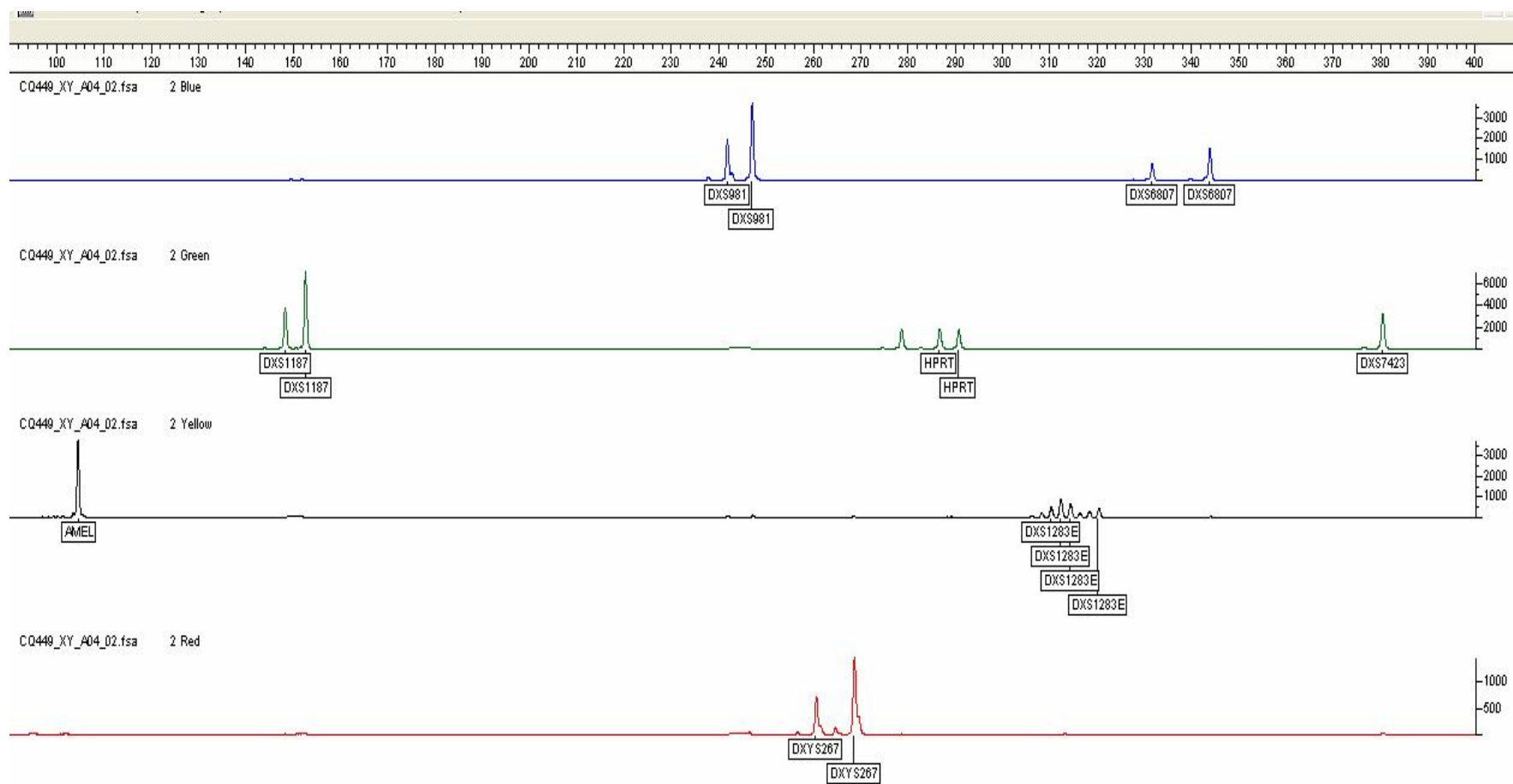
CQ651: Example of a normal female



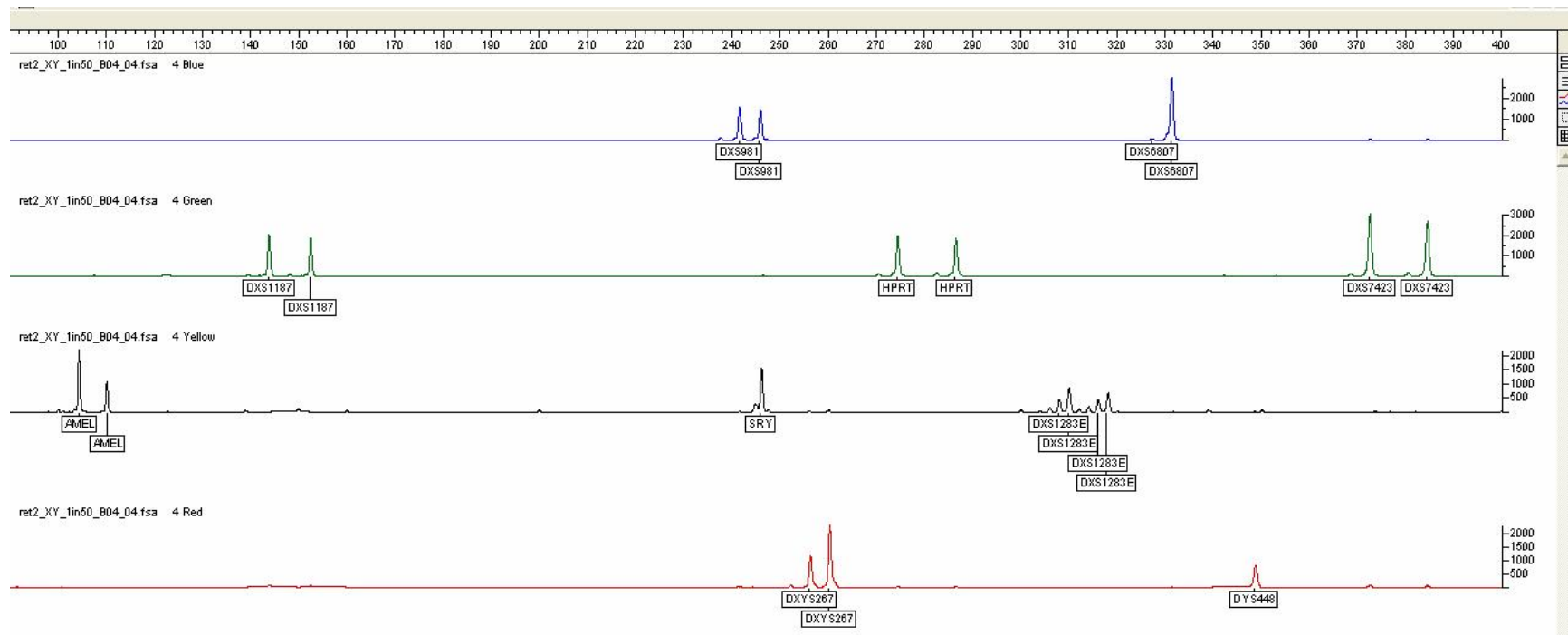
Ret 18: Example of sample with a karyotype of 45,X



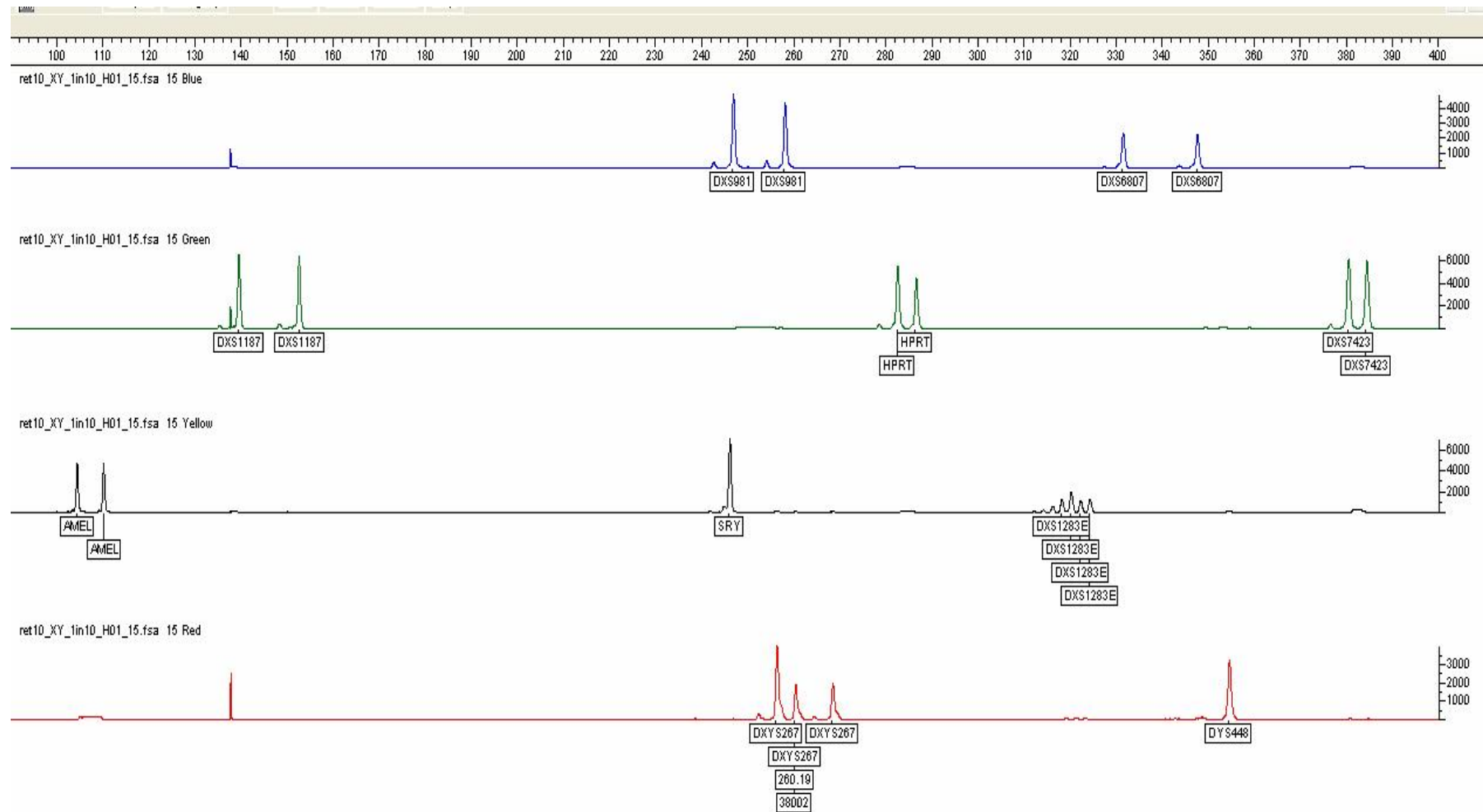
CQ600: Example of sample with a karyotype of 45,X where Y chromosome sequences (circled in red) have been amplified at a low level (c. 10%)



CQ449: Example of sample with a karyotype of 47,XXX



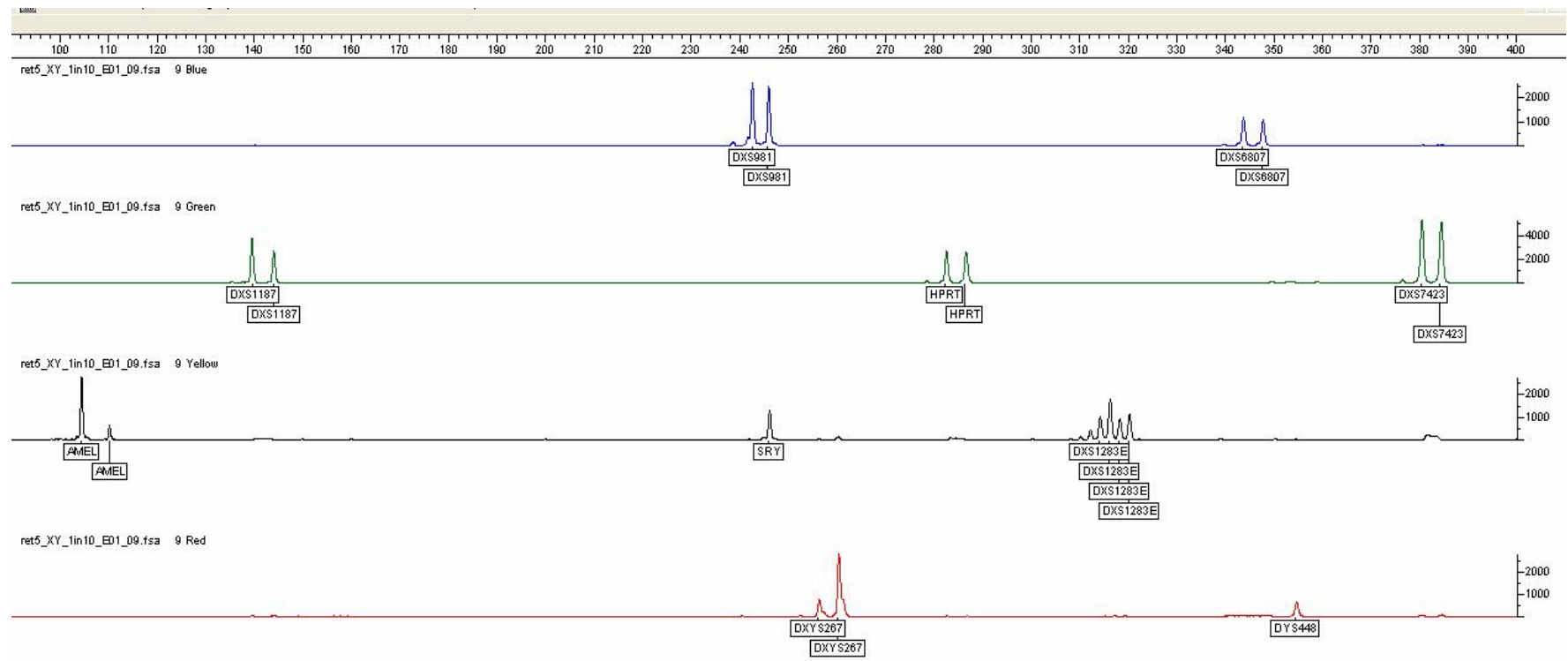
Ret 2: Example of sample with a karyotype of 47,XXY



Ret 10: Example of sample with a karyotype of 48,XXYY



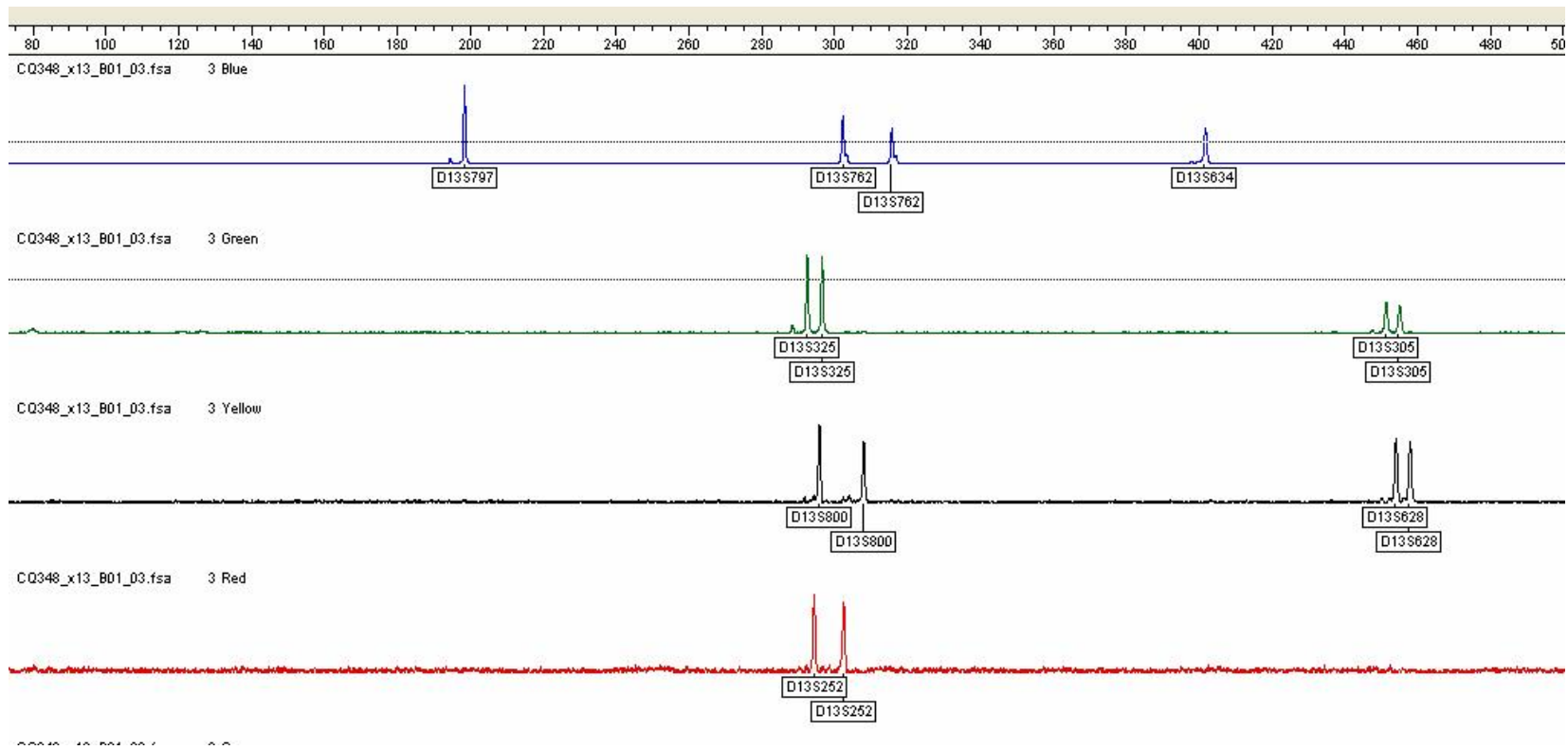
Ret 85: Example of sample with a karyotype of 48,XXXXY



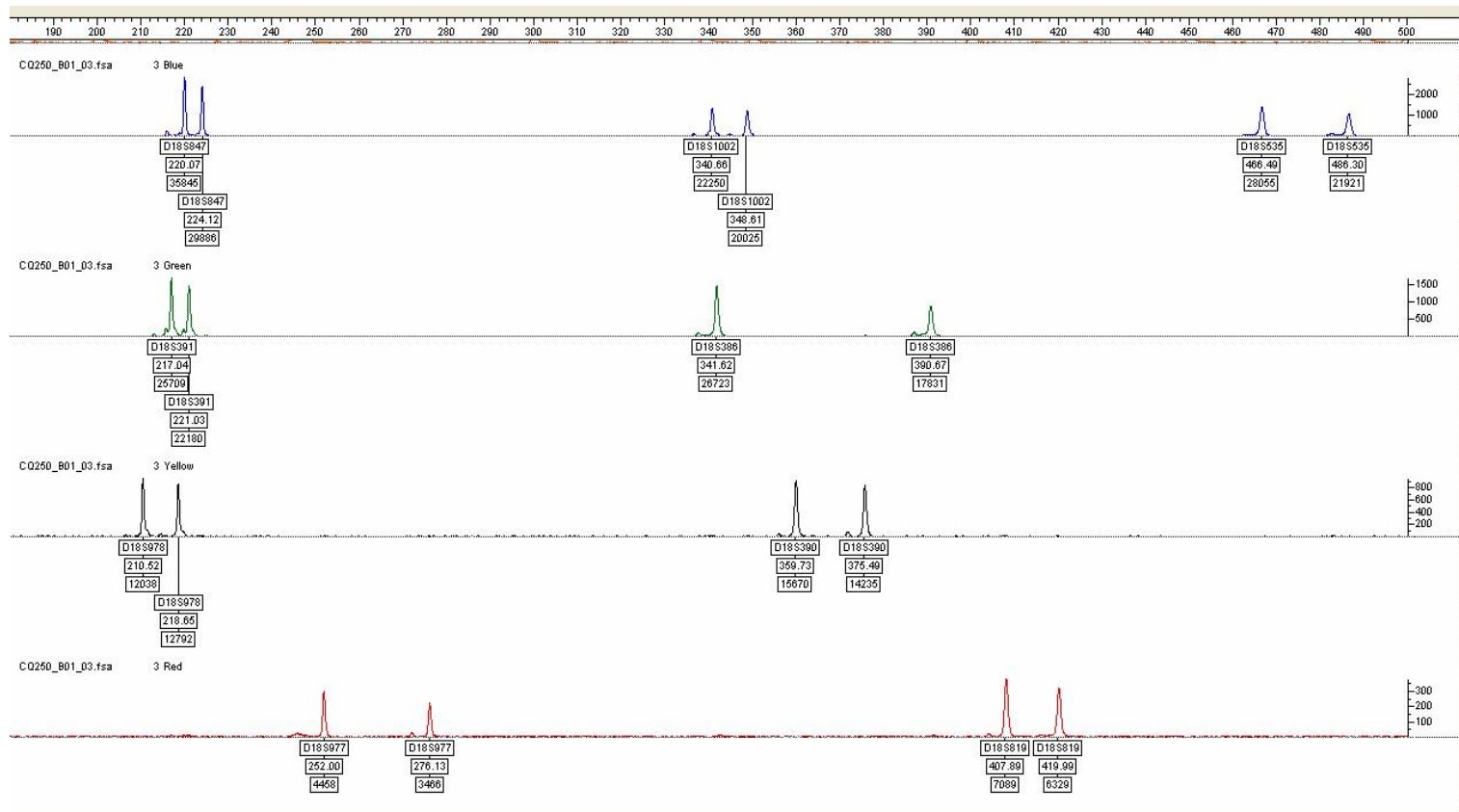
Ret 5: Example of sample with a karyotype of 49,XXXXY

APPENDIX 4

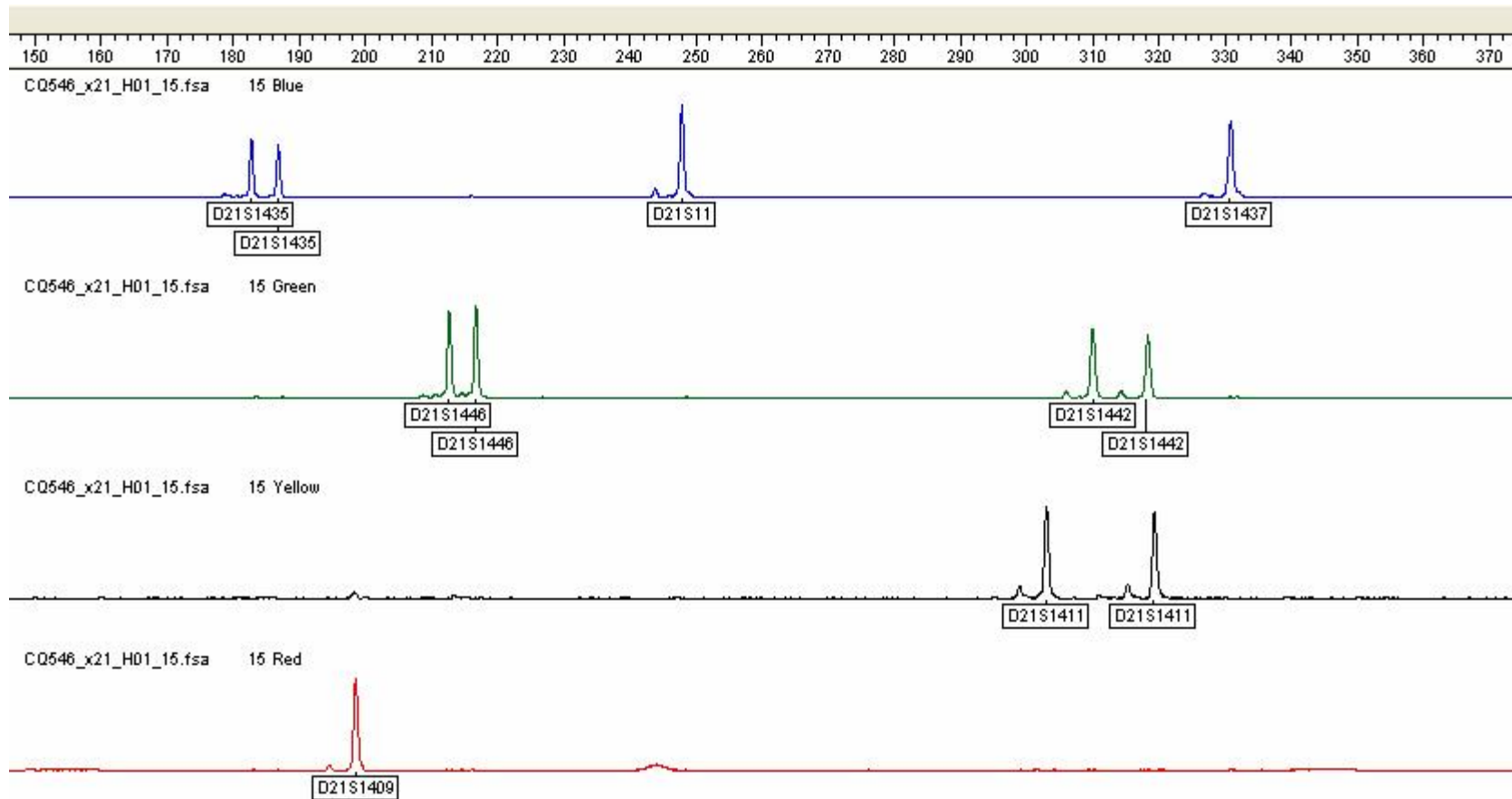
Examples of QST*R-13, QST*R-18 and QST*R-21 traces



CQ348: Example of sample tested with QST*R-13



CQ250: Example of sample tested with QST*R-18



CQ546: Example of sample tested with QST*R-21