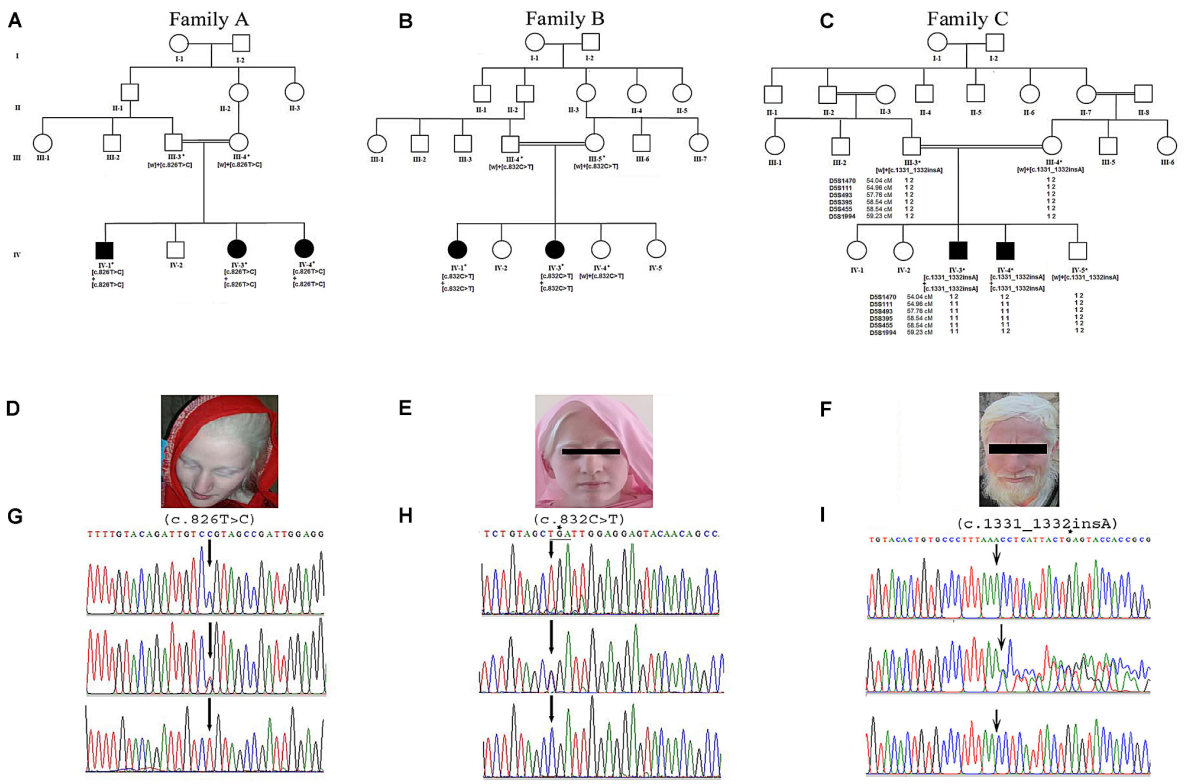
Toolboxes for SuperFastLearning digital contents in STEM

WANT TO LEARN ABOUT GENETIC DIAGNOSIS FROM A TO Z ?



PROBLEM-BASED LEARNING - UNIVERSITY OF BORDEAUX - December 2021
4TTV305U Genetics of Physiological and Pathological Processes :
International Bachelor of STEM, 2nd Year
Coordinator : Pr. Javerzat

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The complex problem :

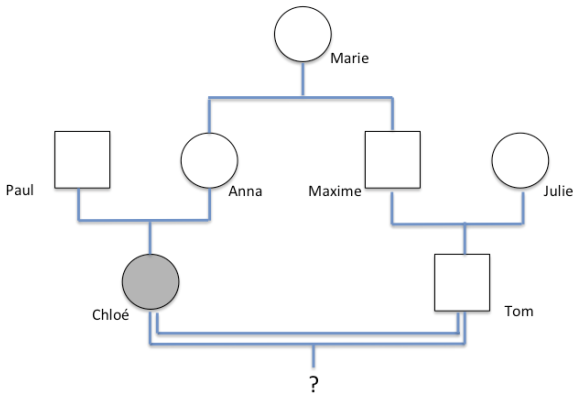
Patients and non-specialist healthcare professionals are increasingly being expected to understand and interpret the results of genetic testing. The reporting of these results is currently done using a wide variety of templates containing different amounts, levels and layouts of information.

Recent guidelines for template design recommend that genetic reports should include two separate sections to be provided independently to 1. the health-care geneticist that referred the patient, 2. the patient and his/her family and family doctor. In each section, genetic and medical information should be carefully delivered taking into account the likely understanding of the recipients.

The medical genetics department of Bordeaux is the national reference center for genetic diagnosis of albinism. They have recently identified a new class of genotype named "R402Q-OCA1" that requires creating a specific report template for patients with such genotype.

The trainee in charge of developing the "R402Q-OCA1" report template should have a thorough understanding of the functional, transmission and population genetics relevant to "classical-OCA1" and "R402Q-OCA1" patients. This knowledge should enable him/her to develop the diagnostic reporting template for clinicians directly in charge of "R402Q-OCA1" patients (section 1, professional section). The information contained in this section should be as accurate as possible for optimal medical follow-up and genetic counselling.

Following the recommendations of editorial guidelines, the trainee should then carefully work on the specific wording, numbers and graphics that might be used within the template to communicate the results and their implications clearly to the patient, his/her family and family doctor (section 2, patient-friendly section). This report format should allow "R402Q-OCA1" patients to understand detailed information about their unique genetic make-up, its consequences for their lives and risks for carriers in the family.



The medical genetics department of Bordeaux provides all necessary information for the project to be completed in due time including the genetics data for the family of Chloé P., a fictional "R402Q-OCA1" patient.

Organisation of this sequence

Phases and steps

Phase	Duration	Steps	Explanations
First phase **In group** **In class** **Dec 2nd 2021, 5-7 pm (total duration 2 hours)**	**10 min**	**Organise the group**	Divide up the roles (see page 6): Depending on the number of students per group, you may take one major and one minor function, eg. "time keeper & facilitator" "secretary & reviewer"
	20 min	**Discover and rephrase the problem**	**Read the document provided** Individual work: everyone does an initial overview of the booklet to familiarise themselves with its contents. **Understand and clarify the problem** What exactly is the problem we are going to address? The scribe starts to note down what appears in the exchanges (keywords, concepts, ideas, etc.).
	30 min	**Design pathway**	Establish a list of relevant questions. Take stock of what the team knows (and does not know) based on both the group and individual backgrounds. If necessary, draw up a list of restrictions to limit the scope of the problem (if necessary, check with the tutor). Draw up a list of expected outputs. Consider different ways of dealing with the task. The activator launches and relaunches the discussion when necessary.
	20 min	**Define the knowledge needed**	**Clarify the learning outcomes** What do we need to (re-)learn / discover to deal with the problem? What questions will each of us need to be able to answer? What will we need to be able to do?
15 min	**Define a working plan**		Determine what information needs to be gathered to confirm or invalidate your first thoughts. List the tasks to be done and deliverables to be prepared by everyone before the next session. The secretary notes what is decided and arranges to communicate it to the other team members.

Phase	Duration	Steps	Explanations
Second Phase **Individually** **At home**	**Dec 2nd to Dec 16th** **12 hours**	**Implement the action plan**	Each student in the group has to work on every step that have been defined in the action plan. You will collect and analyse information in order to be able to explain and solve the problem. **1- Individual home work (estimated time 6 hours)** Each student provides the necessary work defined during the 1st phase (exploitation of resources, research on identified key points, data manipulation). It is highly recommended that communication between students is kept to a minimum during this phase. This will ensure fruitful sharing during the final phase. **2- Preparation of deliverables (estimated time 4 hours)** Each student prepares a detailed report of the results of his or her work to be shared during the final phase. As the individual report should be synthetic, graphics and bullet points are highly encouraged as well as a "one slide per task" format.

Phase	Duration	Steps	Explanations
Final Phase **In group** **In class** **Dec 16th**	**45 min**	**Determine the role of each partner**	Quickly check the roles of each student in the group, make sure that the essential roles are provided. You can change roles if needed.
		Share everyone's production	Each student presents his scientific findings and individual attempt at designing the genetic testing report template. The scribe notes any discrepancies between the individual reports. Common answers and findings are summarized, discrepancies are discussed and a consensus agreed. A vote for the best answer to each scientific question and best template can be organized.
	45 min	**Prepare the deliverable**	The final collective output consists of: - as many slides as initial individual scientific findings (maximum 5). - one slide (animations allowed) for the section 1, professional section of the R402Q-OCA1 reporting template, specifically completed for Chloé's clinician - one slide (animations allowed) for the section 2, patient-friendly section of the R402Q-OCA1 reporting template, specifically completed for Chloé and husband Tom.
	30 min	**Assess**	You will assess alone the group work. Compare all together your answers. Then assess individually your learning outcomes and your work.

What is your role in the group ?

	The driver ensures the progress of the group work. They make sure each step is realised. They alert when a task is forgotten and should find collective solutions when required.
	The time keeper is responsible for the schedule of the collective work. They ensure an optimal use of the time available.
	The scribe writes all the points that have been discussed, whatever they are and all the positions expressed during the meeting(s). They write them on a blackboard, paperboard or any other writing surface that can be viewed by each group member. .
	The facilitator's role is key to ensure that everyone can express their opinion freely, and to carry out the goal of the meeting. They can help to develop a common understanding and to create a pleasant working climate.
	The secretary is in charge of synthesising the main ideas, hypotheses, decisions taken and working results of the group. They are in charge of communicating these data to the group members.
	The speaker talks about the process, work status and the results of the group to the teacher, the tutor, the whole class or to any external person.
	The reviewer is in charge of the progress points. They are reporting what has been done and what remains to be done.
	The observer is looking at the group functioning. They are reporting their observations to improve the group functioning.

Assessment:

Evaluation of the group work

You will assess individually the group work: on a scale from 1 to 5:

Assessment	Totally no	no	yes	Totally yes
About the group's outputs				
Did the group follow each step of PBL sequence?				
Did the group acquire the intended learning outcomes?				
Did the group produce what was asked?				
About the group's organisation				
Were the group climate and interactions favourable for efficiency?				
Could everyone express themselves?				
How could be improve the group's work?				
Did everyone keep their role during the phase?				
Self-assessment				
How qualitative and efficient were my interactions with the group?				
How qualitative and efficient was my production?				
About the complex problem				
Was the topic interesting?				
About the relation with the tutor				
Did the tutor guide the group effectively, by asking the good questions to redirect when needed?				

Evaluation of the problem-based learning sequence

Each student auto-evaluates his learning by answering a questionnaire focused on the Intended Learning Outcomes: (scale 0-nothing 5-a lot)

Example :

1- How much did you know before the lesson about genetic testing?
0 1 2 3 4 5

2- How much do you know now about genetic testing?
0 1 2 3 4 5

Evaluation of the individual work

Please give your individual slides to be graded by the teacher

Evaluation of the group deliverable

Vote for the 5 best slides to create your final deliverable. They will be graded by your teacher.