

## **EJP RD**

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## **AD38**

### **Annual report on implementation of training programs**

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## 1. Delivering training programmes through the ERNs

### 1.1. Calls for research workshop topics

In its objective to deliver training programs in the form of workshops, WP17 has continued to conduct calls in year 3. The aim of those calls is to identify the most suitable topics for the organization of research training workshops targeted to the ERNs. Selected research training workshops will have to train ERN researchers and clinicians in ERN relevant innovative training themes. Training themes may include innovative research methodologies, diagnostic research methodologies, interdisciplinary treatment approaches, such as gene therapy and transplantation, etc. Moreover, the workshops are aiming to provide a cross-ERN added value. The successful applicant receives a financial support to organize a 2-day-workshop.

Two calls for workshop topics were launched in year 3. Table 1 below is summarizing the opening periods of the calls, the number of eligible proposals received, the number of selected applications and the final time point until which the applicants should held their workshop. Due to the Covid-19 outbreak, the initial 9-month period to held the workshop has been extended to 18 months from the date applicants receive a positive answer for funding.

	Opening period	Nr. of eligible proposals received	Date Final SEC evaluation	Nr. of selected applications	Last time point to conduct the Workshop
3. Call Workshop Topics	25.01. - 07.03.2021	11	30.03.2021	8	15.12.2022
4. Call Workshop Topics	06.09.-22.10.2021	6	17.11.2021	5	31.05.2023

Table 1. Summary of calls for research trainings in WP17: Workshops topics

The received proposals are checked for eligibility (persons affiliated to any EJP RD beneficiary institution, ERN-full member or affiliated partner institution) and then evaluated in two steps by the Scientific Evaluation Committee. The first step includes remote evaluation by three members of the SEC and in the second step, the individual evaluations are presented by the three SEC members to the whole committee and a funding decision is made. The selection is based on the following criteria:

#### - Excellence

- Addressing cross-cutting issues relevant for ERNs
- Multidisciplinary aspects
- Fostering ERN collaboration
- No overlap with other EJP RD training activities

**- Impact**

Relevance of the topic for the ERN(s)

Will the topic induce or enhance collaboration within and between ERNs?

Benefits the workshop will bring to the RD community

**- Implementation**

Appropriate training methodology

Appropriate venue of the workshop

The selected workshop topics from both calls launched in 2021 are summarized in Table 2.



Applicant	ERN	Affiliation	Country	Topic	Scheduled / planned date for the workshop
Charlotte Niemeyer	ERN PaedCan	Department of Pediatrics and Adolescent Medicine, University Hospital Freiburg	Germany	GATA2-related Myelodysplastic Syndromes	30 Sept & 01 Oct 2021
Pia Vihinen	ERN EURACA; ERN PaedCan; ERN GENTURIS	Tyks and FICAN West Cancer Centre and Turku University Hospital	Finland	Molecular Tumor Board (MTB), Wide Gene Profiling and AI in the diagnosis and treatment of patients with rare adult cancers	September 2022
Maurizio Scarpa	MetabERN	Azienda Sanitaria Universitaria Friuli Centrale	Italy	The Blood-Brain Barrier: current research and novel therapeutic crossing approaches	1 <sup>st</sup> week of March 2022
Philippe Chevalier	ERN GUARD-HEART ERN EURO-NMD	Hospices Civils de Lyon, CHU Lyon	France	Functional studies of genetics variants found in patients with cardiac and neuromuscular diseases	not yet defined
Donato Bonifazi		Consorzio per le Valutazioni Biologiche e Farmacologiche, Bari	Italy	Modelling & Simulation: Research Methodologies for Small Populations in Rare Diseases	not yet defined
Justine Baccheta	ERKNet, ERN BOND	Pediatric nephrology Unit, CHU Lyon	France	Translational research workshop on bone impairment in rare diseases	9-10 June 2022
Sabrina Sacconi	ERN EURO-NMD	CHU Nice	France	Contemporary outcome measures in neuromuscular diseases	04-05 March 2022

Pascal David Johann / Franck Bourdeaut	ERN PaedCan, ERN EURACAN	Division of Pediatric Neurooncology, German Cancer Research Center, Heidelberg / Institut Curie, INSERM U830, Paris	Germany and France	Strengthening translational research in Atypical teratoid rhabdoid tumors (AT/RT) - ancillary studies to the SIOPE-ATRT01 trial	11 & 12 June 2022
Arvid Irmeijs	ERN GENTURIS	Pauls Stradins Clinical University Hospital, Riga	Latvia	Psychological, molecular and administration aspects of Hereditary breast and ovarian cancer genetic population screening (HBOC GPS)	not yet defined
Isa Houwink	ERN Bloodnet	Leiden University Medical Center, Dept. Public Health and Primary Care	Netherlands	Blended learning: Workshop how to design and evaluate an e-learning for rare diseases taking hemoglobinopathies as an example	not yet defined
Marielle E. van Gijn	ERN RITA	University Medical Center Groningen, Dept. of Genetics	Netherlands	From high throughput sequencing to diagnosis in immune mediated disorders	not yet defined
Ana Caroline Pereira Neves	Endo-ERN	Associação Protetora dos Diabéticos de Portugal (APDP), Lisbon	Portugal	Genetics and precision medicine in rare diseases	not yet defined
David Gómez Andrés	ERN-RND	Hospital Universitari Vall d'Hebron, Dept. Of Child Neurology, Barcelona	Spain	Challenges and barriers for gene therapies in rare neurological disorders: preparing the next generation of clinicians and researchers.	Sept. / Oct.2022

Table 2. Selected workshops from both calls launched in 2021.

Workshops that are currently open for registration can be found here: <https://www.ejprarediseases.org/our-actions-and-services/training-and-education/ern-workshops/>

When informing the selected workshop organizers about the attributed funding, the coordination offices at EKUT and LUMC are providing them with a detailed process description for the organization, containing all the steps going beyond the pure organization and including the reporting and

reimbursement of the costs. The individual steps are linked to a responsible person/team who will carry out the work and can either be the workshop organizer, the project managers of the WP17 coordination team or the EJP RD coordination/communication team. Furthermore, the workshop organizer is receiving several templates (e.g. participants' registration form, website content templates, accreditation templates, satisfaction survey, etc.) who should be filled or adapted to the individual workshop. This process allows a uniform treatment of the ERN Workshops. In order to foster the most efficient collaborative work, all the documents are saved in a shared folder on MS Teams, to which all the three above mentioned parties have an access.

The EJP RD communication team is announcing the opening of the workshops through the EJP RD website and is preparing visual materials for communication via social media channels. Furthermore, the coordination offices of WP17 at EKUT and LUMC are disseminating the opening of the workshops through the ERN specific channels, so that all 24 ERNs can include the bespoke training event in their newsletter/events.

The organizers of the selected workshops that haven't taken place yet are planning to carry out their workshops in accordance with the pandemic situation and the related security measures.

### 1.1.1. Completed workshops

In the year 3 of the EJP RD project (2021), 4 workshops have been successfully conducted. These are summarized in Table 3 (Reports of the completed ERN workshops are available in the internal EJPRD database).

	Name Applicant	ERN	Institution	Country	Workshop Title	Date	Format
Call 1	Joanne Lee, Victoria Headley	ERN EURO-NMD	Newcastle University	UK	Launching Advisory Committees for Therapeutics (ACT) in rare diseases	7-8/07/2021	Online
	Willemijn M. Klein,	VASCERN	Radboud University Medical Center Nijmegen	NL	Intranodal Magnetic Resonance Lymphangiography Studies Workshop.	9-10/09/2021	Online
	Olaf Dekkers	EndoERN	Leiden University Medical Center	NL	Methods to study rare diseases, their causes and treatments: The Basics & Beyond.	30/09-01/10/2021	In person
Call 3	Charlotte Niemeyer	ERN PaedCan	Department of Pediatrics and Adolescent Medicine, University Hospital Freiburg	DE	GATA2-related Myelodysplastic Syndromes	30/09-01/10/2021	Hybrid

Table 3. Successfully conducted workshops in year 3.

In order to monitor the achievement of the defined objective “To deliver research training activities in form of research mobility and 2 days workshops with cross-ERN value” WP17 has set as indicator: ‘Evaluation results of the implemented training measure’. This indicator is measured by sending a satisfaction survey to the workshop participants containing questions on the quality and impact of the workshop contents. The defined target for the quality is 70% of the workshop participants evaluated the workshop very good or excellent. The defined target for the impact is: 70% of the workshop participants evaluated the workshop as highly useful and implement the content/methods learned at least weekly

The analysis of the satisfaction surveys from the four conducted workshops is presented in the Table 4.

Workshop	Rating of the quality of the workshops		Achievement of the target/threshold for the performance indicator ‘quality’ in %:	Rating of the usefulness of the workshop contents for the participant’s future research/professional activity		Achievement of the target/threshold for the performance indicator ‘impact’ in %	Nr. of responses
Advances in diagnostic and therapeutic methods for lymph flow abnormalities_RUMC	11	excellent*	100%	18	highly useful**	100%	20
	9	very good		2	useful		
Launching Advisory Committees for Therapeutics (ACT) in rare diseases_UNEW	11	excellent	100%	13	highly useful	100%	13
	2	very good		0	useful		
GATA2-related Myelodysplastic Syndromes_UK-FR	7	excellent	100%	8	highly useful	100%	9
	2	very good		1	useful		
Clinical Research: The Basics & Beyond_LUMC	7	excellent	93%	11	highly useful	100%	15
	7	very good		4	useful		
	1	satisfactory		/	/		

\*Selecting options: excellent, very good, good, satisfactory, poor, unacceptable.

\*\*Selecting options: highly useful, useful, not at all useful.

Table 4. Analysis of the quality and impact of the ERN Workshops.

## 1.2. Calls for fellowships for research mobility

In year 3, one call for fellowships was completed and another one has been launched and come up to the remote evaluation step by SEC at the end of the year.

	Opening period	Nr. of eligible proposals received	Date Final SEC evaluation	Nr. of selected applications	Last time point to conduct the Fellowship
3. Call Fellowships	15.03. - 26.04.2021	24	01.06.2021	18	15.12.2022
4. Call Fellowships	18.10.- 28.11.2021	20	21. 01 & 04.02.2022	/	18 months after application approval

In comparison with the first two calls, in calls 3 and 4, more flexible eligibility criteria were applied. The aim to make the programme more attractive and train more fellows in RD research from a wider geographical area has been achieved through the following criteria adaptation.

The changes are highlighted in blue. Eligible applicants can be:

- PhD students with a **minimum of one year of research experience** at the time of submitting a proposal
- **Postdocs/research fellows within five years after receiving PhD degree** at the time of submitting the proposal
- Physicians with **at least one year of rare disease specialist training and within five years of completion of clinical training** at the time of submitting the proposal

Furthermore,

- **Either home or host (secondment) institution must be a Full Member or Affiliated Partner of an ERN** at the time when the application is submitted, as well as during the proposed period of the training stay.
- **Both the home and host institutions must be located in EU countries and/or in countries in which EJP RD beneficiaries are located**, which are: Austria, Armenia, Belgium, Bulgaria, Croatia, Czech Republic, Denmark, Estonia, Finland, France, Germany, Georgia, Greece, Hungary, Ireland, Israel, Italy, Netherlands, Latvia, Lithuania, Luxembourg, Malta, Norway, Poland, Portugal, Romania, Serbia, Spain, Sweden, Slovakia, Slovenia, Switzerland, Turkey and UK.
- Duration of training stay is 4 weeks to **6 months**
- **Residency of the applicant must be in a country involved in EJP RD (see above)**

Table 5 gives an overview of selected fellowships from the third call.

Name	ERN	Home Institution	Host institution	Project Title
Francesco Missale	EUROCAN	University of Brescia, Italy (non-ERN)	Netherlands Cancer Institute - Antoni van Leeuwenhoek, Amsterdam, the Netherlands	Salivary gland tumors management and external validation of novel N category staging systems for malignant major salivary gland tumors
Rebecca Pulvirenti	ERNICA	University Hospital of Padua, Italy	Erasmus Medical Center – Sophia Children’s Hospital, Rotterdam, the Netherlands	Towards a blueprint for transitional care in patients with congenital anatomical anomalies (CAA)
Oana-Ofelia Dănilă	endo-ERN	"Carol Davila" University of Medicine and Pharmacy Bucharest, Romania (non ERN)	Leiden University Medical Center, the Netherlands	The influence of Growth Hormone excess secretion on the evolution of Fibrous Dysplasia in patients
Beste Ozsezen	LUNG	Hacettepe University School of Medicine Ankara, Turkey (non-ERN)	"Anna Meyer" Pediatric University-Hospital, Florence, Italy	Lung function in a cohort of teenagers born very preterm
Eszter Lévai	ERKNet	Semmelweis University, Budapest, Hungary	Heidelberg University Hospital, Germany	The role of PARK7 in mesothelial and vascular endothelial barrier integrity and function in peritoneal dialysis
Dmitrijs Rots	ERN ITHACA	Radboud University Medical Center, Nijmegen, the Netherlands	Manchester University Hospitals NHS Trust, UK (non-ERN)	Role of non-Mendelian genetics in Mendelian neurodevelopmental disorders
Jeroen Kerstens	ERN EURO-NMD	University Hospital Antwerp, Belgium	Erasmus MC University Medical Center, Rotterdam, the Netherlands RITA	Neuronal autoantibodies in patients with unexplained ataxia and/or parkinsonism

Ana Filipa Duarte	ERN CRANIO	Erasmus Medical Center, Rotterdam, the Netherlands	King's College London, UK	The FUZ gene in craniosynostosis: from a mouse knock-out model to understanding its contribution in human suture closure
Elke de Boer	ERN ITHACA	Radboud University Medical Center, Nijmegen, Netherlands	University of Burgundy, Dijon, France	Integrative analysis of NGS and transcriptome data to elucidate the genetic etiology of unexplained cases of ERN ITHACA
Annalisa Allegorico	ERN LUNG	Bambino Gesù Children's Hospital, Rome, Italy	University Hospital Muenster, Germany	Immunofluorescence Technique to learn and spread knowledge in Italy
Josias Bastian Grogg	eUROGEN	The Christie NHS Foundation Trust, ManchesterUK (non-ERN)	Universitätsklinikum Hamburg-Eppendorf, Germany; University Hospital Centre Zagreb, Croatia; University College London Hospitals NHS Foundation Trust, UK; Antoni van Leeuwenhoek hospital, Amsterdam, the Netherlands	The OrphAn-Testis-Histology registry (OATH)
Marija Miletic	eUROGEN	University Hospital Centre Sestre milosrdnice, Zagreb, Croatia	UZ Leuven, Belgium	Treatment patterns and real-world evidence for patients with penile cancer in specialized healthcare providers within European Reference Network on urogenital diseases and conditions (ERN eUROGEN) network

Giada De Lazzari	ERNICA	University of Padova, Italy	INSERM - Toulouse, France (non-ERN)	Patient-derived intestinal 3D model for studying rare disorders: the Tricho-Hepato-Enteric Syndrome paradigm (TH3S)
Eva Coopmans	endo-ERN	Leiden University Medical center, the Netherlands	Barts Cancer Institute London and The William Harvey Research Institute (WHRI) London, UK (non-ERN)	GH-secreting pituitary tumours: nup therapy resistance in the bud (pilot-study)
Benjamin Chaigne	ERN reCONNET /RITA	Centre de Référence Maladies Auto-immunes et Maladies Systémiques Rares, Hôpital Cochin, Université de Paris France	Royal Free Hospital Campus - University College of London RITA	Integrated proteomic analysis of blood B cells and skin fibroblasts in systemic sclerosis: toward proteomic B/F signatures of SSc patients.

Table 5. Overview of selected fellowships from Call 3.

### 1.2.1. Completed research mobilities

Out of the 33 fellowships selected for funding over the three calls, 6 have completed their fellowship in 2021 (see Table 6). Some of the others could not start their secondment due to the restrictions related to the COVID pandemics. Almost all of them already delivered their surveys and reports shortly after completing the fellowship, but some forms will follow and are hence not yet included in this document. In the fourth call for fellowships 2021/2022 there were 20 eligible applications. They ran through the first remote evaluation step of the evaluation committee (SEC). The first SEC evaluation meeting took place on 21<sup>st</sup> January 2022 and the second and final one on February 4<sup>th</sup>.

Call	Name	ERN	Home Institution	Host institution	Project Title	Duration of secondment	Status
Call 2	Carlo Wilke	ERN-RND	University Hospital Tübingen, Germany	Radboud University Medical Center, Nijmegen, the Netherlands	Beyond family-based exome analysis: advanced cohort-level bioinformatic approaches for assessing genetic burden in rare neurodegenerative diseases	01.01.2021 - 31.03.2021 (3 months)	completed
	Nuria Duenas Cid	ERN GENTURIS	Catalan Institute of Oncology, Barcelona, Spain	University Hospital Bonn, Germany	Colorectal cancer risk personalization using polygenic risk score analysis in Lynch syndrome individuals	06.06.2021- 24.07.2021 (7 weeks)	completed
	Laura Costa Comellas	ERN EURO-NMD	Hospital Universitari Vall d'Hebron, Barcelona Spain	Bambino Gesù' Children's Research Hospital IRCCS, Rome Italy	Inherited Myopathies: Diagnostic value of clinical deep phenotyping, imaging and muscle biopsy in the era of Next Generation Sequencing	30.08. - 30.10. 2021 (2 months)	completed
	Agne Bubilaite	ERN Skin	Hospital of Lithuanian University of Health Sciences Kaunas Clinics, Lithuania	University of Lübeck, Germany	Management of autoimmune blistering diseases associated with mucous involvement and malignancies based on gained	01.10.2021- 01.11.2021 (1 month)	completed
	Silvia Vallengia	ERN CRANIO	University of Padova, Italy	Erasmus MC, Rotterdam, the Netherlands	Black-bone MRI evaluation of cranial vault and temporal bone in craniosynostoses	04.09.- 30.11.2021 (3 months)	completed
	Vaiva Jariene	ERN Skin	Hospital of Lithuanian University of health sciences, Kaunas, Lithuania	Städtisches Klinikum Dessau, Brandenburg Medical School Theodor Fontane, Germany	Application of ex vivo co-culture systems for studying molecular drug Effectiveness on hidradenitis suppurativa	03.10.- 30.11.2021 (2 months)	completed

Call 3	Alexander Maximilian Bernhardt	ERN-RND	LMU Munich university hospital, Germany	IRCCS, Istituto delle Scienze Neurologiche di Bologna, Italy	Advancing RT-QuIC for the diagnosis of rare neurodegenerative diseases	16.08. - 8.10.2021 (8 weeks)	completed
	Caroline Merckx	ERN EURO-NMD	Ghent University Hospital, Belgium	University Medical Center Göttingen, Germany	Evaluating the therapeutic potential of osmolytes in an in vitro model for Duchenne Muscular Dystrophy	24.10. - 12.11. & 29.11.- 17.12.2021 (6 weeks)	completed

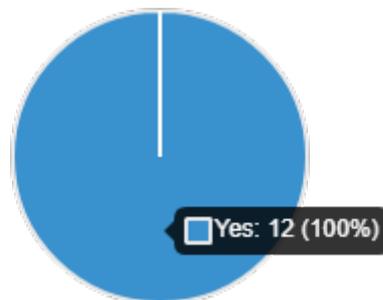
Table 6. Overview of completed fellowships from Call 2 and Call 3.

From the satisfaction and the report survey that the fellows have filled after the completion of the secondment, WP17 could assess the quality (1.) and the impact (2.) of the funded training:

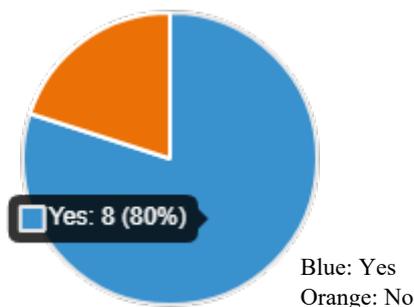
1. 91% of the fellows evaluated the research training stay at the host institution very good or excellent.
2. 91% of the fellows evaluated the research training stay at the host institution as highly useful and were going to implement the content/methods learned at least weekly.

Some selected outcomes from the questionnaires are listed here:

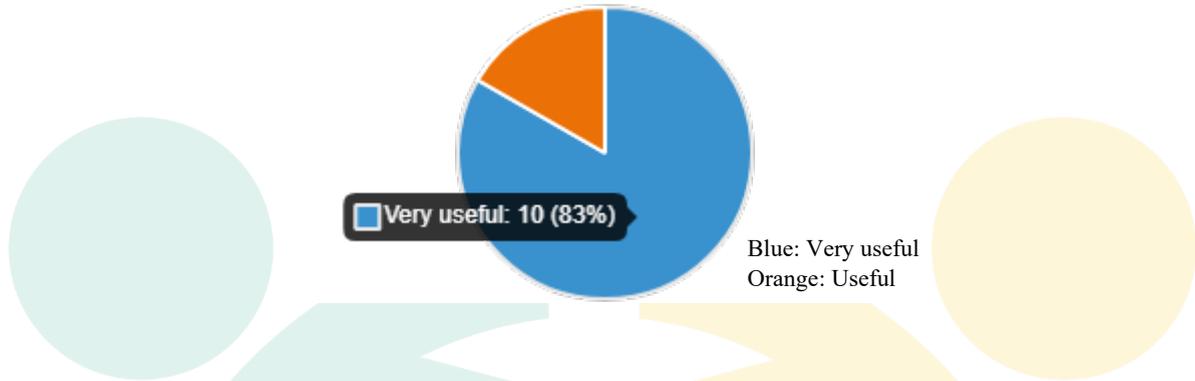
**Q1: Have you learned what you intended to learn?**



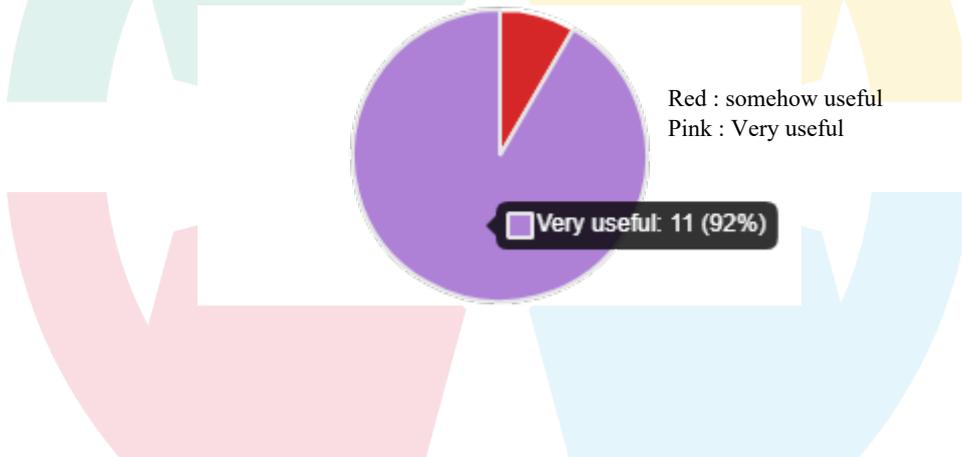
**Q2: Do you anticipate to use the contents learned during your stay regularly, i.e. at least weekly**



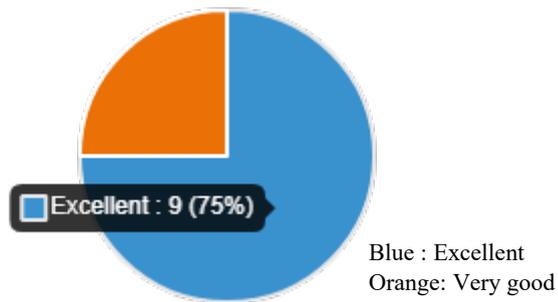
**Q3: Please rate the usefulness of your stay for your future research**



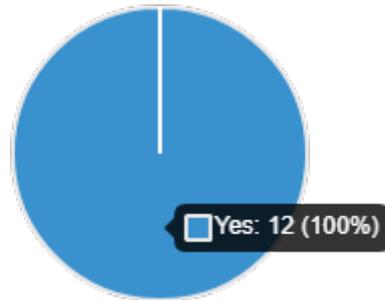
**Q4: How useful was the exchange for your daily work and career?**



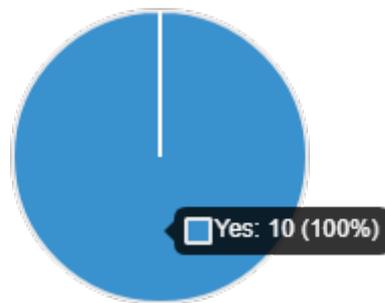
**Q5: Please rate the quality of your stay at the host institution**



**Q6: Did the guidance by the supervisor meet your expectations?**



**Q7: Will this fellowship be of profit for the host and home institution?**



**Q8: Is there anything you would like to share regarding your experience with the fellowship exchange programme?**

Q8: Is there anything else you would like to share regarding your experience with the fellowship exchange program?
I would recommend it to all young researchers
My exchange was a great experience for me - I learnt a lot, met great people and challenged myself. I am grateful from the bottom of my heart to all that enabled my fellowship.
I would again like to thank my supervisor and the host research group for the open welcome and the excellent support, and the EJP-RD Research Mobility Fellowship team for the kind and reliable support.
Throughout my stay at the UKB I have not only felt accompanied on a professional level, but also on a personal level. Both Dr. Aretz and everyone on his team have been very attentive to me and have made me feel at home.
It has been a wonderful experience that I would recommend to any young researcher.
I am so grateful for the chance to participate in a fellowship exchange program, I think it helps to build bridges between centers.
Excellent fellowship exchange program!
The fellowship exchange program has been a unique and extremely positive personal and professional experience. I want to thank and congratulate all the people who make up the project and

have allowed me to enjoy it. The entire process has been easy, fast and comfortable, from start to finish.

The exchange in the COVID pandemic has been a handicap for me, above all logistically and sometimes emotionally. Being away from my family has been difficult at times. But I definitely recommend it 100%

It was a wonderful experience.

I strongly recommend it, but I suggest a longer duration of the fellowship (at least 6 months).

It's a great opportunity! In the future, I think it might be better to add an extra week since it was a very intense fellowship.

Their complete feedback is summarized [in this table](#).

## 2. Establishing an accreditation process for training activities

WP17 has identified the European Accreditation Council for Continuing Medical Education (EACCME) an institution of the UEMS (Union of the European Medical Specialists) as body for the accreditation of the ERN workshops (<https://eaccme.uems.eu/home.aspx>).

The workshop coordination office based at EKUT applied for the first pilot accreditations of two selected workshops: 1. “Contemporary outcome measures in NeuroMuscular diseases, Nice, France” at the end of December 2021 and 2. “Translational research on bone impairment in rare diseases, Lyon, France” in February 2022. The delays for applications of LEEs have been changed from 7 weeks prior to the date of the LEE (valid until the end of 2021) to 12 weeks (valid from 2022). The application process requests information on educational needs, set educational outcome, teaching methods, funding sources, detailed information of the organizing institution, 2 members of the scientific evaluation committee and a fee of 175€ for <100 participants per LEE.

Furthermore, when applying one has to commit to provide EACCME a report on the participants' feedback 4 weeks after the event has taken place. For this purpose, WP17 will use the satisfaction survey form already established for the workshop report and insert minor changes according to the EACCME evaluation form.

At the moment of writing this deliverable the first workshop has successfully received the accreditation. The second workshop is still in a review phase by EACCME.

Since there is no accreditation for medical specialists that is recognized in all EU countries, from where the workshop participants come from, it remains for year 4 to evaluate the necessity and usefulness of the accreditation process.