Student Project Works Q&A#1

Course Evaluation:

Students attending the course

- Class participation and attendance +

Student project work and presentation (60%)

- Written exam (40%)

Students non attending the course

- Written exam (100%)

The learning objectives of the course on Molecular and Cellular Physiology for the Master Degree in Genetics and Molecular Biology are:

1. Understand the mechanisms underlying the comunication of neuronal and glial cells in the homeostatic control and in the pathogenesys of brain diseases

2. Develop the ability to critically analyze and interpret primary literature in molecular and cellular physiology, and to effectively communicate scientific findings through written and oral presentations.

3. Apply molecular and cellular physiology concepts to design and execute a research project, and to develop independent research skills, such as experimental design, data analysis, and interpretation.

Project Work Overview

•Goals of the Project Work

The goal of the project work for the students is to apply the knowledge and skills they have gained during the course to investigate and analyze a specific topic related to molecular and cellular physiology.

The project work is designed to enhance the students' critical thinking, problem-solving, and communication skills, as well as their ability to work independently and collaboratively.

The ultimate goal is to produce a high-quality student project work that demonstrates a thorough understanding of the chosen topic and showcases the students' ability to apply their knowledge to real-world situations.

Student Project Work: Video Podcast

For your final group project, you will produce a video podcast (12–15 minutes) focusing on a rare genetic disorder or a neurodegenerative disease.

Objectives

Your podcast should demonstrate your ability to integrate clinical, molecular, and cellular knowledge with experimental and translational perspectives, while also reflecting on the broader social impact of these diseases.

Task

In groups of 4 students,
you will design and record a video podcast.

Each group member will take on a role:

- **Interviewer (moderator)**

- **Patient or family member (personal/social perspective)**

- **Clinician (clinical and diagnostic perspective)**

- **Researcher (molecular, cellular, and therapeutic perspective)**

Content Guidelines

Your podcast must include:

- **Clinical Phenotype** – Describe the main signs and symptoms.
- **Molecular & Cellular Biology** – Explain the underlying mechanisms.
- **Key Experiments** – Present the discoveries that clarified disease mechanisms, and propose additional experiments you would perform.
- **Therapeutic Strategies** – Summarize current treatments and discuss potential future approaches (drug development, gene therapy, repurposing).
- **Social Impact** – Discuss the burden on patients, families, and society.

- Slides (3–5 max) should accompany the video to illustrate the key points.

Disease List (1)

Each group will be assigned one of the following:

Neurodevelopmental Disorders

- Fragile X Syndrome
- Smith-Magenis Syndrome
- Rett Syndrome
- DiGeorge Syndrome (22q11.2 Deletion)
- Dravet Syndrome
- SYNGAP1-related Disorder
- Kabuki Syndrome
- Angelman Syndrome
- Brugada Syndrome

Disease List (2)

Neurodegenerative Disorders

- Frontotemporal Dementia (FTD)
- Amyotrophic Lateral Sclerosis (ALS)
- Huntington's Disease (HD)
- Parkinson's Disease
- CANVAS (Cerebellar Ataxia, Neuropathy, Vestibular Areflexia Syndrome)
- Early onset Alzheimer's Disease (EOAD)
- Cystic Fibrosis

Timeline

- Groups formed in Week 0
- Check-in/Q&A in Week 3 and Week 6
- Final presentation (January)
- Your video podcast will be screened in class
- Short Q&A session will follow

Evaluation

- Scientific accuracy and depth (clinical + molecular)
- Clarity and creativity in communication
- Integration of experimental evidence and therapeutic reasoning
- Quality of the slides and video production
- Teamwork and role distribution

Generic Video Podcast Script (Any Rare/Neurodegenerative Disease)

Duration: ~12–15 minutes
Roles: Interviewer, Patient/Family member, Clinician, Researcher
Slides: 3–5 to illustrate key points (phenotype, molecular pathway, experiments, therapies, social impact)

Visual support, not full text





0:00–1:00 – Introduction (Interviewer)

Welcome the audience.

- **Introduce the disease (“Today we will talk about X, a rare disease affecting the nervous/cardiac/muscular system…”).**
- **Present the guests: a patient/family member, a clinician, and a researcher.**

Slide 1: Title of the disease + symbolic image (e.g. brain, DNA, heart)

1:00–3:00 – Patient/Family Perspective

Share the clinical experience:

- First symptoms noticed (e.g. seizures, movement problems, developmental delays).
- Daily challenges (school, work, autonomy, therapies).
- Emotional and social impact on the family.

Goal: humanize the disease and highlight its real-life burden.

3:00–6:00 – Clinician Perspective

Systematic clinical description:

- **Key symptoms and progression.**
- **Typical diagnostic process (EEG, MRI, genetic testing, blood biomarkers).**
- **Explain how diagnosis is confirmed and differential diagnoses to consider.**
- **Discuss current management and treatments (symptomatic, supportive, experimental).**

Slide 2: Table or diagram of the main clinical features.

6:00–10:00 – Researcher Perspective

Molecular & Cellular Biology:

The gene(s) involved, protein function, cellular pathway.

How mutations affect the pathway → phenotype.

Key experiments that revealed mechanisms:

Animal models, iPSC-derived cells, imaging, electrophysiology, omics.

Future experiments:

What should be done to clarify unanswered questions?

Slide 3: Pathway diagram or schematic of the cellular mechanism..

10:00–12:00 – Therapies & Future Directions

Current therapies (if any): drugs, gene therapy, symptomatic treatments.

Limitations of existing approaches.

Potential strategies under investigation:

- Drug repurposing
- Gene editing / CRISPR
- Antisense oligonucleotides

Novel biomarkers for early diagnosis

Slide 4: Table “Current vs. Potential Therapies.”

12:00–15:00 – Social Impact & Conclusion

Patient/family: what is the biggest unmet need (awareness, treatment, support)?

Clinician: challenges in healthcare systems for this disease.

Researcher: why translational science and collaborations are crucial.

Final note by interviewer:

“This disease teaches us how biology, medicine, and society must work together to bring solutions.”

Slide 5: Awareness image (rare disease ribbon, brain-heart-DNA icon).

Important:

Even though each student will play a specific character in the podcast, all four members must work together on every aspect of the content preparation.

This means that each of you should contribute to:

- researching the clinical phenotype,
- understanding the molecular and cellular biology,
- identifying key experiments,
- discussing therapies,
- reflecting on social impact.

The roles are performative only, to make the podcast engaging. The scientific preparation is a shared responsibility.