TARGETED ANALYSES OF A CANDIDATE GENOMIC REGION DETERMINING THE LACK OF PIGMENTATION IN GILTHEAD SEABREAM (Sparus aurata)

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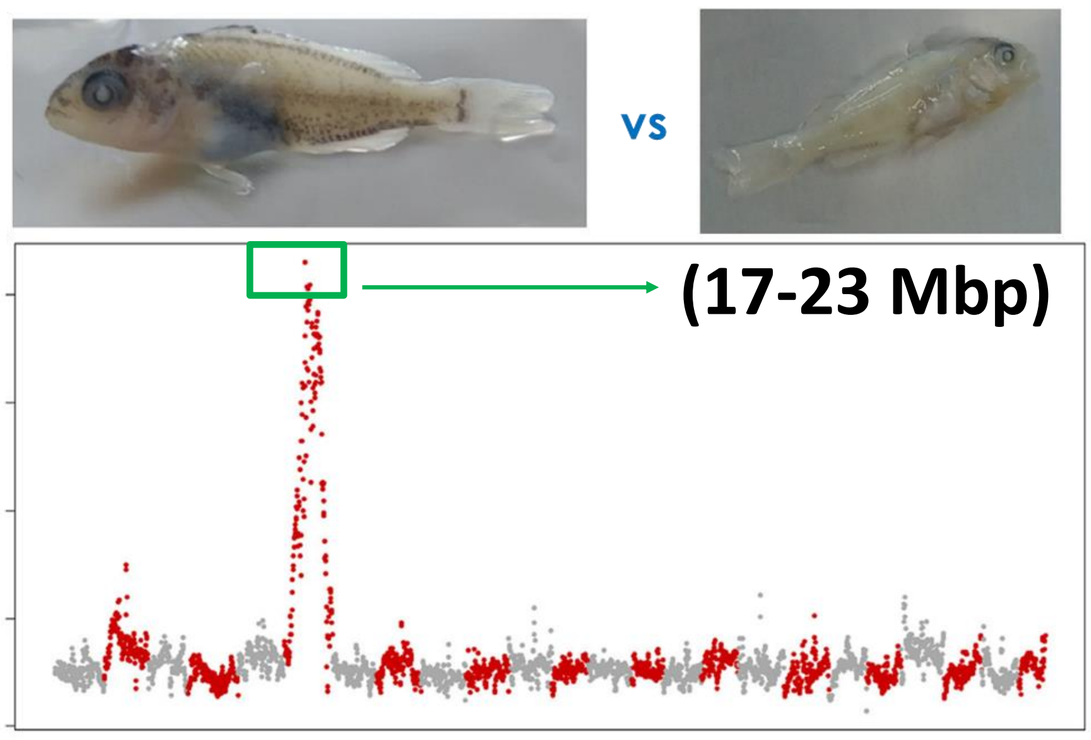
1 - Introduction

- High rates of phenotypic abnormalities in gilthead seabream during early development cause significant economic losses
- Increased deformities in inbred populations suggest a genetic component is involved
- While skeletal deformities are well-studied, pigmentation discolourations also affect consumer acceptance
- Fish pigmentation is controlled by various chromatophores, including melanophores, xanthophores, erythrophores, and iridophores
- **The aim of the study was to characterize a genomic region linked with lack of pigmentation**

2 – Materials and Methods

Genome scan of normal (n.30) vs depigmented (n. 20) fry derived from the same broodstock nucleus detected a region on chromosome 6



1) Retrieve all SNPs and INDEL included in the region
2) Detect genes included in the most comprehensive list of pigmentation-related genes so far available (https://www.ifpcs.org/colorgenes/
3) Perform variant effect predictor of the variants near those genes

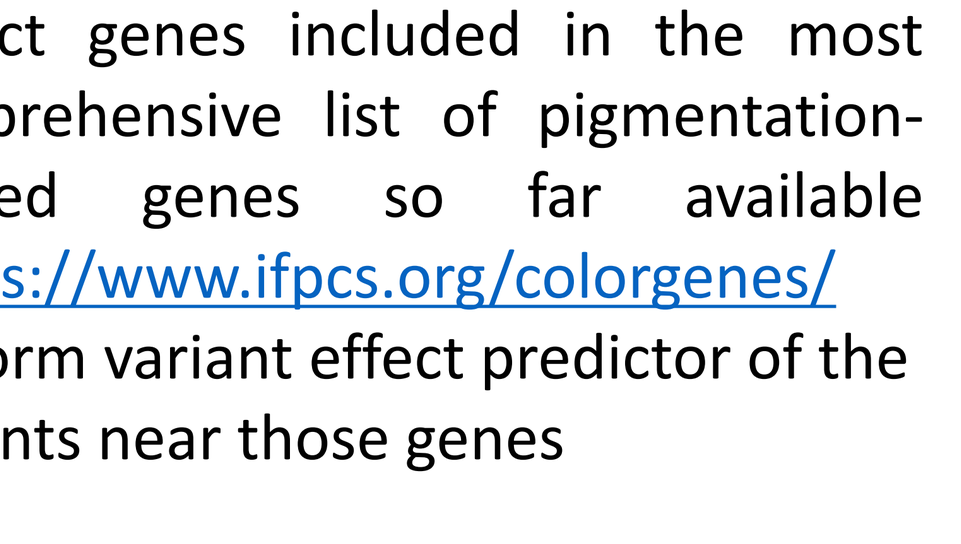
3 – Results

The analyses identified :

1) 4,810 SNPs and 541 INDELs
2) Six genes involved in pigmentation (**dstyk, erbb3, fancd2, parp3, rab7, slc2a11**)

The slc2a11 gene has been linked to the absence of xanthophores in zebrafish and medaka

3) Most of the variants are linked to intronic and upstream/downstream gene variants, with no SNPs or INDELs identified as having significant impacts



4 – Conclusions

The causative mutation(s) may be linked to regulatory regions rather than functional changes in protein structure. Further analyses of these genes are necessary