**A** COL12A1 expression at rs594012 and rs240736 in OA hip cartilage

- **Major allele**
- **Minor allele**

<table>
<thead>
<tr>
<th>Genotype at rs9350591</th>
</tr>
</thead>
<tbody>
<tr>
<td>CC n = 9 CT n = 2</td>
</tr>
</tbody>
</table>

**B** COL12A1 expression at rs594012 and rs240736 in OA knee cartilage

- **Major allele**
- **Minor allele**

<table>
<thead>
<tr>
<th>Genotype at rs9350591</th>
</tr>
</thead>
<tbody>
<tr>
<td>CC n = 32 CT n = 4</td>
</tr>
</tbody>
</table>

**C** COL12A1 expression at rs594012 and rs240736 in NOF cartilage

- **Major allele**
- **Minor allele**

<table>
<thead>
<tr>
<th>Genotype at rs9350591</th>
</tr>
</thead>
<tbody>
<tr>
<td>CC n = 4 CT n = 1</td>
</tr>
</tbody>
</table>

**D** TMEM30A expression at rs41269315 in OA hip cartilage

- **Major allele**
- **Minor allele**

<table>
<thead>
<tr>
<th>Genotype at rs9350591</th>
</tr>
</thead>
<tbody>
<tr>
<td>CC n = 3 CT n = 4</td>
</tr>
</tbody>
</table>

**E** TMEM30A expression at rs41269315 in OA knee cartilage

- **Major allele**
- **Minor allele**

<table>
<thead>
<tr>
<th>Genotype at rs9350591</th>
</tr>
</thead>
<tbody>
<tr>
<td>CC n = 3 CT n = 6</td>
</tr>
</tbody>
</table>

**F** TMEM30A expression at rs41269315 in NOF cartilage

- **Major allele**
- **Minor allele**

<table>
<thead>
<tr>
<th>Genotype at rs9350591</th>
</tr>
</thead>
<tbody>
<tr>
<td>CC n = 1 CT n = 1</td>
</tr>
</tbody>
</table>
**MYO6 expression at rs1045758 and rs699186 in OA hip cartilage**

- **G**: Allelic expression imbalance
  - Major allele
  - Minor allele
  - Genotype at rs9350591
  - n = 6
  - n = 4

  *p* = 0.019

**MYO6 expression at rs1045758 and rs699186 in OA knee cartilage**

- **H**: Allelic expression imbalance
  - Major allele
  - Minor allele
  - Genotype at rs9350591
  - n = 16
  - n = 9

  *p* = 0.224

**MYO6 expression at rs1045758 and rs699186 in NOF cartilage**

- **I**: Allelic expression imbalance
  - Major allele
  - Minor allele
  - Genotype at rs9350591
  - n = 4
  - n = 0

**SENP6 expression at rs71561434 and rs17414687 in OA hip cartilage**

- **J**: Allelic expression imbalance
  - Major allele
  - Minor allele
  - Genotype at rs9350591
  - n = 11
  - n = 1

  *p* = 0.939

**SENP6 expression at rs71561434 and rs17414687 in OA knee cartilage**

- **K**: Allelic expression imbalance
  - Major allele
  - Minor allele
  - Genotype at rs9350591
  - n = 9
  - n = 11

**SENP6 expression at rs71561434 and rs17414687 in NOF cartilage**

- **L**: Allelic expression imbalance
  - Major allele
  - Minor allele
  - Genotype at rs9350591
  - n = 11
  - n = 0

**Additional file 11. Allelic expression imbalance analysis stratified by disease state and joint site.**

The data were stratified by OA hip, OA knee and NOF for COL12A1 (A, B and C), TMEM30A (D, E and F), MYO6 (G, H and I), and SENP6 (J, K and L). Stratification did not result in any changes to the analysis of combined data. Statistical significance was assessed using the Mann-Whitney U test and is not corrected for multiple testing.