European Commission
Directorate-General for Health and Food Safety (DG SANTE)

ITHACA

ITHACA Board Meeting

2020 December 10 – 12

Satellite Meetings : Friday Dec. 11th

WG 8 Teaching & Training

From 11 to 12:50 AM

European Reference Networks

(WG8) Work Groups / Satellite Meetings : Friday December 11th

Time Slot (Virtual Zoom Rooms)	W8 : Teaching & Training Chair : Laurence Faivre /Jill Clayton Smith	SPEAKER PM contact : Anne Hugon
11h00	EJP Genetic	Laurence Faivre
11h20	Master of genetic CIBERER course	Pablo Lapunzina
11h30	Syndromic Games & "Vite Extra-ordinarie"	Giuseppe Zampino
11h40	Discussion : webinars & other productions	All Attendees
12h00	Updates on Serious Game	Anne Hugon
12h20	YGN in Europe	Florence Riccardi
12h20 - 12h50	Future objectives for Y 4-5	All attendees

EJP-RD - Online course (WP16) - MOOC Diagnosis

- Developed in the context of EJP RD WP16
  - Online academic course
  - Series of 5 MOOCs (Diagnosis, Innovative therapies, Translational research, Clinical trials methodologies + additional topic)
  - Coordinated by French Foundation for Rare Diseases

- 1st MOOC on Diagnosis of rare (genetic) diseases
  - Scientific coordination
    - ERN ITHACA (Prof Laurence Faivre)
    - ERN GENTURIS (Dr Chrystelle COLAS)

  - Targets
    - PhD Students (medicine/research) & Post-Doc
    - Academic researchers, PAO representatives, others

EJP-RD - Online course (WP16) - MOOC Diagnosis

- +/- 15 hour online course (3 hours/week)
- Free course, no pre-requisite
- Academic credits will be explored after 1st session(s) run
  - Based on audience reached
  - Based on Universities, Academic organisations, Other organisations interest
- Creative Commons license

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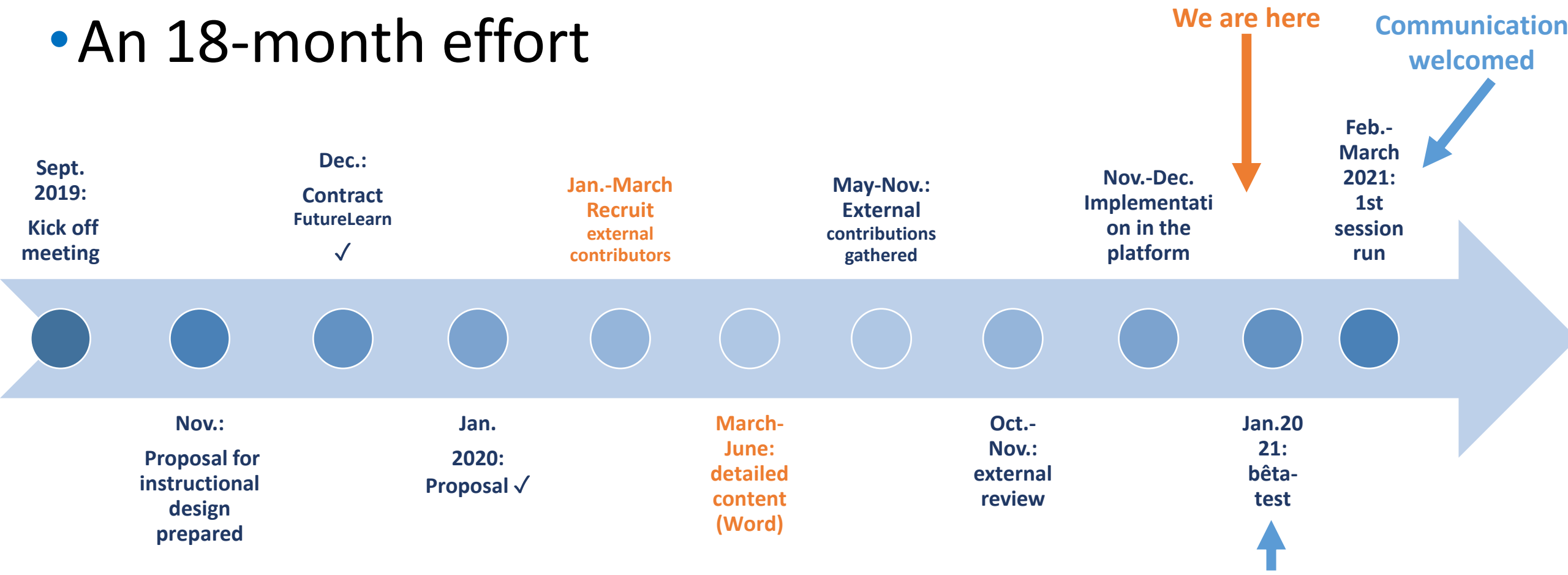
EJP-RD - Online course (WP16) - MOOC Diagnosis

- An 18-month effort



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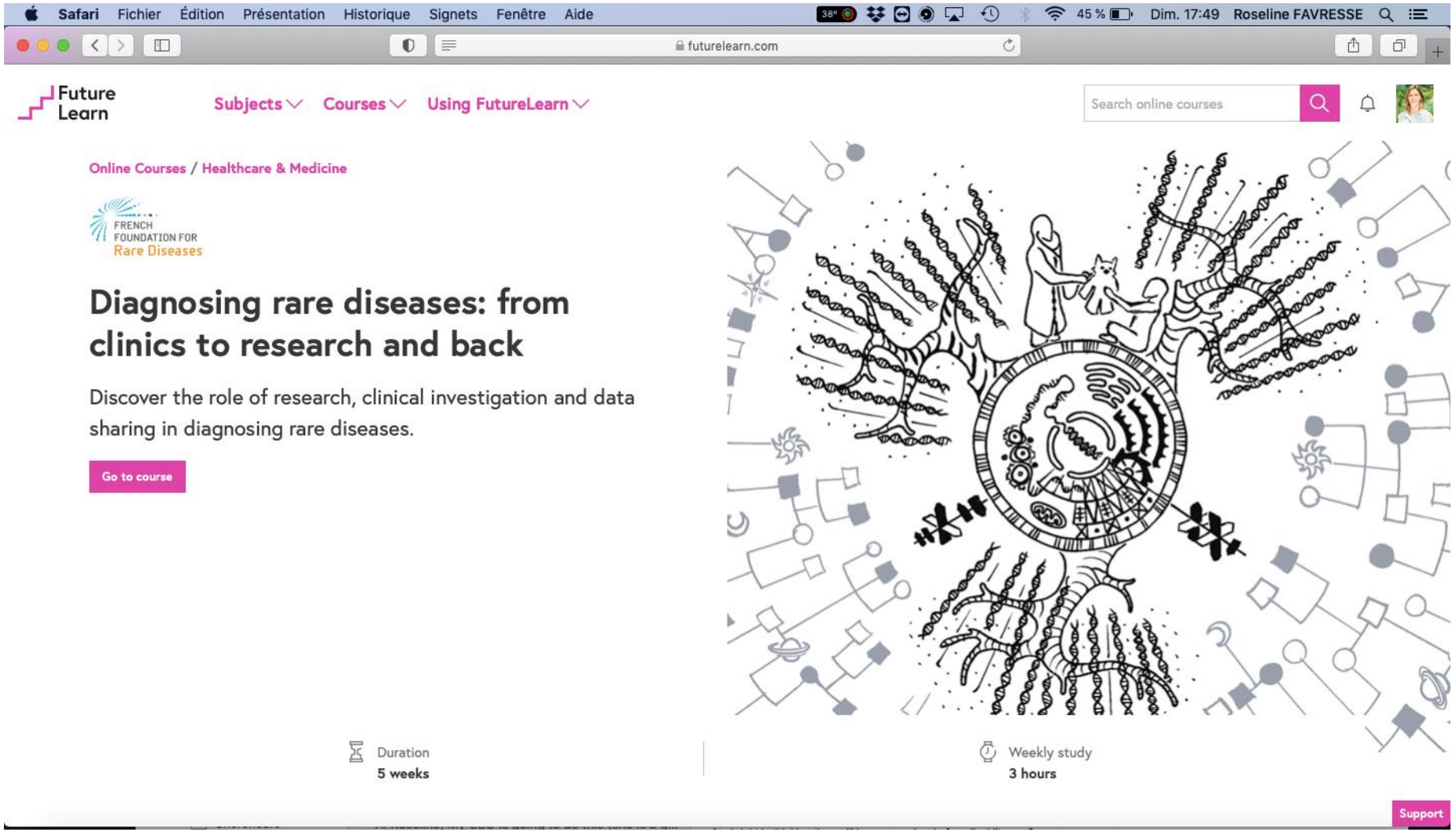

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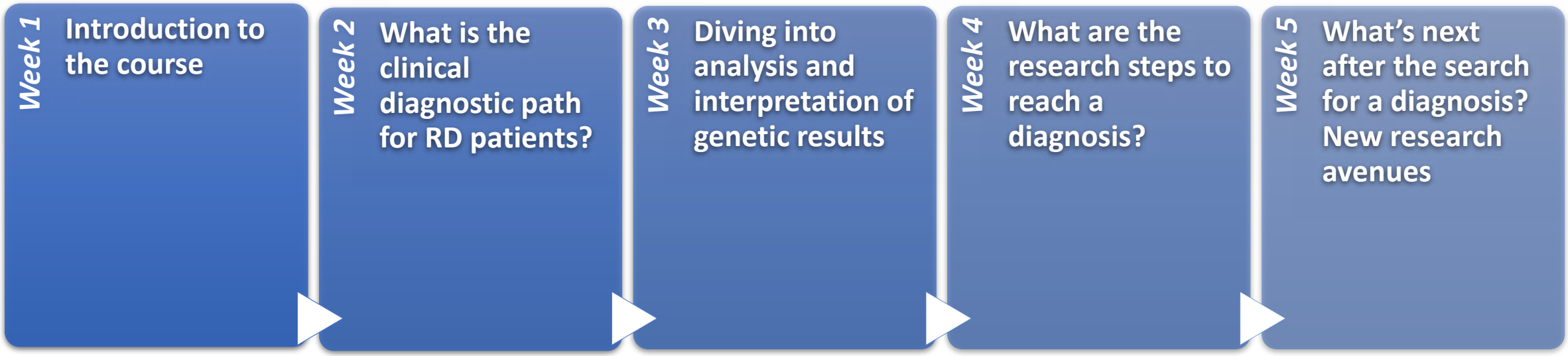
EJP-RD - Online course (WP16) - MOOC Diagnosis

- Organisation of MOOC content
  - *Researching a diagnosis: from clinics to research and back*



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Week 1 | Week 2 | Week 3 | Week 4 | Week 5

Hi Roseline, here's the last step you visited:

4.13 Week 4 Quiz QUIZ

Continue

Diagnosing rare diseases: from clinics to research and back - Dec 2020 (provisional) / Week 3: Diving into analysis and interpretation of gene

Week 2: What is the

Introducing dif
for 3 different

All along week 2, we v
different situations tha
geneticists facing rare

2.1 Introduction

Week 3: Diving into analysis and interpretation of genetic results

From prescribing a genetic test to reporting the results: what are th
steps and which challenges are met? In this week first part, you will
the current available tests in clinical practice to diagnose a rare dise
also learn how Next Generation Sequencing techniques have been a
in clinical genetic services. You will exercise by proposing genetic te
In the second part of the week, you will dive into the interpretation
results. You will be led from the explanation of the human genome v
challenges met when interpreting data, including the different interp
You will also get introduced to the basic principles of bioinformatics and biological analysis to support the genetic analysis. Finally you will learn from different patients' pathways the different situations that can be met when reporting results from genetic testing.

Edit week | View week

Week 3: Diving into analysis and interpretation of genetic results

What are the currently available tests in clinical practice to diagnose a rare disease?

In this item, you will learn about the main modalities, challenges, performance, advantages and disadvantages of the currently available tests to diagnose a rare disease of presumably genetic origin in the clinic.

- 3.1 Introduction ARTICLE
- 3.2 Chromosomal genetic tests ARTICLE
- 3.3 Molecular genetic tests I - Sanger Sequencing ARTICLE
- 3.4 Molecular genetic tests II - Next generation sequencing (NGS) ARTICLE
- 3.5 Definitions: "coverage" and "depth" ARTICLE
- 3.6 Molecular genetic tests III - Targeted gene panels ARTICLE
- 3.7 Targeted Gene Panel: a patient's case ARTICLE

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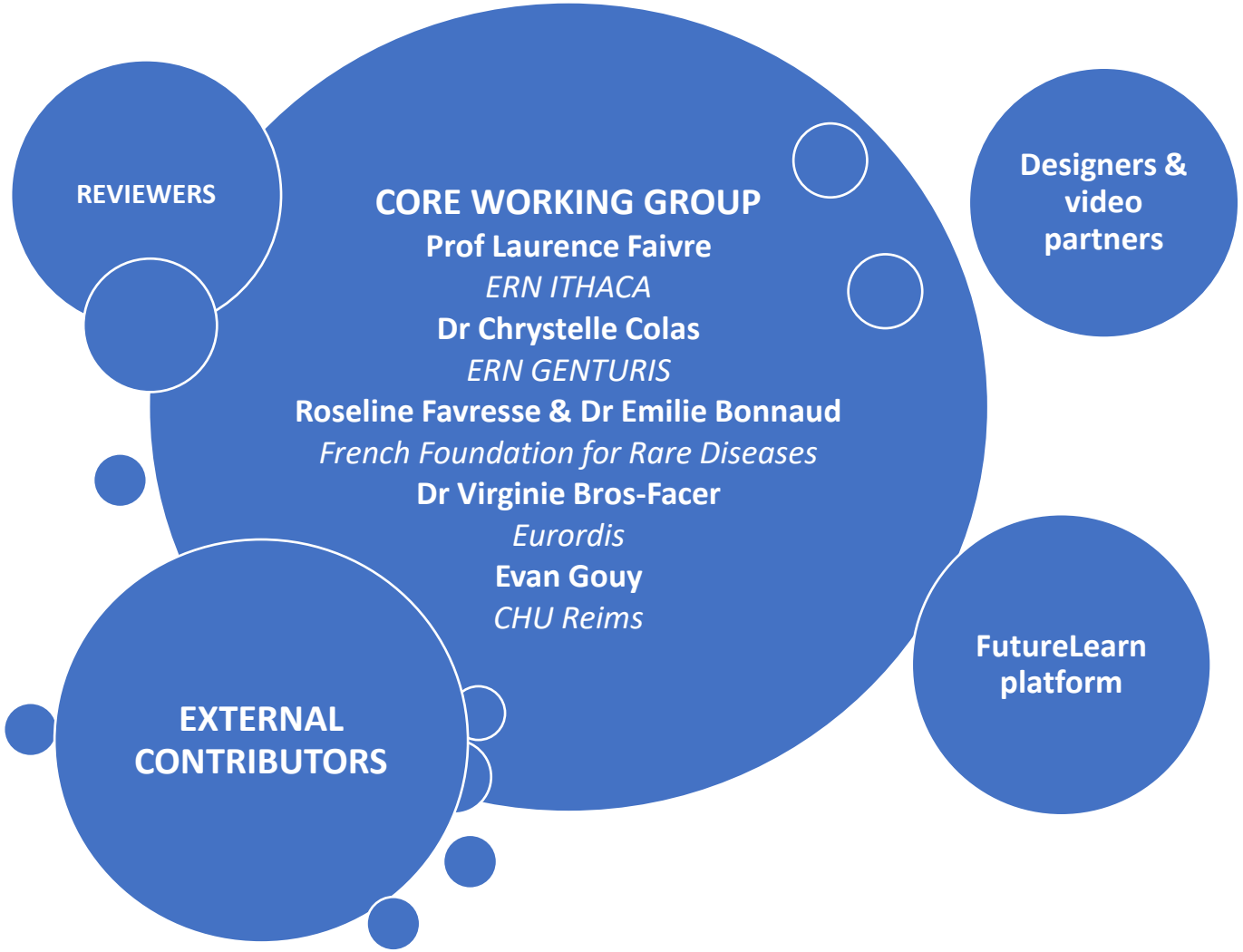
- The way we worked



Special thanks to **ERN ITHACA reviewers**

- Prof Jill Clayton-Smith
- Prof Alain Verloes
- Prof Sylvie Odent
- Prof Annick Toutain
- Dr Sandra Mercier

Special thanks to **ERN ITHACA Contributors**

- Prof Jill Clayton-Smith
- Prof Han Brunner

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EJP-RD - Online course (WP16) - MOOC Diagnosis

- Key Clinicians & Researchers interviewed
  - Prof Jill Clayton-Smith, Honorary Professor in Medical Genetics, University of Manchester, UK
  - Prof Han Brunner, Professor & Head of the department of Human Genetics at Nijmegen University, NL
  - Prof Antonis Antoniou, Professor of Cancer Risk Prediction, University of Cambridge, UK
  - Prof Joris Veltman, Dean of Biosciences Institute, Newcastle University, UK
  - Dr Holm Graessner, Executive Director of the Rare Disease Centre, University of Tübingen, ERN-RND Coordinator, DE
  - Prof Laurence Faivre, Professor & Head of the department of Medical Genetics, Dijon University Hospital, FR

⚠ **Covid-19 obliged us to rely on filming onsite teams (travels not possible for filming team at X locations)**

⚠ **Access to facilities not always granted → audio recording**

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EJP-RD - Online course (WP16) - MOOC Diagnosis

- Patients' testimonies embedded all along the course

Testimony - Eva's story

0 comments

You will now learn from patients about their personal experience of being diagnosed with a rare disease.

Eva is a woman in her thirties from South America who was diagnosed a few years ago with CMMRD (Constitutional mismatch repair deficiency syndrome). This is a rare, inherited cancer-predisposing syndrome characterized by the development of a broad spectrum of malignancies during childhood, including mainly brain, hematological and gastrointestinal cancers.

"It all started when I went back to my homeland to spend some time with my family. I had left my homeland two year prior to study in France without being able to come back and was thrilled to finally have some time off.I started feeling poorly with constant stomach aches.

I saw several doctors, who gave me common drugs for such symptoms, but nothing worked.

1.15 You've completed 1 step in Week 1

Testimony - Emily's Story Part I

0 comments

There is nothing more powerful than a parent's testimony explaining the feelings of the family during his/her child's diagnostic odyssey.

Let us share with you Emilie's wonderful testimony in her search for a diagnosis for her daughter affected with an undiagnosed rare disease.

Emilie is the mother of a 7-year-old child at the time of this first testimony (mid-2016).

Testimony: Joan's story: "I like to see a doctor who sees the bigger picture"

0 comments

"I like to see a doctor who sees the bigger picture"

You will now learn about Joan's story. Joan is a patient diagnosed with PTEN Hamartoma Tumour Syndrome (PHTS) (OMIM 158350).

PHTS is caused by pathogenic germline variants in the *PTEN* (Phosphatase and TENsin homolog) gene with an **autosomal dominant transmission**. PHTS encompasses mainly two syndromes: Cowden syndrome and Bannayan-Riley-Ruvalcaba syndrome. It is a multi-system disorder predisposing to the development of hamatomatous growths in different tissues and an increased risk of breast, thyroid, endometrial, renal and colorectal cancers at an adult age.

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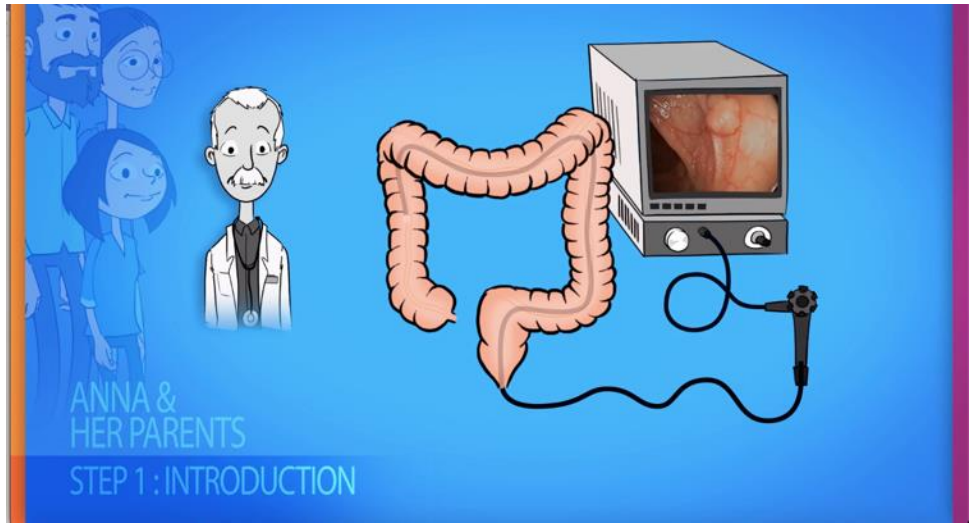
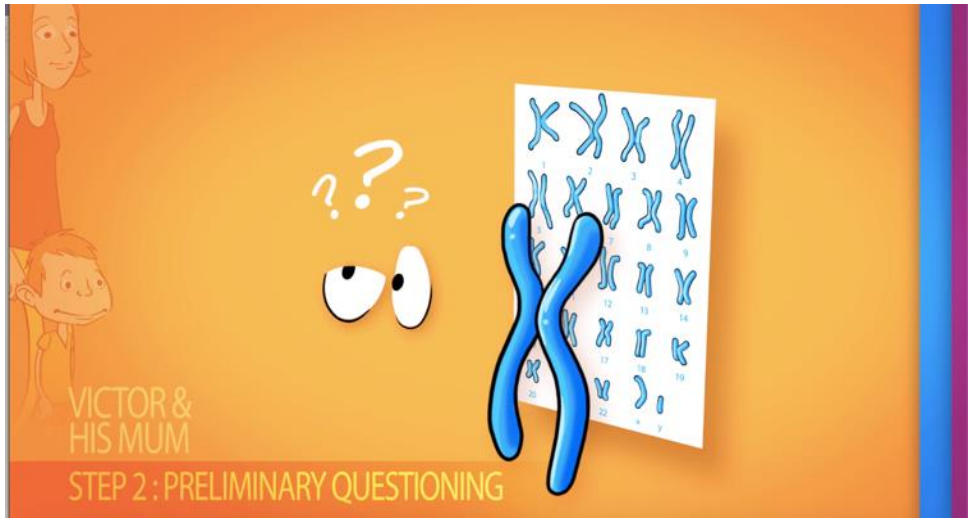
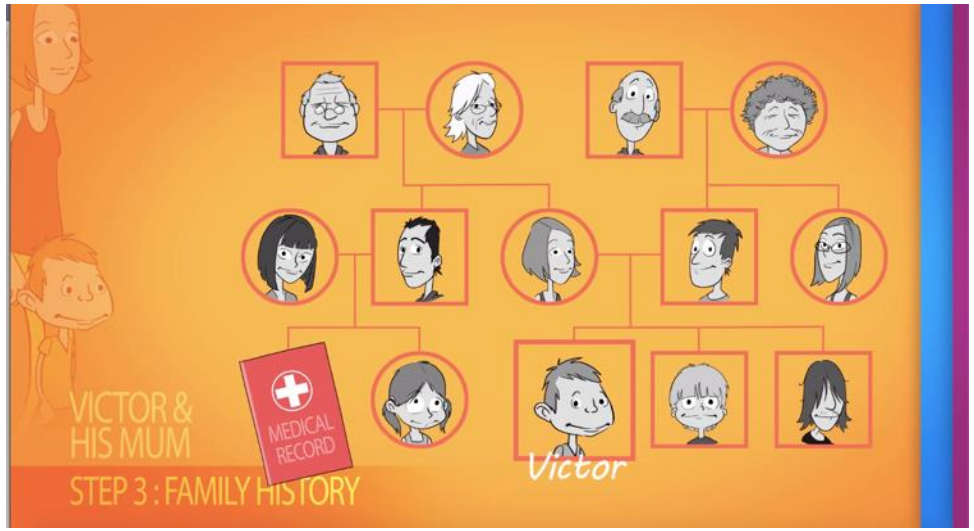
EJP-RD - Online course (WP16) - MOOC Diagnosis

- 2D video supporting week 2 throughout the steps of a genetic consultation







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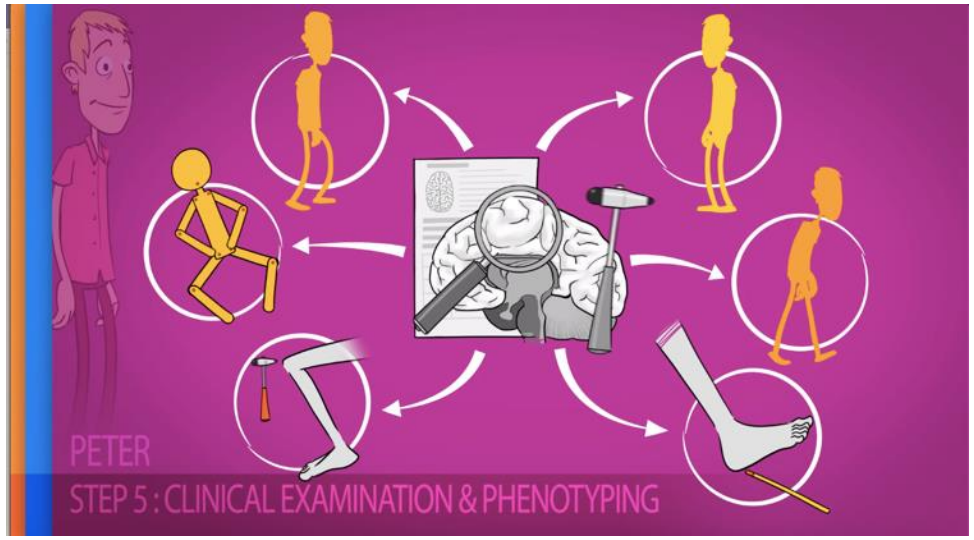
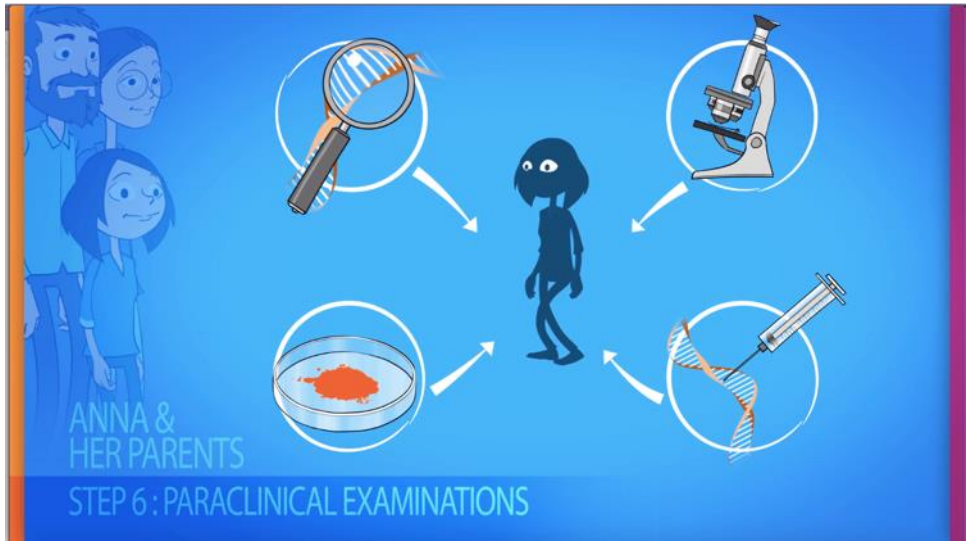
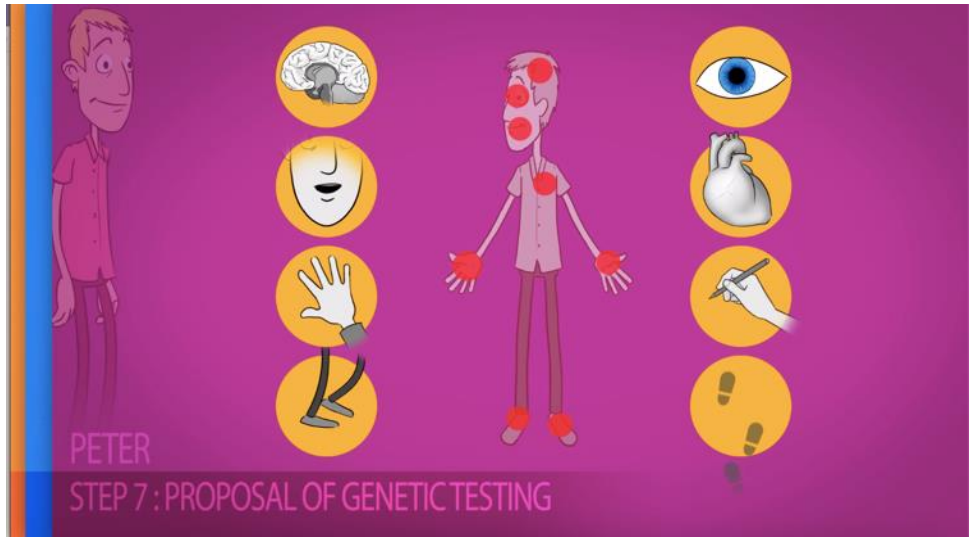
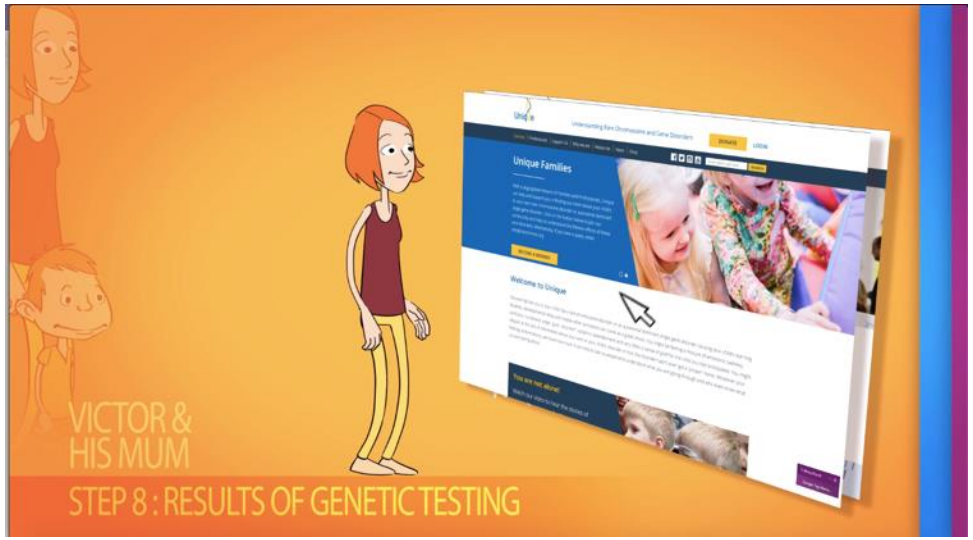
EJP-RD - Online course (WP16) - MOOC Diagnosis

- 2D video supporting week throughout the steps of a genetic consultation









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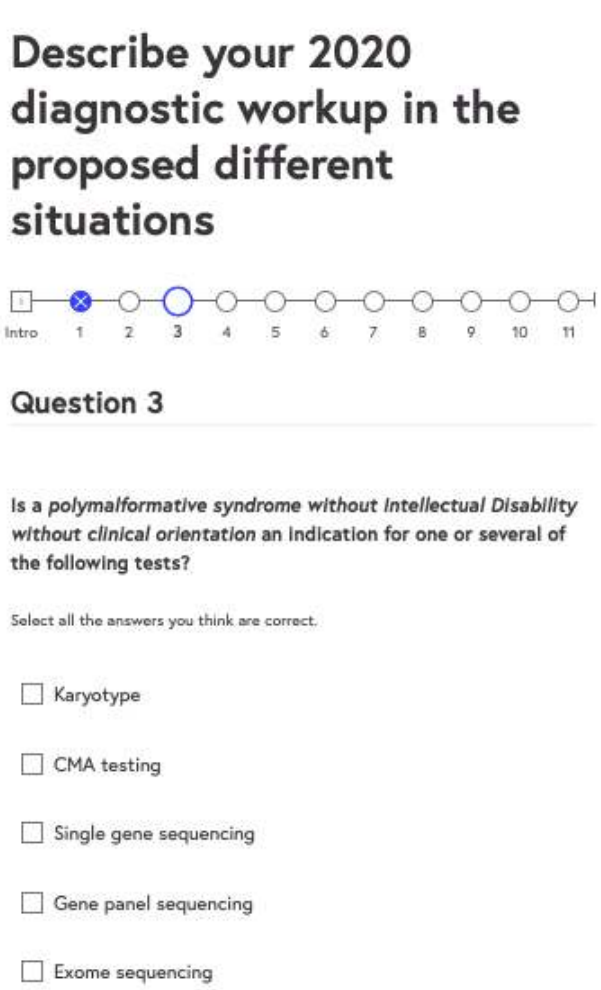
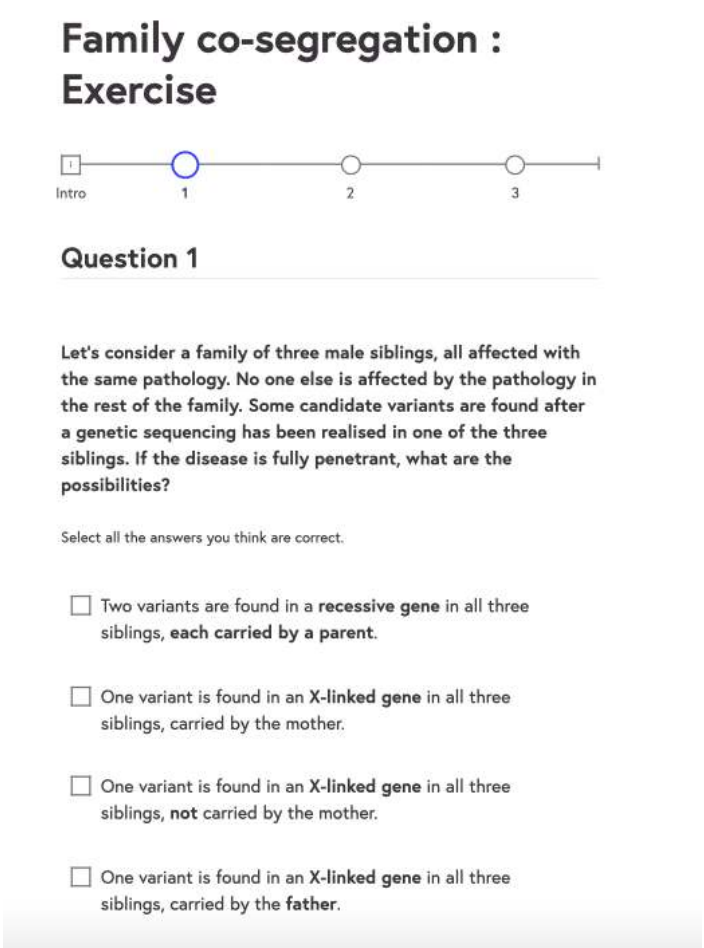
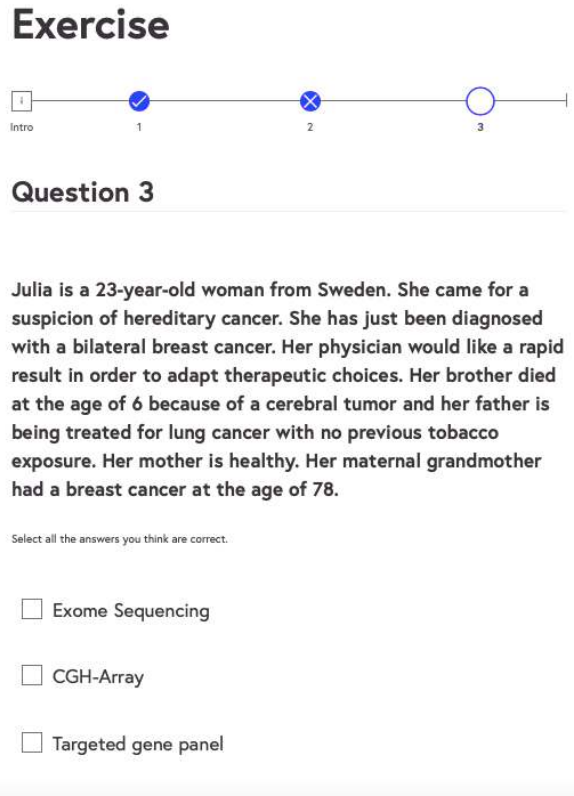
EJP-RD - Online course (WP16) - MOOC Diagnosis

- Numerous exercises & quizzes each week







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Pilot « Master of genetic », Pablo Lapunzina

- There is an idea to set up an European Master of Genetics with contribution of ERNs (mainly ITHACA) together with CIBERER (Spanish National Centre of Biomedical Research on Rare Diseases).
- At this moment, there will be 3 different potential scenarios:

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Pilot « Master of genetic »

Scenario 1. Just an additional European Course, taking advantage of both meeting (CIBERER and ITHACA) to optimize the presence of faculty and students.

Scenario 2. To include the Course into a current, already accepted Master: possible but students attending the Course will not have the official recognition of a University Master degree….

Scenario 3. To set up a new European Master (it will take about 2 year to get the accreditations...).

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Pilot « Master of genetic »

How? Preparing a blended (online+face-to-face) Master (e.g. 80% online classes with printed material and powerpoint lectures (either live or recorded) and 20% face-to-face classes in 2-3 weeks along the year (*e.g.* in two different European locations: Madrid, and wherever).

Needs: Master Thesis work and assistance to labs/clinics

University/ies: One University functions as the leader/coordinator and 2-3 additional Universities as partners.

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Pilot « Master of genetic »

What do we need from the partners?

1-Interested physicians and researchers from ITHACA, please contact us to indicate your interest.

2- Anyone with close relation to his/her University and with experience in international or European similar activities, please tell us.

3- We will explore the Spanish Universities with agreements (partnerships) with other European Universities and tell you soon.

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Syndromic Games & Vite Extra-ordinarie
Giuseppe Zampino

- How to increase the interest on rare disease of medical students?
- How to increase the knowledge of different aspects of disability?
- How to increase the medical competence?

- The first idea was to use a competition-game as an instrument to increase competences:

The Syndromic Games.

- The aim is to teach a comprehensive care of syndromic complex children. The Games consist in a competition among teams of paediatrics and geneticists, in which the different aspects as etiophatogenetic diagnosis, the functional diagnosis, the approach to the most important problems jointed to disability and the communication of diagnosis were faced.

- The second idea was that if a condition is connected with an emotion it will never be forgotten. We use films as didactic instrument :

“Vite Extraordinarie”

- are films describing various syndromes and social, psychological and human aspects connected with them.
- Before every film a clinical geneticist or a neurologist or a paediatrician explains the clinical condition that is described in the film and then a psychologist or a sociologist or a bioethicist analyses the human aspects.

- Third idea : **A cartoon for children in which are explained various medical processes.**

Ex: made by the **Associazione Italiana sindrome di Beckwith Wiedemann** has created to explain to the children, the importance of blood sampling.

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Syndromic Games

The Syndromic game

- It consists of randomly assigned teams.
- At the very beginning, each participant will pick a piece of the puzzle and put it aside.
- Puzzles are 10 in total and each one of them consists of 5 pieces that, once reassembled, will show the image of a child affected by a syndromic disease.

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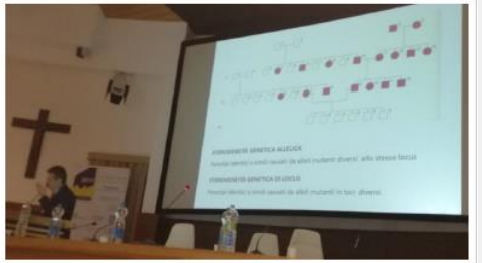
Syndromic Games – Step 1

1 STEP ETHIOPATHOGENIC DIAGNOSIS

- 4 round of presentations lasting 40 minutes each:
- 1)Dysmorphological semeiotics;
- 2)Anamnesis and genealogical tree;
- 3)Request for and interpretation of genetic tests;
- 4)20 syndromic clinical cases (each syndrome will be described in 4 minutes).

- At the end of all presentations, a televote with 40 questions will be held. A score based on both the rapidity and the correctness of their answer will be assigned to each participant.



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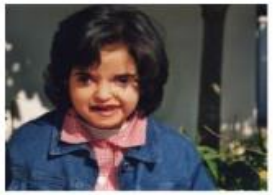
Syndromic Games – Step 2

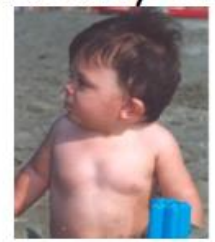
2 STEP GESTALTIC recognition.

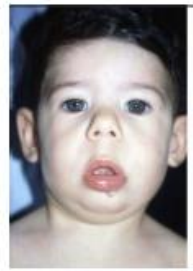
10 teams of 5 individuals each will be formed.

In the room, there will be 10 stations, one for each syndrome, and all participants will go to their station in order to reassemble the puzzle.

At that point, teams will be formed and will get a cumulative score, corresponding to the sum of the individual scores previously obtained with the televote.







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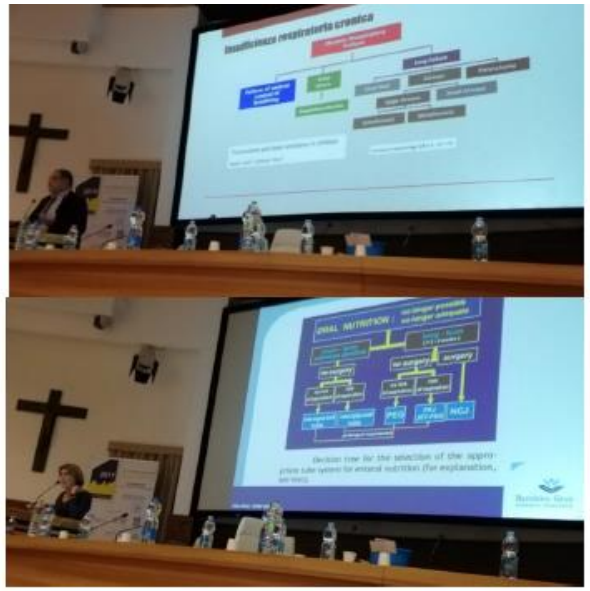
Syndromic Games – Step 3

3 STEP – MANAGEMENT of common clinical difficulties faced by the DISABILITY PEDIATRICIAN

1) Management and organization of clinical care in complex children;
2) Respiratory difficulties in children with malformative syndromes;
3) Nutritional and gastroenterological issues in children with malformative syndromes.

Two stations with mannequins will be placed: 1) tracheostomy management; 2) PEG management. For each station, teams will have to give the solution to a randomly selected question.



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Syndromic Games – Step 4

4 STEP – CLINICAL CASE of a COMPLEX child

Teams will have to solve a clinical case of a complex child, starting from the diagnosis to its clinical management.

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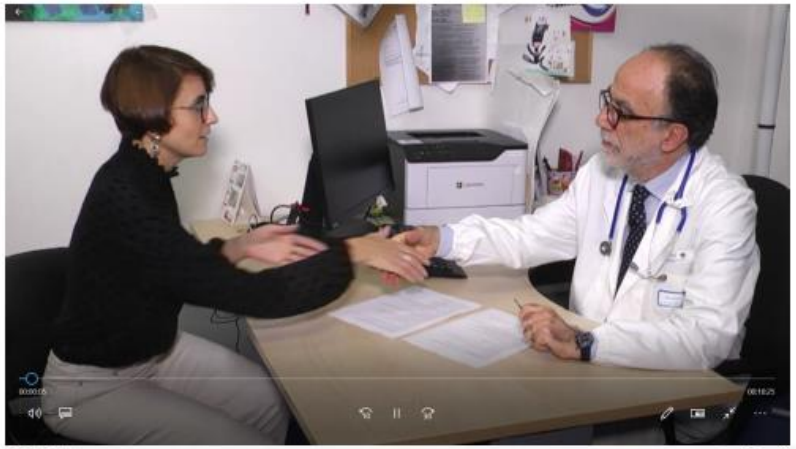
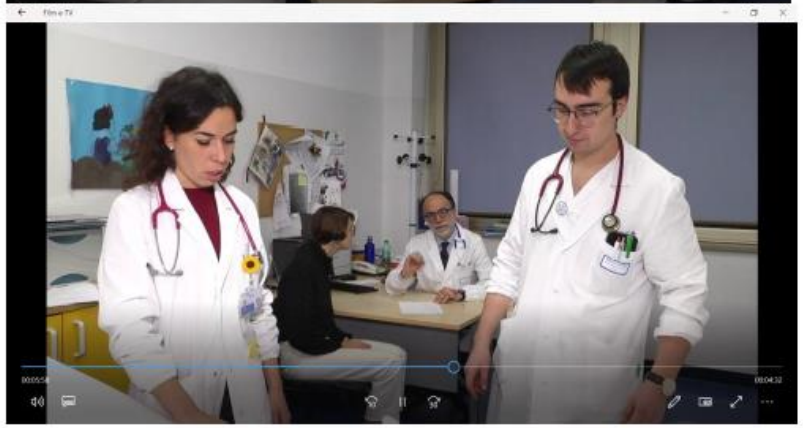
Syndromic Games – Step 5

5 STEP – COMMUNICATING the DIAGNOSIS

2 videos will be shown about how to communicate a diagnosis. In these videos, several mistakes will be purposefully made and teams will be asked to identify as many mistakes as possible.





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Syndromic Games - *final*

Team with the highest score, obtained by the sum of the televote score, the mannequin practical test, the solution of complex case and the communication test, will be the winner of the Syndromic Game.

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Vite Extra-ordinarie

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Think of Chidren / Cartoon
& Associazione Italiana sindrome di Beckwith Wiedemann

- Third idea : **A cartoon for children in which are explained various medical processes.**

Ex: made by the **Associazione Italiana sindrome di Beckwith Wiedemann** has created to explain to the children, the importance of blood sampling.

Together with the Association of families we can develop different productes for children in which are explained various medical processes. Associations are proud to make a product of utility for their children

- Idea of cost is ~ 2.500 euro
- We can do cartoons to explain
- 1) Echocardiogram and the association of noonan syndrome can pay for it
- 2) MRI and the association of NF1 can pay for it
- 3) cistoscopy and the association of Costello can pay for it
- 4) polysonnography and the association of Smith magenis can pay for it.
- 5) etc.

Discussion - For ITHACA, we could adapt the material to English and other languages (while keeping your group acknowledged for the paternity of the document. ?

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Discussion on webinars 2021

Discussion on new training needs and goals and Topics proposed :

Two propositions must be discussed with the group

1. **Webinars "what I know best" For a specialized audience**
   - Following the idea of the importance of training the younger generations (young geneticists' young specialists), and isolated fellows
   - Starting from a basic education of reflexes for good professional practice
   - Share knowledge's on diagnostic strategies, general care of ID, syndrome
   - How to set up webinars: on a secure platform or as a video meeting.

2. **Webinars "what I know best" For a more general audiences (paramedics, nurses, patient's organization's, children) Layman versions** (link to a new proposal from the EU ERN WG Group Knowledge generation to T&T group **)**
   - An ERN new Strategy on Training and Education on Rare Diseases and Complex Conditions
   - Promotion of patient education, generalist vision co-constructed with the OPs
   - Patient education should become one of the major focus of the ERNs' educational activities

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Immediate action on : webinars & other productions

“What are our best experiences to be shared “and How !

We propose the WG to suggest interesting subjects for the community and to define some speakers, and fix an Agenda for next year

1. Share initiatives focusing on ultra rare diseases
2. Commons needs in most disorders (sleep, behaviour, case management, coordination of care, gastrointestinal)
3. Teach children the necessity to know about themselves
4. Disabled children
5. Aging and intellectual disability
6. Covid ?

Question

- Be aware that most HCP are geneticists' doctors, we need to identify others expertise to complete our goals
- We may also discuss whether English language is a barrier to understanding depending on the audience.
- More open Webinars / general

Practical appointments:

1. timeline development : 1x/trim
2. recommendations
3. task assignments for the participants
4. searching representatives of missing disciplines
5. coordination
6. administrative support
7. meeting schedule

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Updates on Serious Game

A virtual game, and an innovative training Tools to facilitate cross-border interactions

Learning goals

- Understand the issues of the diagnosis
- Think of a rare disease facing a neurodevelopmental disorder
- Discover the prescription way of care
- Way to announce diagnosis and understand its impacts

Targets : professionals of care wishing to upgrade their knowledge,
General practitioners and pediatricians, parmedical professionals, students.

Training content : The player is a “general practitioner”. He discovers four young patients, Lina, Tom, Alex and Gaël, whose developmental trajectory questions him. From the identification of the first warning signs to the search for the etiological diagnosis, the different scenarios allow him to deepen his knowledge in the field of neurodevelopment and to understand the role of the general practitioner and the pediatrician, the coordination of the course.
Designers : medical experts, Patient organisations, and other users from the network
Budget = 80 000 € Co-financed ITHACA (INEA CALL) + DefiScience

Pr Vincent des Portes/ Marie pierre Reymond/ Caroline Immesoete / Anne Hugon

Updates on Serious Game

1. **ID-Autism/ Tom, 14 months and Alex, 18 months.** Two worried families. You're taking over the practice of a colleague, Dr Joseph, who's retired. You will successively accompany the journey of Tom and Alex, who seem to present quite similar symptoms.
2. **22q11 syndrome/ Lina, 18 years old**. When one disorder can hide another. You are taking over the practice of a colleague, Doctor François. You are going to accompany Lina who is experiencing difficulties in her school career.
3. **Angelman syndrome/ Gaël 12 months/8 years oldc and 20 years old.** Thunderbolt in a serene sky. You are taking over the practice of a colleague, Doctor Sylvani, who has retired. You accompany Gaël throughout his life. You get to know him and his mother when his life is turned upside down by a probable epilepsy.

These situations are based on real facts. Any resemblance with "real life" would not be fortuitous!

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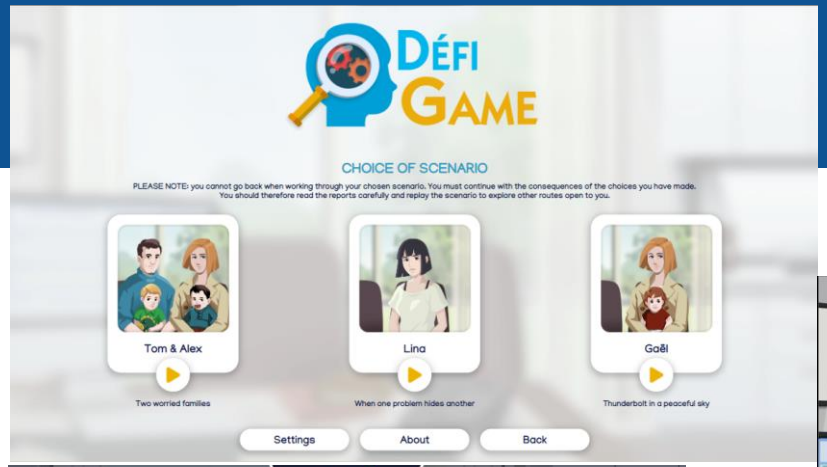
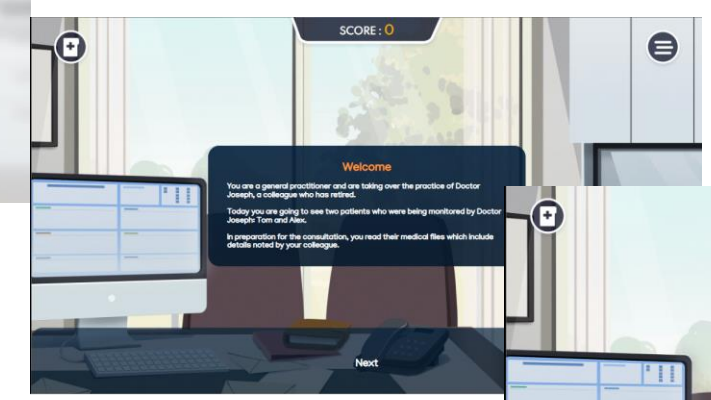
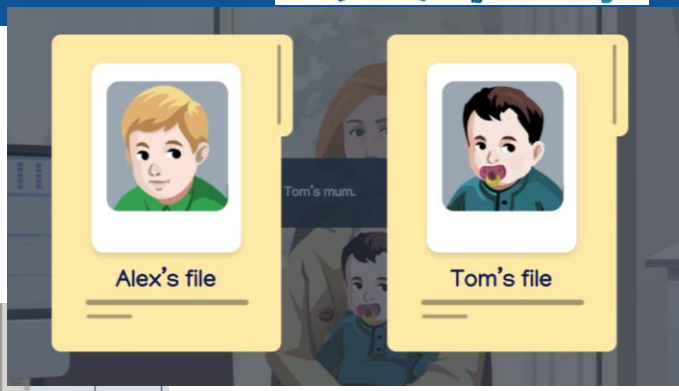
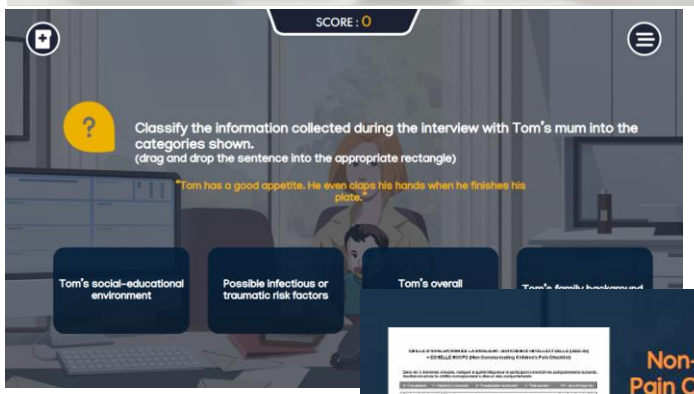
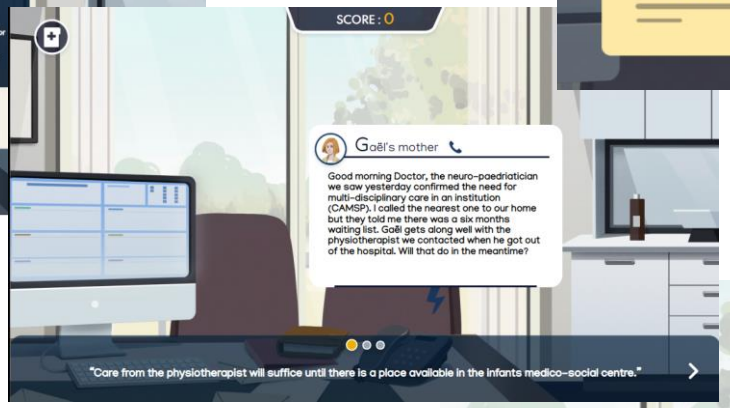
https://tnd-cases.dowino.com/app/#/generic

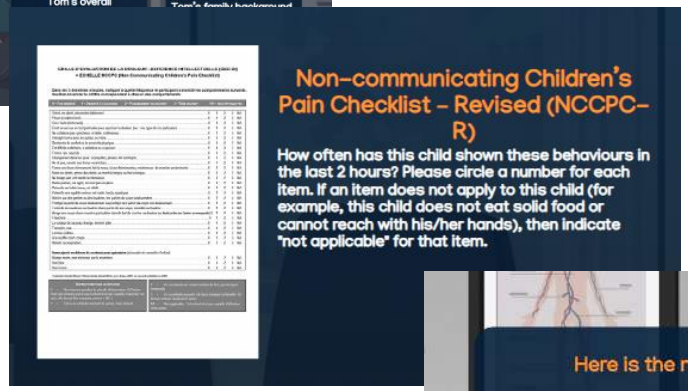


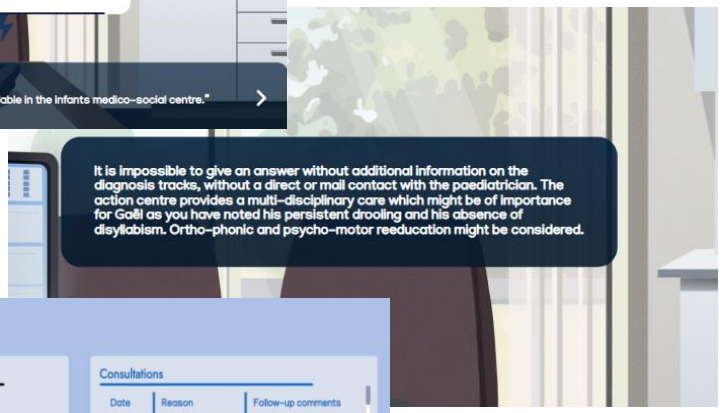


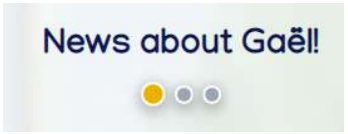


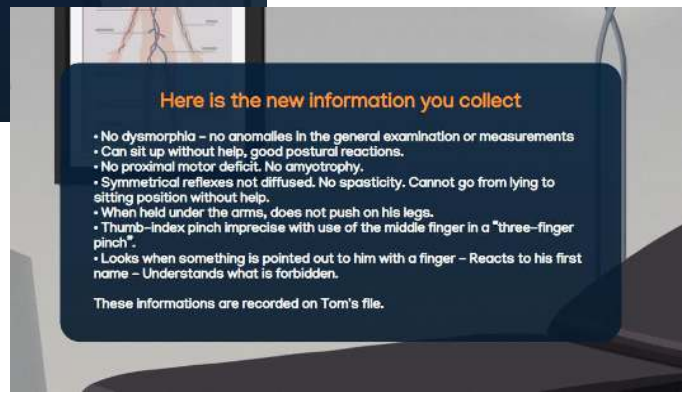


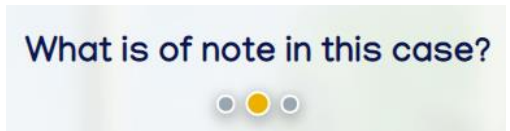












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Updates on Serious Game

NL°10 dec 20: last news

- ✓ the French version of Défigame is validated !
  - ✓ the returns of the first test period are integrated and adjustments have been introduced
  - ✓ the result is consistent and faithful to the work of the design groups

- ✓ It is a real training tool based on real clinical cases, with validated materials such as medical reports, quizzes and resources to complete the training.
- ✓ It takes 1 hour of learning per situation, or 3 hours for the complete game

- ✓ **All of the game's content is translated in english.**
- ✓ **The final and complete version of the Game will be available in early 2021.**

Made for any professionals who must assist in the identification and guidance of patients suspected of neurodevelopmental disorders

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Young Geneticist Network (YGN)

Where do we come from ?

2017 Sept. — ITHACA meeting Manchester

2018 Feb. — YGN Fb group

2018 Jun. — 1st meeting ESHG Conference Milan

2018 Oct. — ITHACA meeting Paris

2019 Jun. — 2nd meeting ESHG Conference Göteborg

2019 Nov. — ITHACA meeting Dusseldorf

2020 Jun. — 1st Worshop Virtual ESHG Conference

2020 Dec. — Virtual ITHACA meeting

Special thanks to Claudia Ciaccio, Berardo Rinaldi and Aurélien Trimouille.

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Young Geneticist Network (YGN)

- Facebook group created 2018
- December 2020
  - 2,4k members in 2020, December

Training in Human Genetics is very heterogeneous

Isolated trainees learn highly-specific expertise areas

: Connect young human geneticists around the world

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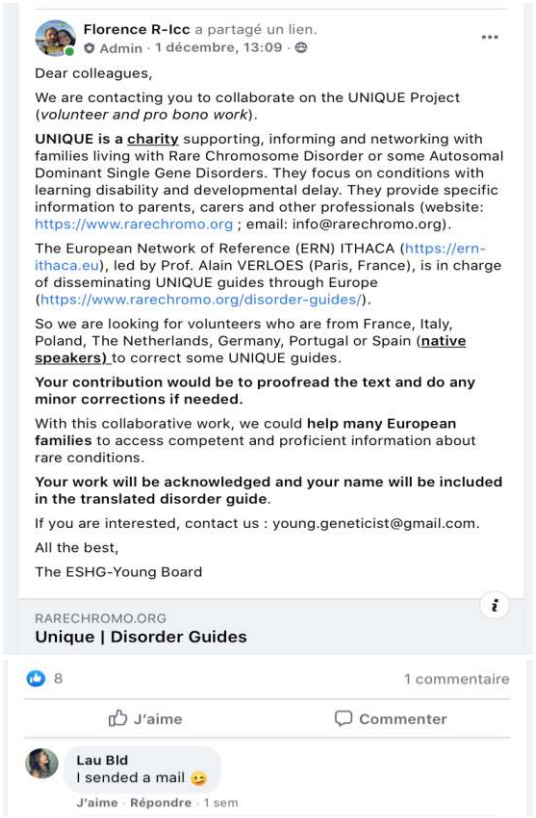
Network Intellectual Disability and Congenital Malformations (ERN ITHACA)

Young Geneticist Network (YGN)

- Facebook group created 2018
- December 2020
  - 2,4k members in 2020, December
  - of the last 60 days:
    - ✔ 62 publications
    - ✔ 44 comments
    - ✔ 168 reactions

➢ Membership questionnaire implemented in 2020

➢ Cleaning of the group in progress...



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YGN in Europe

European referees

EU-27

-France: Mario Abaji, marioabali@hotmail.com
-Italy: Berardo Rinaldi, berardo.rinaldi@gmail.com
-Romania: Ioana Streata, ioana.streata@yahoo.com
-The Netherlands: Margot Reijderd, margot.reijnders@mumc.nl
-Portugal: Celia Soares, cmas00@gmail.com
-Germany: Malou Schadeck, malouschadeck@gmail.com
-Belgique: Laura Boulard, laura.bourlard@erasme.ulb.ac.be
-Spain: Toni Martinez Monseny, afmartinez@sidhospitalbarcelona.org
-The Czech Republic: Marek Turnovec, marek.turnovec@lfmotol.cuni.cz
-Lithuania: Rasa Traberg, rasa.traberg@kaunoklinokos.lt
-Luxembourg: Arthur Sorlin, arthur.sorlin@gmail.com
-Poland: Aleksandra Pietrzyk

And

-United-Kingdom: Zerin Hyder, zerin.hyder@doctors.org.uk
-Russian Federation : Elena Vereshchagina , ev-veresh@rambler.ru

Creation of the first official office 'ESHG-Young' in 2019/2020

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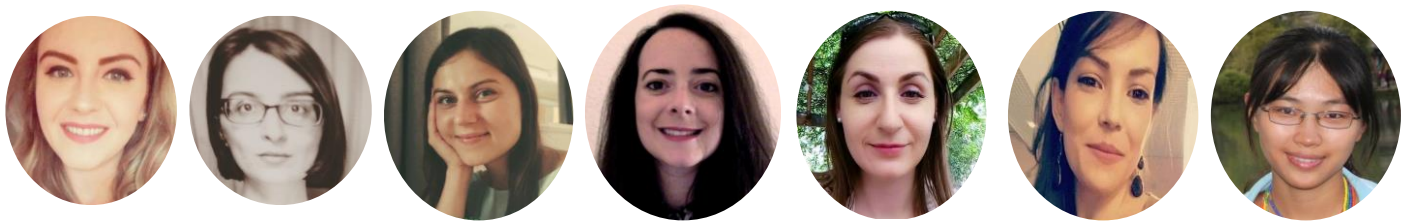
ESHG-Y

- **2020/2021 board**
  - 7 young ladies
  - Different countries and backgrounds



Patricia CALAPOD
Romania

Celia SOARES
Portugal

Ruta MARCINKUTE
UK

Florence RICCARDI
France

Elena AVRAM
Romania

Juliana MIRANDA
Finland

Can DING
Germany

- Official committee within ESHG
  - Since November 2020
  - Statutes approval in progress.
  - ESHG-Y website page in process (https://www.eshg.org/eshgy)

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- ESHG Conferences:
  - 2020: W05 ESHG Young Board and YGN - Professional Development for the Next Generation
  - 2021: Educational session (E0?) > 'Human organoids as genetic disease models'

- ESHG 'Education Committee' :
  - 2 members of the ESHG-Y for a period of one year
  - Call for volunteers for 4 years > 3 other members of the board have applied

- Link with ERN ITHACA
  - Correction of translation of documents Unique +/- MOOC BIG +/- Orphanet
    - ✔ 4 volunteers for the MOOC BIG
    - ✔ 6 emails for the Unique guide project
  - Eurodysmorpho Conference

YGN and ESHG-Young

- **Perspectives**

  - Raising awareness about our community
  - Facilitating access to training in Human Genetics
  - Getting the younger generation involved into ERN,

    in particular ERN ITHACA !

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T&T deliverables Y 4-5 Futur objectives

Sarting from 2021 we will be preparing a new 5-year workplan for ERN ITHACA

WP8	D8.1	Set up a module in a European Master's in Genetics programme	Create a Master's level module covering new advances in ITHACA-specific disorders and research domains for European students	Other	Public	28 Feb 2022	Pending

- **Pilot Project – Madrid 2021**

WP8 + CEF 2	D8.2	Develop a web-based learning tool for non-specialists to handle patients with ID/developmental anomalies	Development and availability online of a serious game on handing rare ID patients for non-specialists (partly funded through CEF eHealth call)	Websites	Public	28 Feb 2021	Pending
WP8 + CEF 4	D8.3	Develop an eLearning module for specialists to learn how to handle ID/developmental anomalies	Create MOOC on bioinformatics methods and protocols to study ITHACA disorders in collaboration with the Young Geneticists Network (ESHG-Young) (partly funded through CEF eHealth call)	Websites	Public	28 Feb 2021	Pending

D8.2 and D8.3 of ERN ITHACA were turned into INSTEAD project

- In development: MOOC + Serious game
- Not a deliverable but requested for evaluation
  - **Webinars (diagnostic strategies, general care of ID,…) : volunteers**

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WG ID projet forecast

- New members expectations
- Looking for new co-deputy chair
- Agenda next meeting 2021/22 - *Bimonthly TC/or 3/4 per years*

Do not hesitate to send your feed back
Thankyou for your participation: PM contact - Anne.hugon@aphp.fr

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