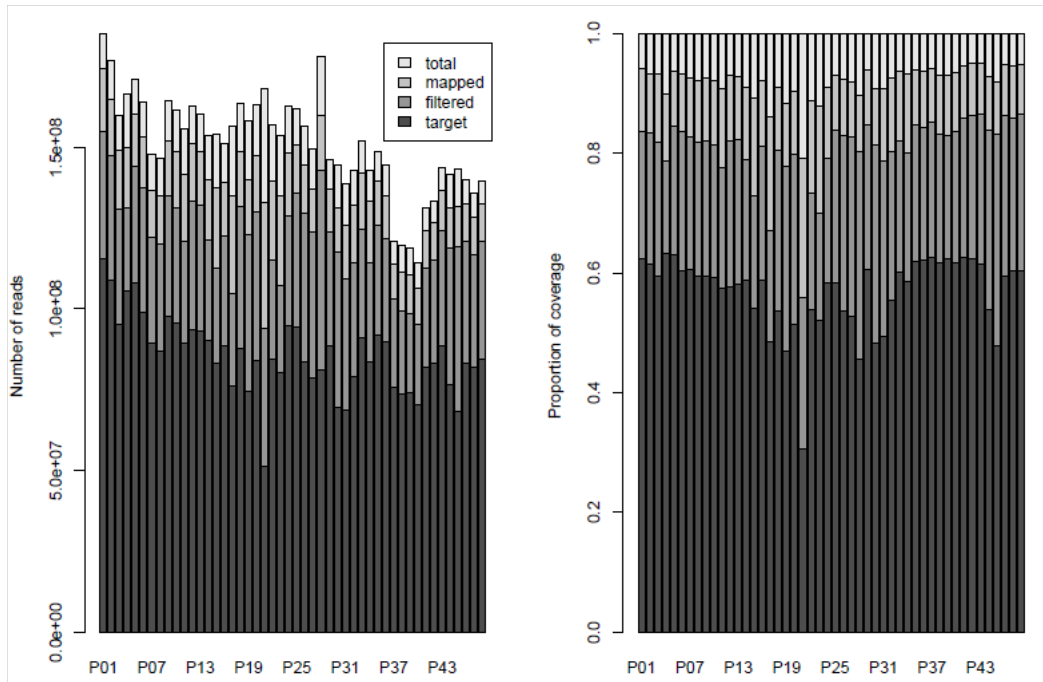


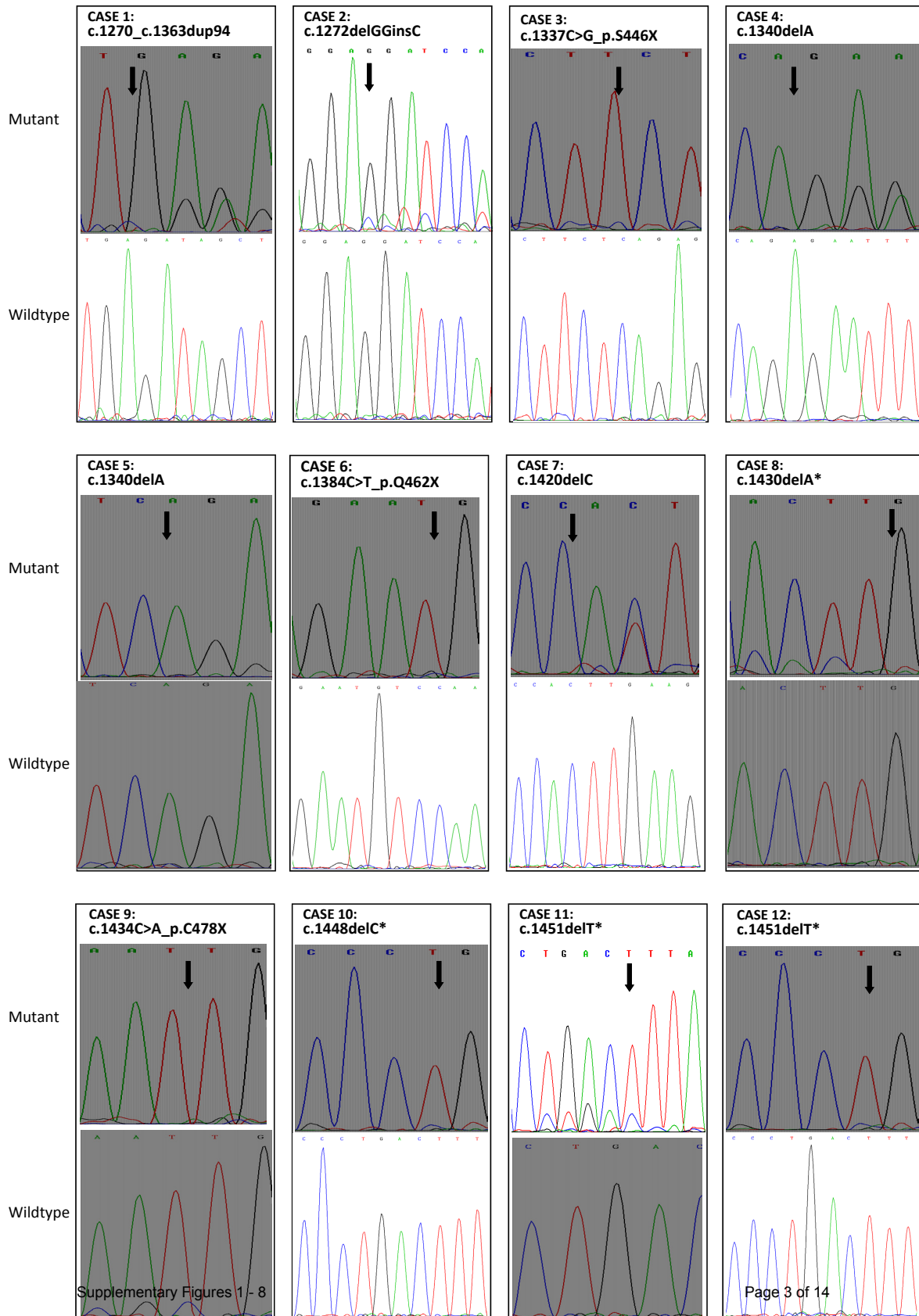
**Supplementary Figure 1.** Samples, sequencing methods and *PPM1D* PTVs identified in different phases of the experiment

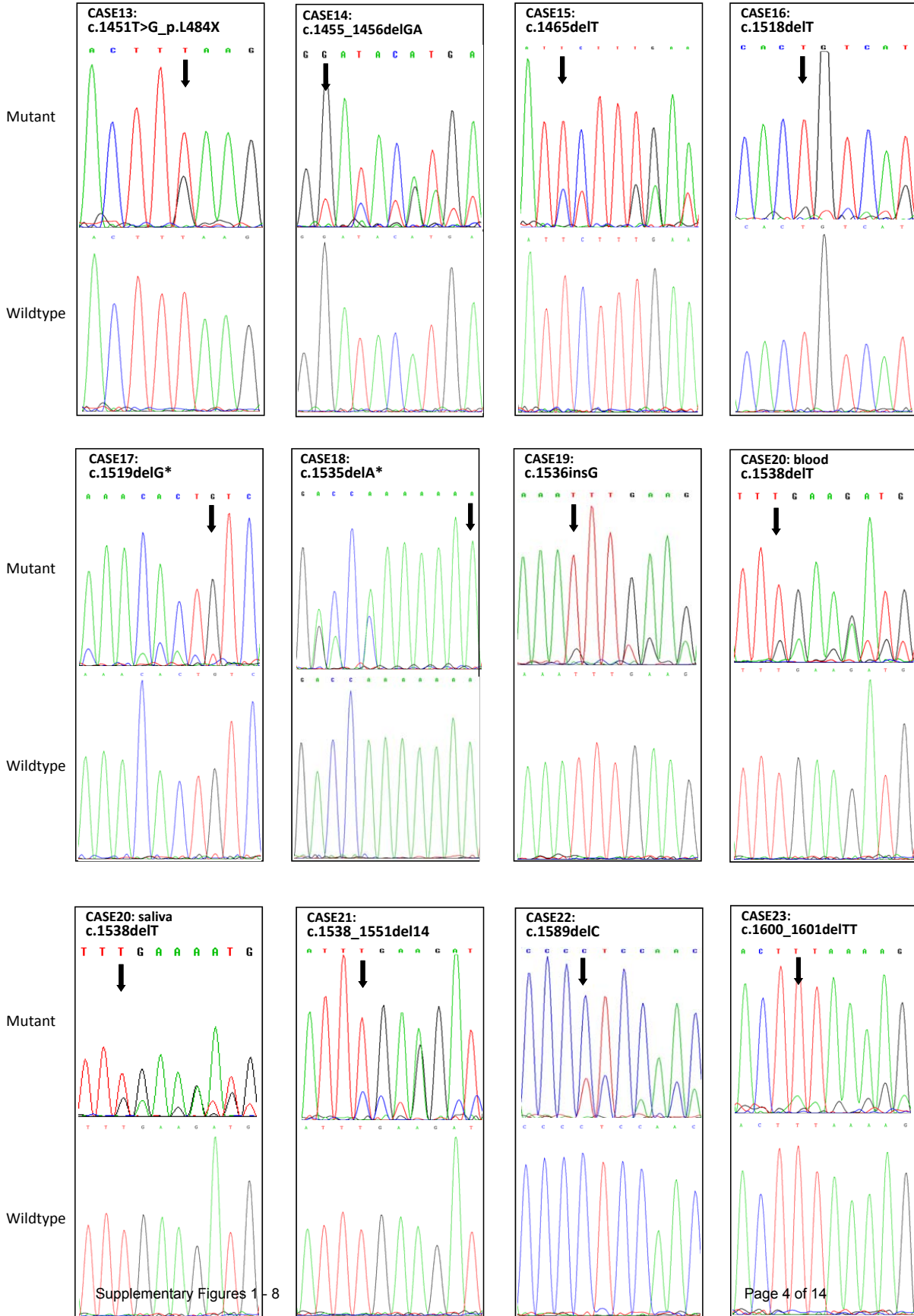
<b>Phase 1— case only DNA repair panel sequencing</b>		
<b>Method</b>	NGS of custom pulldown including 507 DNA repair genes	
<b>Samples</b>	1150 cases (in 48 pools of 24 samples)	
<b>PPM1D PTVs</b>	5 cases	
<b>Phase 2— case-control <i>PPM1D</i> sequencing</b>		
<b>a)</b>		
<b>Method</b>	<i>PPM1D</i> full gene Sanger sequencing	
<b>Samples</b>	2456 cases	1347 controls
<b>PPM1D PTVs</b>	10 cases	0 control
<b>b)</b>		
<b>Method</b>	<i>PPM1D</i> mutation cluster region (MCR) Sanger sequencing	
<b>Samples</b>	5325 cases	4514 controls
<b>PPM1D PTVs</b>	15 cases	1 control

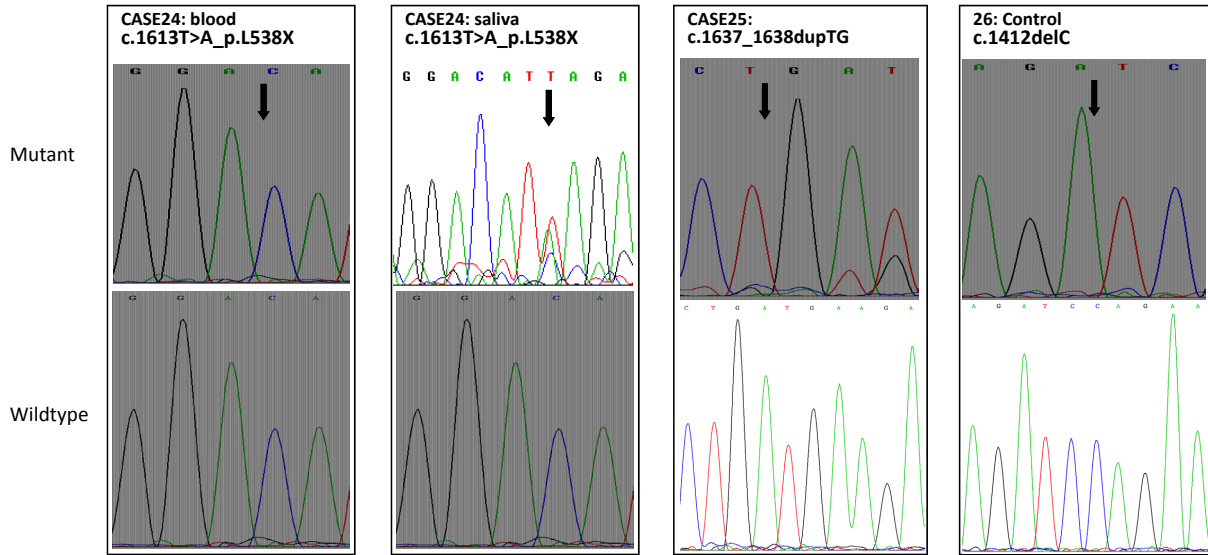
**Supplementary Figure 2: Coverage of custom pulldown by pool**

Bars indicate the absolute (left) and relative (right) number of reads obtained from sequencing the total number of reads (total), mapped to the reference (mapped), remaining after application of quality filters (filtered) and high quality reads mapped to the target regions (target). Mapped reads were filtered to remove ambiguous alignments with a quality score of 0 and bases with a call quality below 22 were masked. Of the remaining reads of each pool 50-60% fell within the target regions, except for Pool 21, where the on target percentage was significantly lower.

**Supplementary Figure 3.** Sanger sequencing chromatograms for 26 *PPM1D* mutations

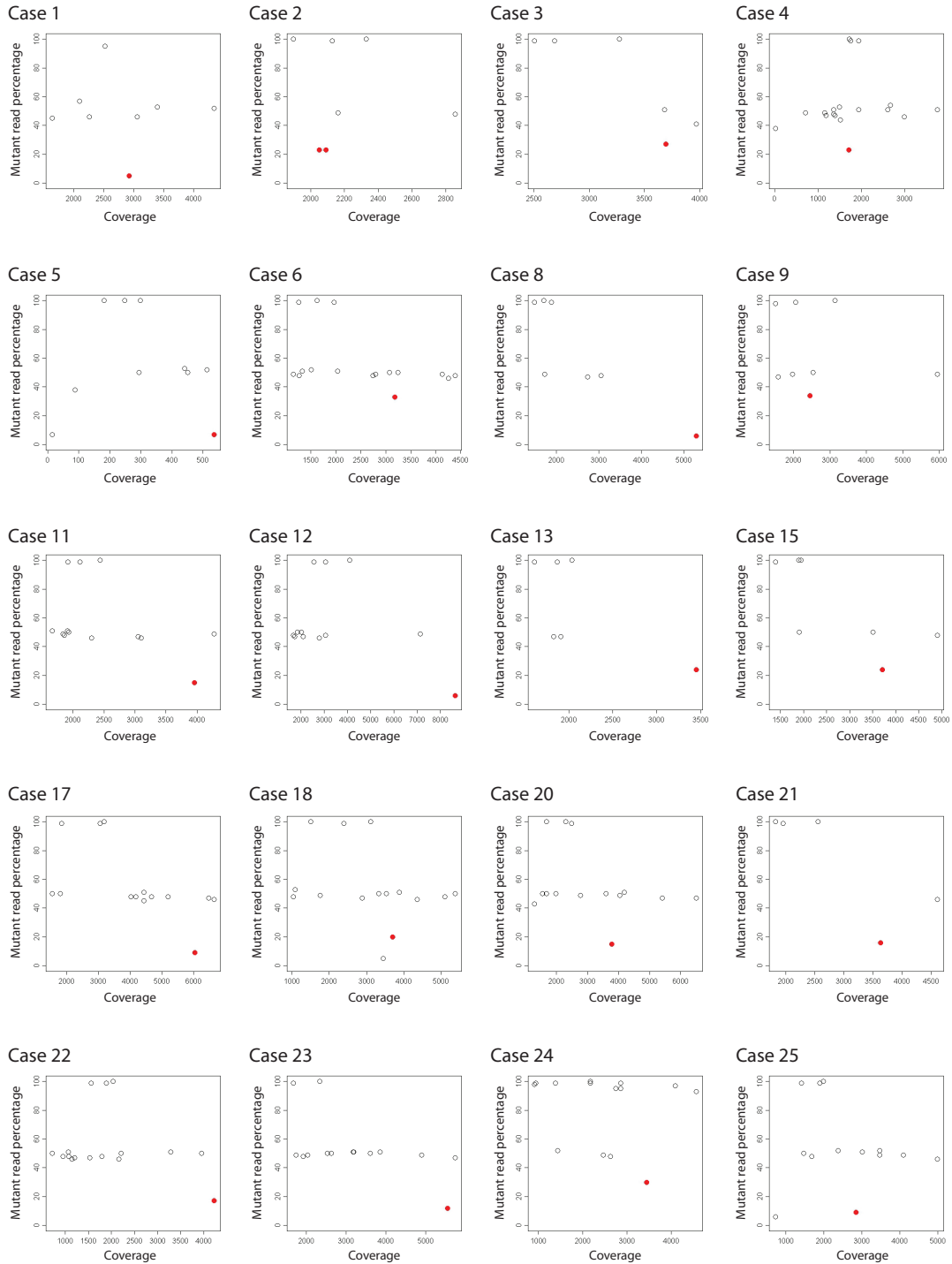






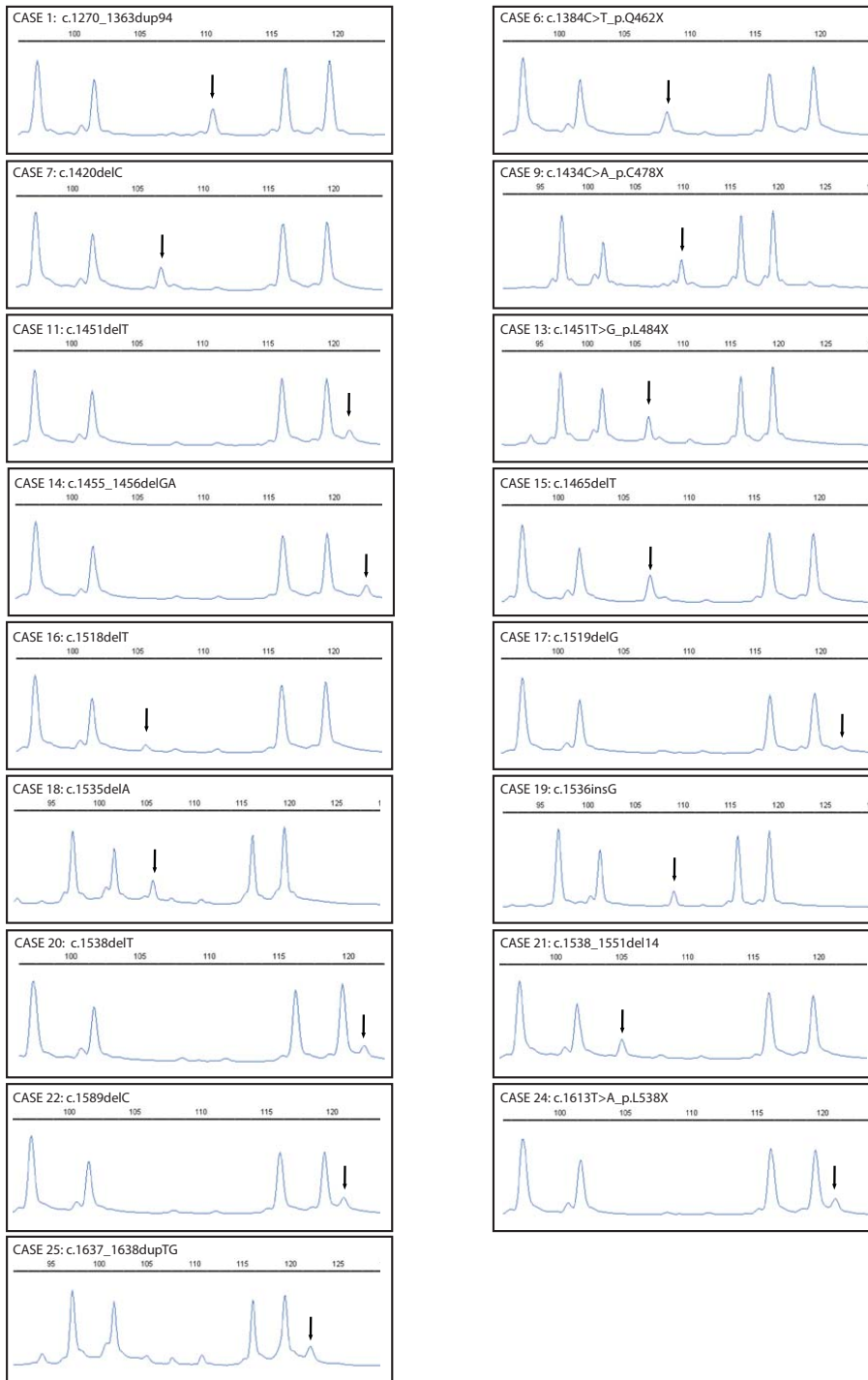
The mutant allele is lower than typical of heterozygous mutations, consistent with mosaicism.  
 \*indicates that the reverse sequencing trace is presented.

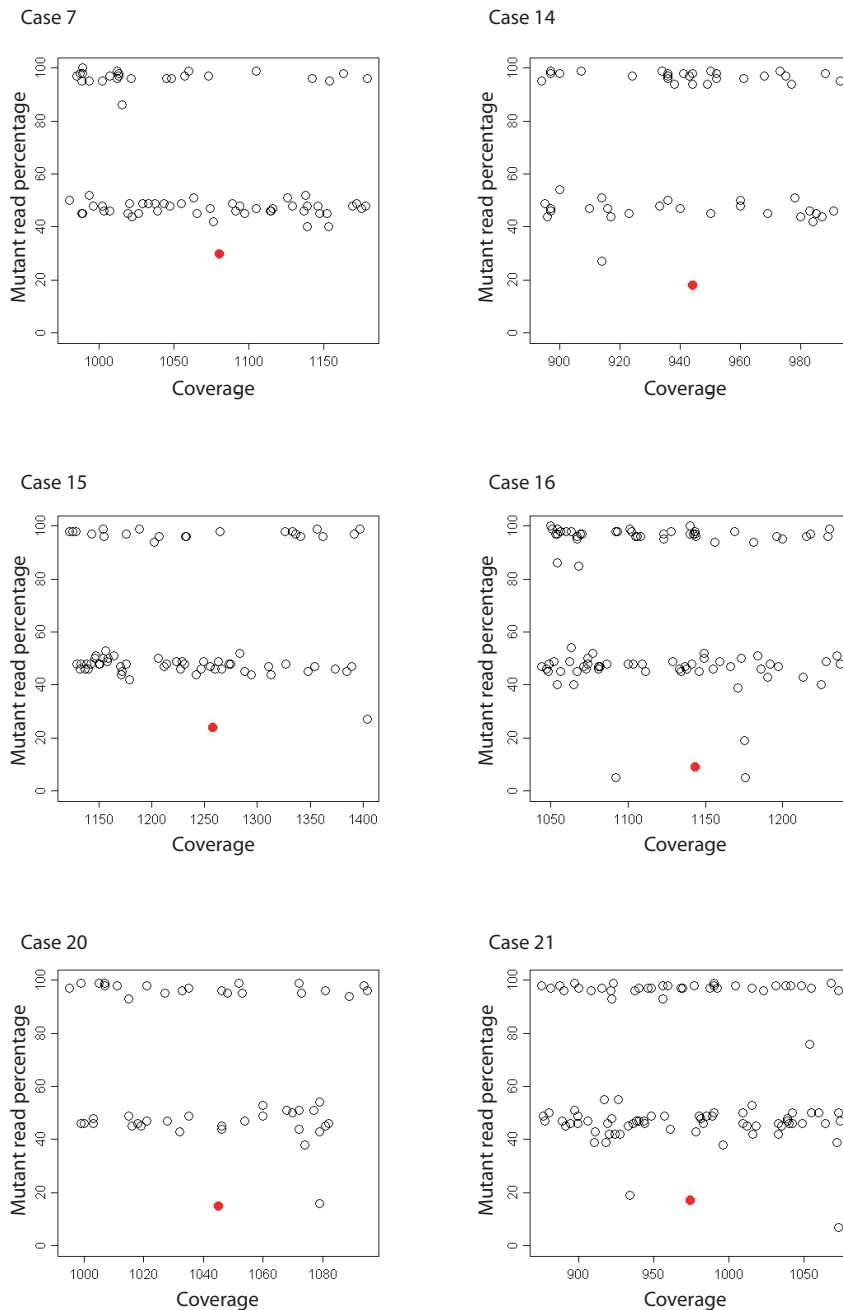
**Supplementary Figure 4.** Deep PCR amplicon sequencing of *BRCA1*, *BRCA2* and *PPM1D* cluster region showing mosaic mutations.



Mutant read percentage is calculated as the proportion of reads containing the variant. The red dot indicates the *PPM1D* mutation. In case 2, the complex indel was called as two different mutations and thus two red dots. Variants were censored at 5%. All mutations have a consistently lower mutant read percentage, indicating mosaicism. Open dots represent variants in *BRCA1* or *BRCA2*.

**Supplementary Figure 5.** MLPA profiles showing *PPM1D* mutations.



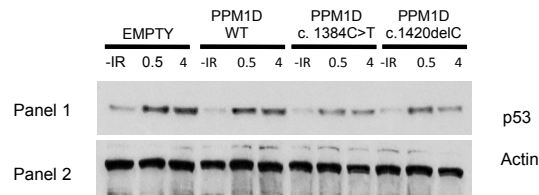
**Supplementary Figure 6.** DNA repair panel individual sequencing in six *PPM1D* mutation carriers.

Mutant read percentage was calculated as in Supplementary Figure 4. Read coverage was recorded at all variant sites, counting only bases within reads with a mapping quality of at least 20 and a base quality of at least 22. A window around the mutation containing at least 50 variants with similar coverage was identified. The red dot indicates the *PPM1D* mutation. Open dots represent other variants in the custom pulldown which were not validated. All *PPM1D* mutations were consistently lower, indicating mosaicism. Mutant read percentages for cases 15, 20 and 21 matched those in Supplementary Figure 4.

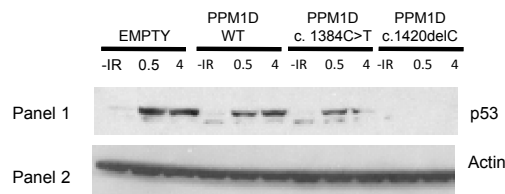


**Supplementary Figure 7: The effect of mutant PPM1D isoforms on p53 activation**

### 293T



### Hela

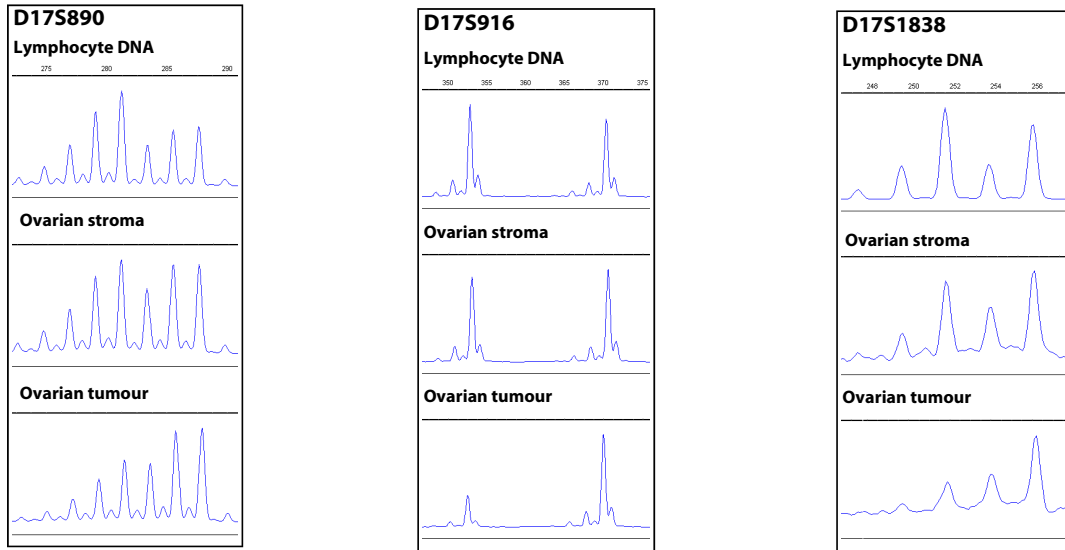


p53 wild type HeLa and HEK293 cells were transfected with *PPM1D* cDNA expression constructs and exposed to ionising irradiation (5 Grays). At 30 minute and four hour intervals after IR exposure whole cell lysates were generated and western blotted to estimate the IR induced activation of p53. Western blots showing p53 and actin (loading control) protein levels at different times (in hours) after IR exposure are shown. 'Empty' represents cells transfected with an empty expression construct, 'PPM1D WT' represents cells transfected with a wild type *PPM1D* cDNA expression construct and 'PPM1D c.1384C>T' and 'PPM1D c.1420delC' represent cells transfected with mutant *PPM1D* cDNA constructs. The suppression of p53 was enhanced in cells transfected with the mutant constructs suggesting these alleles encode hyperactive PPM1D isoforms.

**Supplementary Figure 8. Tumour haplotype analysis, Sanger sequencing and MLPA analysis**

**Case 11 c.1451delT**

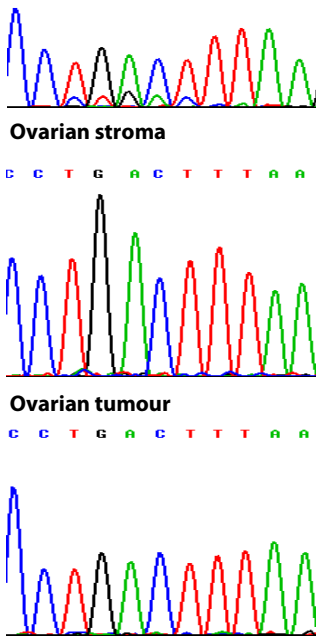
**Microsatellite intensities**



**Sanger sequencing chromatograms**

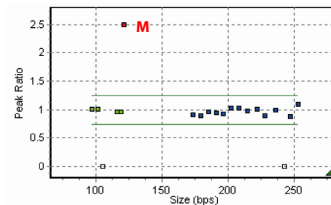
**Lymphocyte DNA**

C C T G A C T T T A A

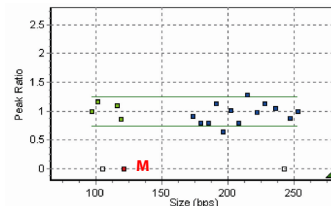


**MLPA dosage plots**

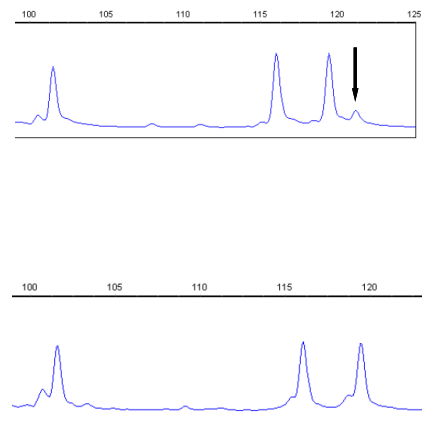
**Lymphocyte DNA**



**Ovarian tumour**

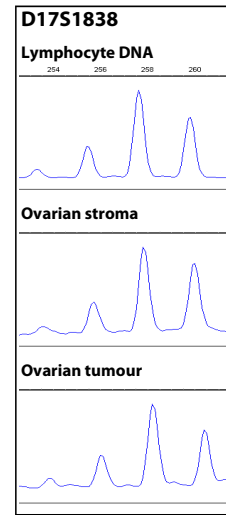
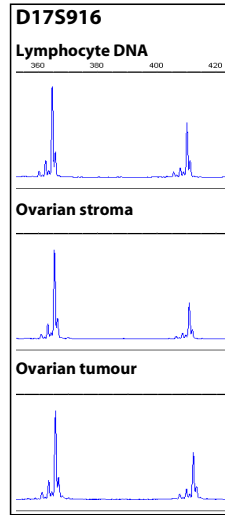
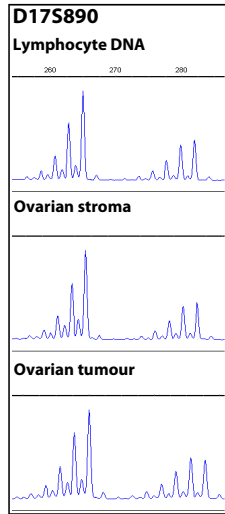


**MLPA intensities**



**Case 15: c.1465delT**

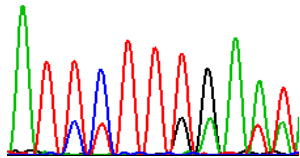
**Microsatellite intensities**



**Sanger sequencing chromatograms**

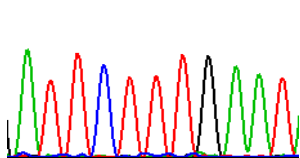
**Lymphocyte DNA**

A T T C T T T G A A T



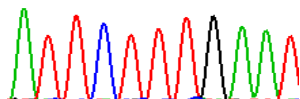
**Ovarian stroma**

A T T C T T T G A A T



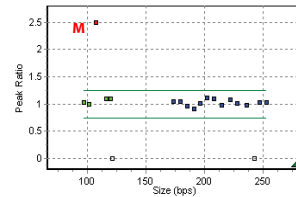
**Ovarian tumour**

A T T C T T T G A A T

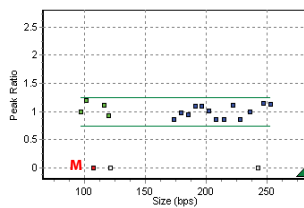


**MLPA dosage plots**

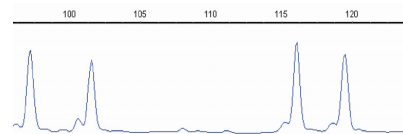
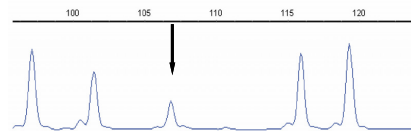
**Lymphocyte DNA**



**Ovarian tumour**

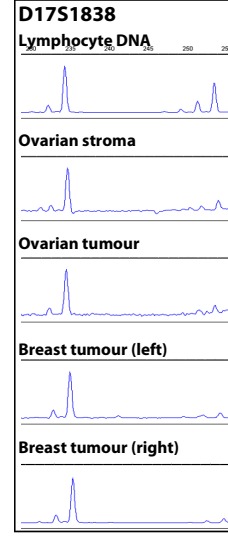
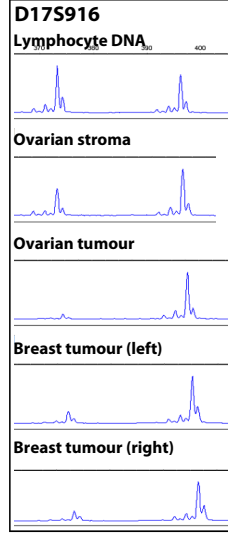
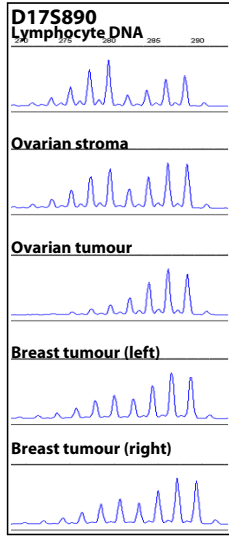


**MLPA intensities**

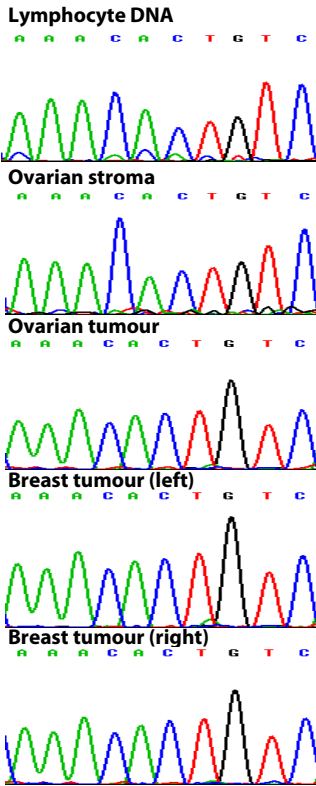


**Case 17: c.1519delG**

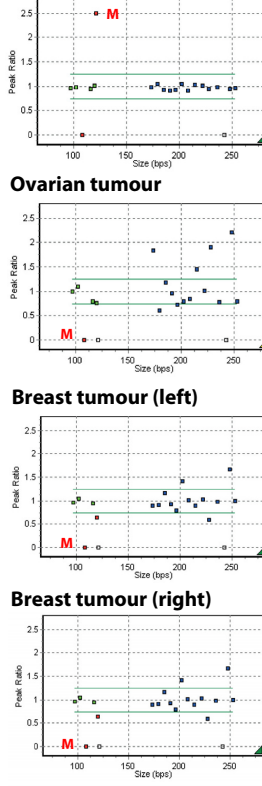
**Microsatellite intensities**



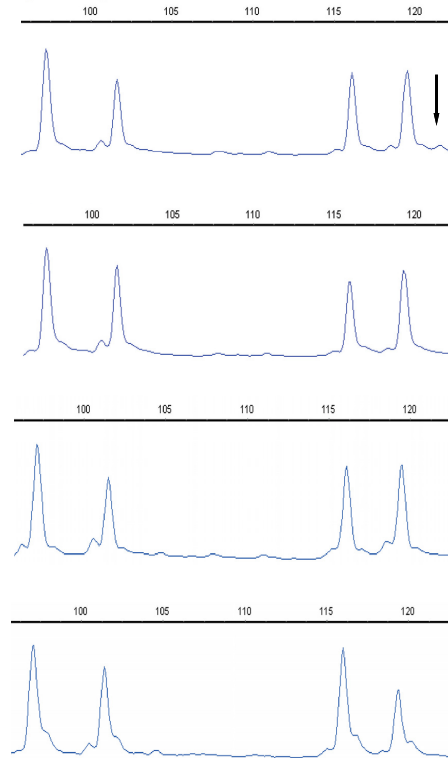
**Sanger sequencing chromatograms**



**MLPA dosage plots**

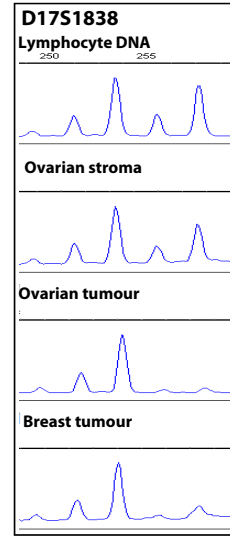
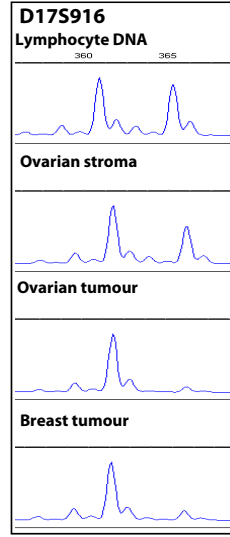
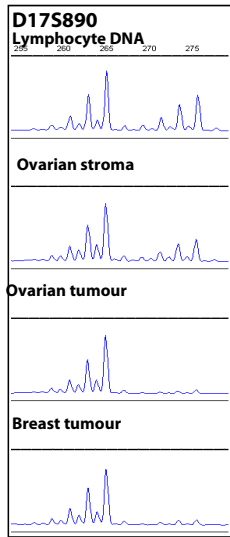


**MLPA intensities**

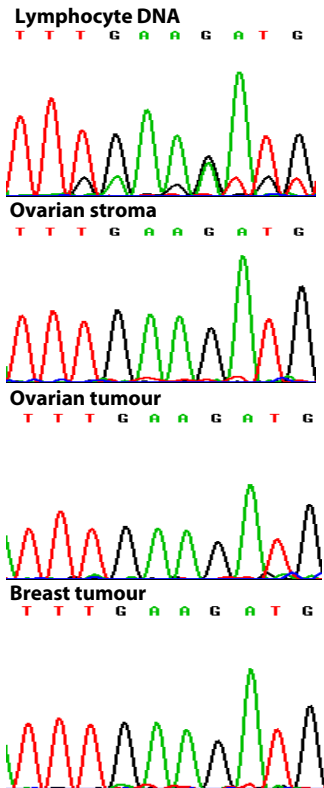


Case 20: c.1538delT

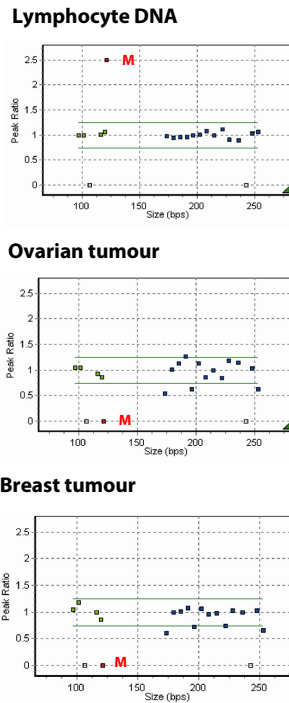
Microsatellite intensities



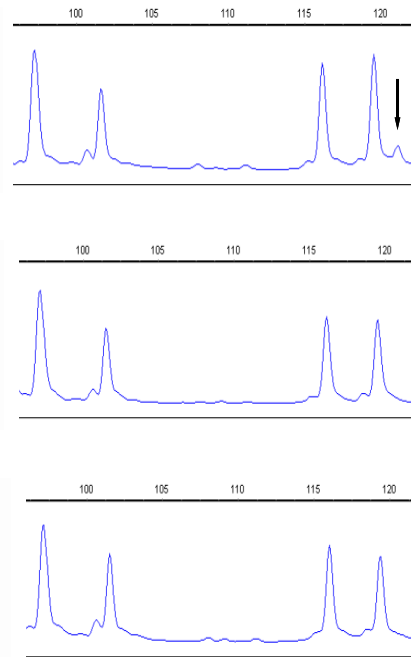
Sanger sequencing chromatograms



MLPA dosage plots

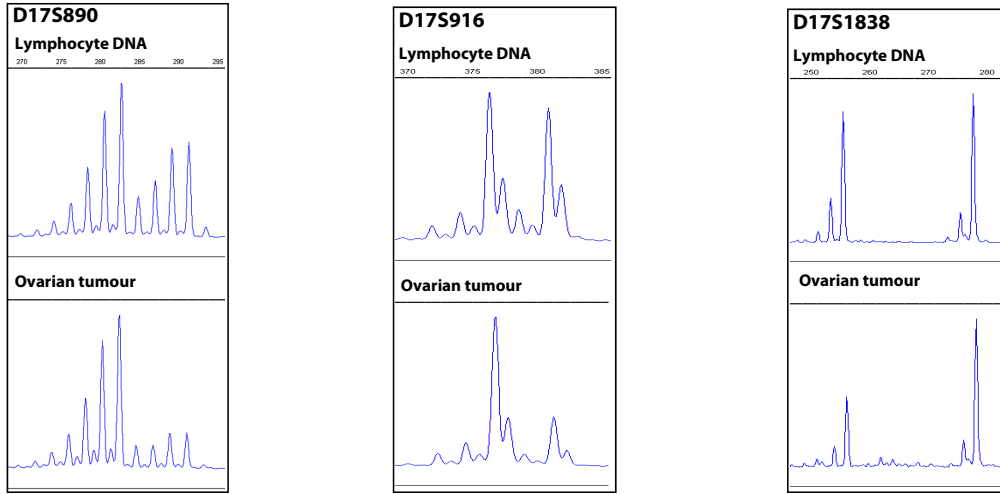


MLPA intensities



**Case 1 c.1270\_1363dup94**

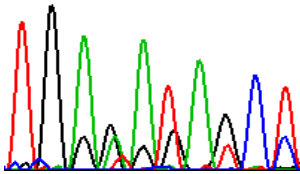
**Microsatellite intensities**



**Sanger sequencing chromatograms**

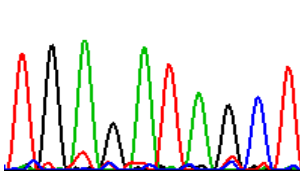
**Lymphocyte DNA**

T G A G A T A G C T



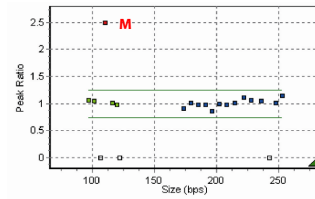
**Ovarian tumour**

T G A G A T A G C T

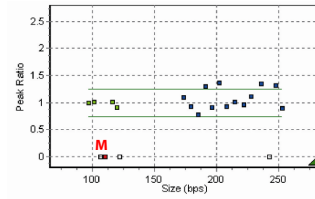


**MLPA dosage plots**

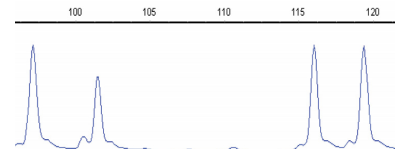
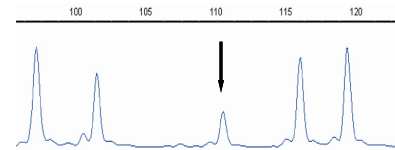
**Lymphocyte DNA**



**Ovarian tumour**



**MLPA intensities**



Microsatellite data demonstrates two alleles in DNA from lymphocytes and stroma and loss of heterozygosity in tumours in cases 1, 11, 17 and 20. There is no loss of heterozygosity in the tumour DNA from case 15. The Sanger sequencing and MLPA data demonstrate that the mosaic mutations in lymphocyte DNA are not detectable in stromal or tumour DNA. The data from deep PCR amplicon sequencing of tumour and stromal DNA is presented in Supplementary Table 5.

## The Wellcome Trust Case Control Consortium

The following individuals are part of the Wellcome Trust Case Control Consortium+

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Hocking<sup>33</sup>, Chris Holmes<sup>5</sup>, Eleanor Howard<sup>1</sup>, Philip Howard<sup>34</sup>, Joanna MM Howson<sup>17</sup>, Debbie Hughes<sup>16</sup>, Sarah Hunt<sup>1</sup>, John D Isaacs<sup>35</sup>, Mahim Jain<sup>18</sup>, Derek P Jewell<sup>36</sup>, Toby Johnson<sup>34</sup>, Jennifer D Jolley<sup>3,4</sup>, Ian R Jones<sup>29</sup>, Lisa A Jones<sup>19</sup>, George Kirov<sup>29</sup>, Cordelia F Langford<sup>1</sup>, Hana Lango-Allen<sup>2</sup>, G Mark Lathrop<sup>37</sup>, James Lee<sup>12</sup>, Kate L Lee<sup>34</sup>, Charlie Lees<sup>21</sup>, Kevin Lewis<sup>1</sup>, Cecilia M Lindgren<sup>8,18</sup>, Meeta Maisuria-Armer<sup>17</sup>, Julian Maller<sup>18</sup>, John Mansfield<sup>38</sup>, Jonathan L Marchini<sup>5</sup>, Paul Martin<sup>7</sup>, Dunecan CO Massey<sup>12</sup>, Wendy L McArdle<sup>39</sup>, Peter McGuffin<sup>14</sup>, Kirsten E McLay<sup>1</sup>, Gil McVean<sup>5,18</sup>, Alex Mentzer<sup>40</sup>, Michael L Mimmack<sup>1</sup>, Ann E 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