

# General Introduction

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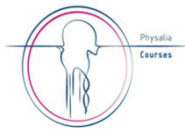


OscarGenomics



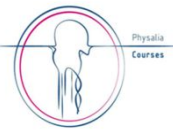
# Christian - in a slide

- University of Gießen (*BSc Agricultural Science*)
- University of Gießen (*MSc Crop Production*)
- University of Gießen (*PhD Plant Breeding*)
- Roslin Institute, University of Edinburgh (*Post-doc in QG*)
- Excellence in Breeding, CGIAR / CIMMYT (*QG Scientist*)



# Oscar- in a slide

- **Agricultural Engineer** (*Madrid*)
- **Animal Science** (*MSc degree, Madrid*)
- **Animal Genetics & Quantitative Genetics** (*UPM-Madrid & UW-Madison*)
- **University of Wisconsin-Madison** (*Post-doc*)
- **Dept. Environment and Primary Industries-Melbourne, Australia** (*Senior Research Scientist*)
- **INIA - Madrid** (*Senior Research Scientist*)
- **The Roslin Institute - Edinburgh** (*Full Professor*)



# Filippo - in a slide

- Roma (*born*)
- Perugia (*MSc degree*)
- Cork, ICBF (*Web-design & Database*)
- Cremona, ANAFI (*Quantitative Genetics*)
- Guelph, CGIL (*Visiting Scientist*)
- Wageningen, WUR (*PhD*)
- Göttingen University (*post-doctoral researcher*)
- Lodi, PTP (*'omics in animals, plants, humans*)
- Milan - CNR (*tenured researcher*)
- Cardiff University (*biostatistician*)
- Milan - CNR (*senior researcher*)
- Bruxelles - ERC (*seconded national expert*)
- Milan - CNR (*director of research*)





# to keep your appetite - a success story

From GWAS to CRISPR-based therapy for sickle-cell disease & beta-thalassemia ([Frangoul et al. 2021](#))

GWAS for HbF (fetal haemoglobin) & disease severity → target gene: **BCL11A**

→ subsequent molecular characterization of the gene (transcription factor that represses  $\gamma$ -globin expression and fetal hemoglobin in erythroid cells)

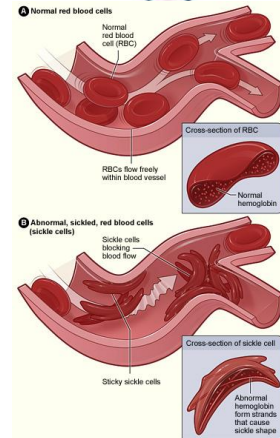
## References:

[BCL11A and F-cell production](#): (Menzel et al 2007)

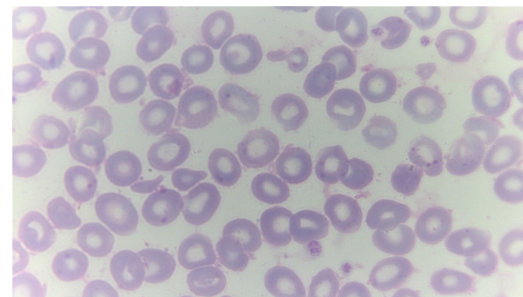
[BCL11A associated with Hbf and beta-thalassemia severity](#) (Uda et al 2008)

[BCL11A associated with HbF and sickle cell disease](#) (Lettre et al 2008)

[BCL11A and HbF expression](#) (Sankaran et al 2008)



[https://en.wikipedia.org/wiki/Sickle\\_cell\\_disease](https://en.wikipedia.org/wiki/Sickle_cell_disease)



[https://en.wikipedia.org/wiki/Beta\\_thalassemia](https://en.wikipedia.org/wiki/Beta_thalassemia)

# now you – round of introduction

- who
- where from
- type of research / work
- what's your interest on GWAS / motivation



# course - in a slide

the course [website](#):

- Introduction to GWAS
- Linear regression, linkage disequilibrium, population structure, ...
- Experimental design, sample size and statistical power
- Data exploration and preprocessing
- Imputation of missing genotypes
- Basic GWAS models (in R)
- Linux and the Shell
- Scaling up: building a pipeline for GWAS
- Collaborative exercise
- post-GWAS analysis
- GWAS model extensions

Flexibility!



# laying out the **topic**

- [https://filippob.github.io/introduction\\_to\\_gwas/](https://filippob.github.io/introduction_to_gwas/)
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## introduction\_to\_gwas

[https://filippob.github.io/introduction\\_to\\_gwas/](https://filippob.github.io/introduction_to_gwas/)

[View the Project on GitHub](#)  
filippob/introduction\_to\_gwas

## introduction\_to\_gwas

**Material for the course “Introduction to genome-wide association studies (GWAS)”**

Instructors: *Filippo Biscarini, Oscar Gonzalez-Recio, Christian Werner*

This course will introduce students, researchers and professionals to the steps needed to build an analysis pipeline for Genome-Wide Association Studies (GWAS). The course will describe all the necessary steps involved in a typical GWAS study, which will then be used to build a reusable and reproducible bioinformatics pipeline.

Each day the course will start at **14:00** and end at **20:00** (CET). As a general rule, we'll have a longer break (30 minutes) at 16:00 and two shorter breaks (10-15 minutes) later on during the day (to be decided flexibly depending on the sessions).

