

Groups and project topics:

Functional genetic study of:	Group	Students, teachers
Anosmia in Kallman syndrome	1	
Adenomatous polyposis coli (APC)	2	
Situs inversus in Kartagener's syndrome	3	
Hypohidrotic ectodermal dysplasia (HED)	4	
Muscle atrophy or hypertrophy	5	
Scoliosis	6	
Amyotrophic lateral sclerosis (ALS)	7	
Achondroplasia	8	

The length of the final research plan should be 2000 words.

The research plan must include (in the following order):

I Title, the names of authors and the number of the group.

II Abstract (ca 100 words)

- o Short summary of the research plan
- o Graphical abstract

III Introduction (ca 300 words) (300-400)

- o Motivation of the study: description of the phenotype of the disorder, and its implications to understanding biology and health.
- o Which molecular mechanisms or gene/regulatory pathway is the disorder associated with? (there can be multiple). For example, you can discuss the genetic linkage, the gene product and the cellular functions of the protein.
- o Is the affected gene and its functions conserved? Describe and compare the functions of your candidate gene in vertebrate and invertebrate organisms.

IV Planned research (ca. 1000 words) (1000-1300)

1. Define your hypothesis: Formulate the research question. In your research plan, focus on one candidate gene or pathway. For example, the research question can be:

- o How does the mutation affect your gene/protein function?
- o What is the role of your gene of interest in normal development (in the context of the disease)?
- o What are the mechanism by which the linked gene/pathway manifests the disorder?
- o What are potential additional genes and pathways associated with the disorder?

2. Design experiments to answer your research questions. Plan 1-2 experiments that address the research question, taking advantage of the models and methods discussed on the course in experiment design.

- o Describe the experiment – focus on the principle, you do not need to describe the method in great detail
- o What kind of information do you expect to produce with the experiment?
- o What kind of controls are needed?

V Discussion and impact (ca. 300 words) (300-500)

- o Discuss the strengths and weaknesses of your experiments
- o What kind of studies would be needed to further confirm your results?
- o Summarize how the proposed experiments would benefit understanding of disease mechanisms, developmental processes, or genetic pathways

VI Summary of the individual contributions of the authors (not included in word count)

VII Reference list (not included in word count)

- The figure legends and text on the figures are not included in the word count. However figure legends must be concise.

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Schedule and tasks

The research plan is prepared as independent work in pairs, mentored by a teacher. In the first week of the course, each pair will select a topic and the gene/genetic pathway that will be studied in the project.

Please arrange your feedback meetings with your teacher, as outlined below.

1. First meeting (12.1.):

- Choose the main research topic: gene(s) or pathway(s) associated with the disease.
- Think of possible hypotheses and discuss them with the teacher.
- Find one article to work through in addition to the reference(s) provided by teachers.

Independent work before the next meeting:

Students:

- Summarise the article and find additional references.
- When summarising the literature, explain how it is related to the study topic (the disease) and your research hypothesis. What is known about the disease mechanism, and what is unknown?
- Think of possible experiments to study the disease mechanism. Use methods taught in course program: either in lectures or lab practicals.
- Formulate the hypothesis/research question
- Write a one-page (600-800 words) outline of your project and send it to your teacher. The outline must include: the genetic pathway associated with the disease, the gene mutation or cellular process of study and the aims of the project (research hypothesis).

Teachers:

- Read the students' project draft.
- Find one strength and one area of development you would like to discuss about the research plan.
- If necessary, find additional literature on the research topic to recommend to students.

2. Second meeting (week 6)

- Exchange feedback on the writing task until the second meeting.
- Define the research hypothesis.
- Discuss the experiments addressing the research aims with the teacher. How do the experiments test the hypothesis?

Independent work before the next meeting

Students:

In your written research plan:

- Describe your experiments: the samples, controls, measurements, and expected results.
- Describe the possible interpretations of the expected results.
- Explain how the experiment addresses the research question.
- Include a graphical abstract.

Send the research plan text to the teacher. The research plan should include all required sections (I-VII).

Teachers:

- Read the students' research plan.
- Assess the research aims, described experiments and the description of the expected results.
- List the main strengths and weaknesses of the research plan (2-5 strengths and weaknesses).
- For the points that need development, explain why the approach would not work and think of the ways it could be improved.

3. Third meeting (week 7)

- Exchange feedback on the writing task.
- Discuss the key strengths and any important weaknesses of the presented research plan.
- Discuss the advantages of and disadvantages of your approach.
- Discuss alternative approaches for the study.

Student presentations

Prepare a 30-minute presentation of your research plan.

Present your project to the rest of the course on 13.2.2025.

After the presentations:

Students finalise the project based on the given feedback. Submit the final research plan in Moodle by 28.2.

Assessment

Project presentation and the final project text will be assessed. Each written project is evaluated by two teachers. Assessment guidelines for teachers are found in Moodle.