



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.

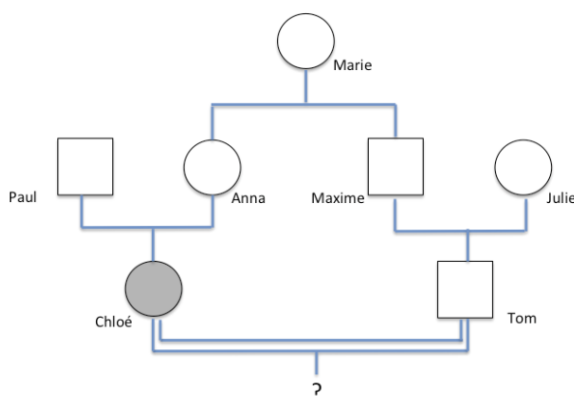


Recent guidelines for template design recommend that genetic reports should include two separate sections to be provided independently to 1. the health-care geneticist that referred the patient, 2. the patient and his/her family and family doctor. In each section, genetic and medical information should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

Phase	Duration	Steps	Explanations
First phase In group In class Dec 2nd 2021, 5–7 pm (total duration 2 hours) 15 min	10 min	Organise the group	Together, define the role of each group member (see page 6): Depending on the number of students per group, you may take one major <u>and</u> one minor function, eg. “time keeper & facilitator” “secretary & reviewer”
	20 min	Discover and rephrase the problem	Read the document provided /Understand and clarify the problem After reading the context alone, define the keywords and questions that may arise regarding these keywords in a group. What exactly is the problem you have to solve? What are you being asked to do?
	30 min	Design a pathway	Define which elements and concepts need to be explained, propose a list of relevant questions, identify a pathway to reach the solution, and define a list of expected deliverables.
	20 min	Define the knowledge needed	Clarify the learning outcomes Clearly define what is known and what is unknown. What data is missing to solve the problem? What are the intended learning outcomes? What deliverables do you need to produce in this case?
	15 min	Define a working plan	Which steps do each of you need to follow in order to collect the information needed (research, experiments, interviews...)?

Phase	Duration	Steps	Explanations
Second Phase Individually At home	Dec 2nd to Dec 16 th 12 hours	Implement the action plan	Each student in the group has to work on every step that has been defined in the action plan. You must collect and analyse information to explain and solve the problem.











			<p>1- Individual home work (estimated time 6 hours)</p> <p>Work defined during the 1st phase (exploitation of resources, research on identified key points, data manipulation).</p> <p>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.</p> <p>2- Preparation of deliverables (estimated time 4 hours)</p> <p>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.</p>
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Phase	Duration	Steps	Explanations
Final Phase In group In class Dec 16 th	45 min	Determine the role of each partner	If necessary, you can change some roles
		Share everyone's production	<p>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.</p> <p>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.</p>
	45 min	Prepare the deliverable	<p>The final collective output consists of:</p> <ul style="list-style-type: none"> - 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 assess the group work alone. Compare your answers together. Then assess your learning outcomes and your work individually.



What is your role in the group?

	<p>The driver guides the progress of the group work. They ensure that each step is completed.</p> <p>They put out an alert if a task is forgotten, and need to find collective solutions when required.</p>
	<p>The timekeeper is responsible for keeping to the collective work schedule. They ensure optimal use of the time available.</p>
	<p>The scribe writes down 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 surface that can be viewed by all the group members.</p>
	<p>The facilitator's role is key to ensuring that everyone can express their opinion freely and to meet the goal of the meeting. They can help to develop a common understanding and to create a pleasant work atmosphere.</p>
	<p>The secretary is responsible for summarising the main ideas, hypotheses, decisions taken, and working outcomes of the group.</p> <p>They are in charge of communicating these data to all the group members.</p>
	<p>The speaker reports the process, work status, and the group's results to the teacher, the tutor, the whole class, or to any external person necessary.</p>
	<p>The reviewer is in charge of the progress points. They report what has been done and what remains to be done.</p>
	<p>The observer looks at how the group functions. They report their observations to improve the way the group operates.</p>



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 the PBL sequence?				
Did the group achieve the intended learning outcomes?				
Did the group produce what was asked?				
About the group's organisation				
Did the group atmosphere and interactions facilitate efficiency?				
Was everyone able to express themselves?				
Could the group's work be improved?				
Did everyone stick to their role during the different phases?				
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 relationship with the tutor				
Did the tutor guide the group effectively by asking the right questions to refocus 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.