



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.



### Piscirickettsia salmonis

- The principal bacterial agent that affect the Atlantic salmon Chilean production.
- Produces a systemic aggressive infection that involves several organs, including lead kidney and liver.

79%

- It has been observed ria bacteria in commercial and ex
- Little is known abou molecular point of vie

- l of resistance to this tal populations.
  - o this disease from a

### SRS challenge



## De novo assembly



### **ASE detection**



### Assembly results

Species

N50	1,711	
N° of nucleotides	50,867,482	
Contigs>1000 bp	17,294	
N° of annotated contigs	16,614	
GO terms	81,791	

Salmo salar Danio rerio **Oreochromis niloticus** Lepisosteus oculatus Neolamprologus brichardi Maylandia zebra Xiphophorus maculatus Haplochromis burtoni Others 0 5 15 20 25 10 30 **BLAST Top-Hits (%)** 

#### **Top-Hits Species distribution**

### **KEGG enzymatic annotation**

N° sequences with enzymatic annotation	4,173
Metabolic pathways	126

### Pathways with greater number on enzymes

Purine metabolism



GO category	Terms with differences	
Molecular Function	19	
<b>Biological Process</b>	86	
Cellular Component	34	



- ≻Immune response
- >Antigen processing and presentation
- ≻Cell death
- ≻Response to stress



Terms with differences		



≻Activation of immune response ≻Cytokine production



**Biological Process** 

# **SNPs**

	SI h	NPs identified in lead kidney and		
	Туре	liver N° of SNPs	N° of SNPs	%
Transition	A/G	969	1909	64
	С/Т	Validation of SNPs in set B of	1303	
Transversion	A/T	samples <b>25-3</b> 20)		
	C/G	218	1067	36
	A/C	305		
	G/T <sub>v</sub>	alidated in set B		
	A/C G/T <sub>v</sub>	305 2,976 SNPs alidated m set B		

ASE





GO-distribution of genes with ASE in all samples (Biological process, level 3)





### 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.

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