Allele specific expression on liver and head kidney of *Salmo salar* with differential susceptibility to the challenge with *Piscirickettsia salmonis*.

Dettleff, P. & Martínez, V.
Santiago de Compostela, 2015
ISGA XII
Cis and Trans regulation

- **Trans-regulatory** elements regulate the expression of distant genes (e.g. transcription factors).

- **Cis-regulatory** elements are regions of DNA which regulate the transcription in the same gene or nearby genes (e.g. enhancers and promoters).

- **Cis-acting** regulatory variation can play a role in phenotypic variation.
Allele specific expression

- The allele specific expression (ASE) determines the existence of imbalances in the expression of one allele relative to the other.

Approach to study the impact of genetic variation on gene expression.

- The ASE is affected by factors that act in cis.

Cis-acting factors

Expressed coding SNPs

Allele specific expression

Individual, allowing exclude differences due to external factors.
Piscirickettsia salmonis

- The principal bacterial agent that affect the Atlantic salmon Chilean production.

- Produces a systemic aggressive infection that involves several organs, including head, kidney, and liver.

- It has been observed a variable level of resistance to this bacteria in commercial and experimental populations.

- Little is known about the resistance to this disease from a molecular point of view.
SRS challenge

Head kidney and liver samples

Resistant families

Susceptibles families
De novo assembly
ASE detection

- Mapping
- SNPs detection
- SNPs validation in set B of samples
- ASE analysis
- Unambiguously mapped reads counts
- Heterozygotes SNPs
Assembly results

<table>
<thead>
<tr>
<th>Metric</th>
<th>Value</th>
</tr>
</thead>
<tbody>
<tr>
<td>N50</td>
<td>1,711</td>
</tr>
<tr>
<td>N° of nucleotides</td>
<td>50,867,482</td>
</tr>
<tr>
<td>Contigs &gt; 1000 bp</td>
<td>17,294</td>
</tr>
<tr>
<td>N° of annotated contigs</td>
<td>16,614</td>
</tr>
<tr>
<td>GO terms</td>
<td>81,791</td>
</tr>
</tbody>
</table>

Top-Hits Species distribution

- Salmo salar
- Danio rerio
- Oreochromis niloticus
- Lepisosteus oculatus
- Neolamprologus brichardi
- Maylandia zebra
- Xiphophorus maculatus
- Haplochromis burtoni
- Others

BLAST Top-Hits (%)
<table>
<thead>
<tr>
<th>KEGG enzymatic annotation</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>N° sequences with enzymatic annotation</td>
<td>4,173</td>
</tr>
<tr>
<td>Metabolic pathways</td>
<td>126</td>
</tr>
</tbody>
</table>

![Pathways with greater number on enzymes](#)
 Immune response
 Antigen processing and presentation
 Cell death
 Response to stress
- Activation of immune response
- Cytokine production

<table>
<thead>
<tr>
<th>GO category</th>
<th>Terms with differences</th>
</tr>
</thead>
<tbody>
<tr>
<td>Molecular Function</td>
<td>12</td>
</tr>
<tr>
<td>Biological Process</td>
<td>19</td>
</tr>
<tr>
<td>Cellular Component</td>
<td>3</td>
</tr>
</tbody>
</table>
SNPs

<table>
<thead>
<tr>
<th>Type</th>
<th>N° of SNPs</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td>Transition</td>
<td></td>
<td></td>
</tr>
<tr>
<td>A/G</td>
<td>969</td>
<td>64</td>
</tr>
<tr>
<td>C/T</td>
<td>940</td>
<td>64</td>
</tr>
<tr>
<td>A/T</td>
<td>253</td>
<td></td>
</tr>
<tr>
<td>Transversion</td>
<td></td>
<td></td>
</tr>
<tr>
<td>C/G</td>
<td>218</td>
<td>36</td>
</tr>
<tr>
<td>A/C</td>
<td>305</td>
<td></td>
</tr>
<tr>
<td>G/T</td>
<td>291</td>
<td></td>
</tr>
</tbody>
</table>

SNPs identified in head kidney and liver

2,976 SNPs validated in set B

Validation of SNPs in set B of samples (n=20)
ASE

173 SNPs with ASE in all samples
GO-distribution of genes with ASE in all samples (Biological process, level 3)
- Ribosomal proteins
- Complement factors
- Hemoglobin subunits
Conclusions

- The results showed that common ASE genes in all fish are mostly connected with different metabolic processes.

- The ASE genes by group include protein metabolic processes, with several ribosomal proteins and presenting a low number of immune genes.

- Complement factors and hemoglobin subunits could be interesting genes involved in survival to the disease.

- Further studies are required to understand if these cis-acting factors could affect disease resistance in practice.
Acknowledgments

CONICYT
Ministerio de Educación
Gobierno de Chile

FONDECYT
Fondo Nacional de Desarrollo Científico y Tecnológico

Scholarship to PhD studies.

Project number 11020608.

FAVET
inbiogen

DOCTORADO EN CIENCIAS SILVOAGROPECUARIAS Y VETERINARIAS

Travel bursary.