

## ERN Support and training programma-WP17-EJPRD

### Report Workshop From high throughput sequencing to diagnosis in immune mediated disorders ESID/ERN-RITA Workshop

Imagine Institute, 24 Boulevard du Montparnasse, Paris, France  
**May 30-31, 2022**

#### Date/Venue /Format

Dates: May 30-31, 2022  
Venue: Imagine Institute, 24 Boulevard du Montparnasse, Paris, France  
Format: Hybrid (Face-to-face, combined with online)

#### Numbers (participants, ERN's represented, speakers including patient representatives)

Total participants (ESID+EJP): 141 (42 on site, 99 online)  
EJP participants: 14 onsite, 7 online; originating from Netherlands, France, Italy, Norway, Portugal and Spain.  
ERNs represented by EJP participants:  
1. ERN RITA (n=11)  
2. ERN EYE (n=1)  
3. ERN GENTURIS (n=2)  
4. ERN EuroBloodNet (n=1)  
5. Participants from centers belonging to >3 ERN's, such as ERN RITA but also ERN LUNG;MetabERN;ERN EURACAN;ERN EpiCARE; ERN EURO-NMD;ERN ReCONNET; ERKNet; (n=6)

Speakers:

#### **Day1 Focus on genome analysis including innovative molecular and informatics tool research and diagnostics methodologies**

Prof Isabelle Meyts, Dr Aurélie Cobat, Dr Alexander Hoischen, Dr Anna Mensa, Prof Janna Saarela, Dr Yuval Itan, Dr Esteban Ballestar, Dr Marielle van Gijn, Dr Jérémie Rosain, Prof Roger Colobran, Dr Andres Caballero Garcia de Oteyza.

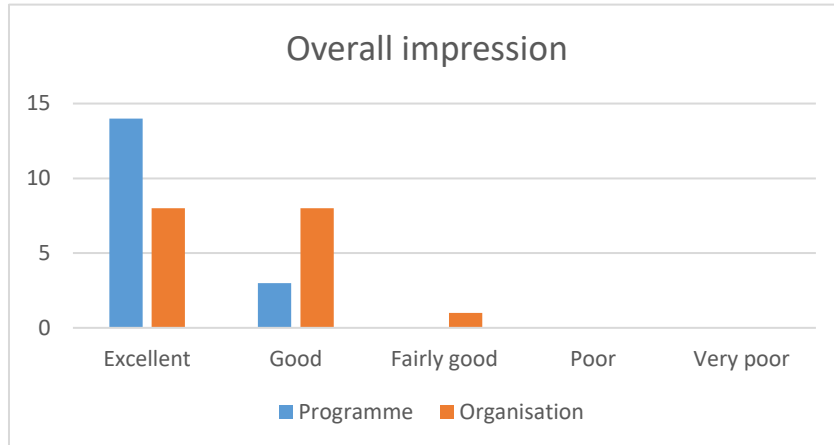
#### **Day 2: Functional validation strategies an evaluation of VUS/Patient perspective**

Prof Holm Uhlig, Prof Sophie Hambleton, Dr Gigliola Di Matteo, Dr Kimberly Gilmour, Dr Vanessa Sancho-Shimizu, Dr Mirjam van der Burg, Julie Power, Martine Pergent, Dr MarcoCrimi.

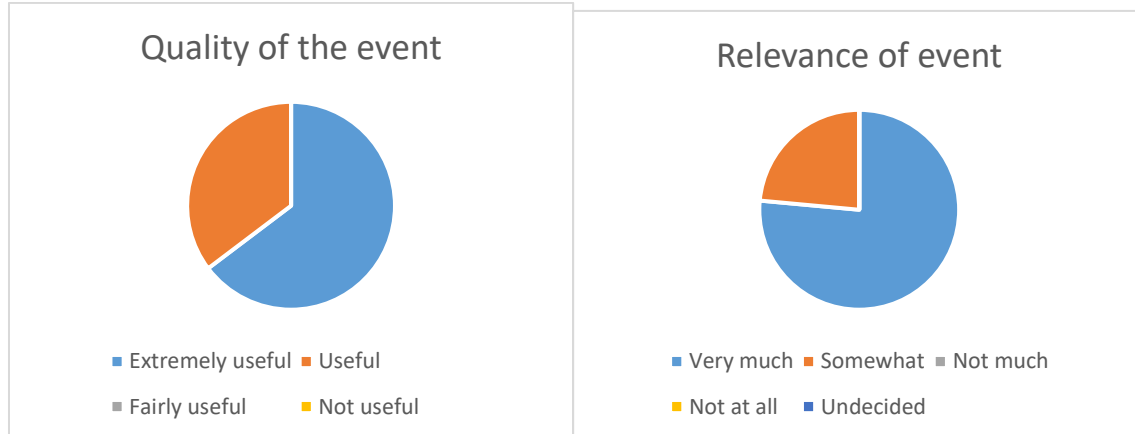


## Analysis of the workshop satisfaction survey

After the workshop, a survey was sent to all participants, both to onsite and online participants. The survey was filled out by 17 EJP participants, of whom 3 online participant. The results of these participants is described. Overall, the outcome of the survey was positive.

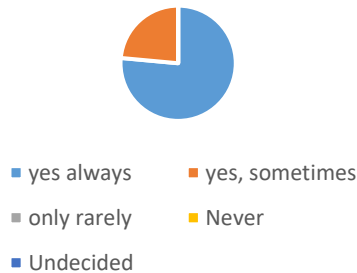


The results from the overall impression is shown above. Almost all participants rated the overall quality of the workshop as excellent or good. The lowest rating of the organization of the workshop was 'fairly good' (n=1).

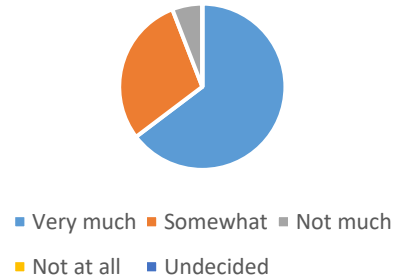


The event was considered useful for participants professional activity (extremely useful n=11; useful n=6) (see above left) For three-quarters of participants the event fulfilled their educational goals and learning outcomes and the workshop was considered highly relevant (see above right). All participants (n=17) found the presented information well balanced and consistently supported by a valid scientific evidence base (not shown).

### Suitability of formats used during the event



### Ways the event affects clinical practice



All participants found the format of the event suitable ((almost)always n=13, sometimes n=4); there was adequate time available for discussion, questions& answers and learning engagement (above left).

The event is likely to affect clinical practice. Sixteen of 17 participants reported the information learnt will be implemented in clinical practice (very much n=11 and somewhat n=5) (above right). Example quotes on how the event influenced future practice were:

- 'I learnt new software and analysis strategies'.
- 'Interpretation of the sanger electropherogram for mosaicism will help to solve some of my unsolved cases'.
- 'I will improve my molecular approaches into challenging cases'.
- 'Will definitely look at HPO and gemma database (first time I heard it)'.

As the best aspect of the event were mentioned the **hybrid format** and the **interactive discussions regarding the clinical cases**. Few points of improvement were mentioned by the survey respondents:

- 'The workshop was quite dense, the same program in more days is suggested'.
- 'Maybe more hands on sessions would be nice'.
- 'Regarding the organization; there was no gluten free food'.

### 2-3 participants' testimonials

'Very complete program with interesting sessions and case presentations'.

'Wonderful interaction during case presentation'.

'As a clinician it helped better understand all molecular tools to evaluate difficult clinical cases'.

