As of July 1st 2017, our laboratory will be updating variant annotations to be consistent with HGVS notation. In HGVS notation, the +1 coding position refers to the first Methionine.

These changes will be implemented for the following genes:

- **RB1**: Reports will include the coding DNA sequence (c.) number based on NM_000321.2 instead of the genomic coordinate (g.) from Genbank accession number L11910.
- **F9**: Reports will include the coding DNA sequence (c.) number based on NM_000133.3 instead of the genomic coordinate (g.).
- **F8**: Reports will include a new amino acid numbering system to reflect HGVS notation (+1 position refers to the first Methionine). F8 has a proprotein which is 19 amino acids shorter on the 5’ end; our legacy numbering excluded these 19 amino acids. The mRNA transcript is NM_000132.3.
- **ENG**: Our reports will no longer be referring to exons 9A and 9B; these exons will now be referred to as exons 9 and 10. All subsequent exon numbers after 10 will be increased by 1, for a total of 15 exons. There will be no change in coding DNA sequence or amino acid numbering which is based on transcript NM_001114753.2.

### Summary

<table>
<thead>
<tr>
<th>Test</th>
<th>Gene</th>
<th>Change</th>
</tr>
</thead>
<tbody>
<tr>
<td>Retinoblastoma</td>
<td>RB1</td>
<td>Coding DNA sequence (c.) number will be reported instead of genomic coordinate (g.)</td>
</tr>
<tr>
<td>Hemophilia</td>
<td>F8</td>
<td>Amino acids will be shifted by 19 residues</td>
</tr>
<tr>
<td></td>
<td>F9</td>
<td>Coding DNA sequence (c.) number will be reported instead of genomic coordinate (g.)</td>
</tr>
<tr>
<td>HHT</td>
<td>ENG</td>
<td>Exons 9A and 9B will now be referred to as exons 9 and 10. Subsequent exon numbers after 10 will be increased by 1.</td>
</tr>
</tbody>
</table>