EJP RD monitoring of Networking Support Scheme

2. General Information

1. Personal Information

Principal Applicant Name

Last name : Smeets
First name : Bert

Country

The Netherlands

Institution

Maastricht University

Email Address

bert.smeets@maastrichtuniversity.nl

Title of the event

LAMA2 Muscular Dystrophy: Paving the road to therapy

Date(s) of the event

March 17-19, 2023

Location of the event

Spain

2. What is your field of research?

Musculoskeletal diseases

3. Was your meeting

Hybrid

4. Was your meeting a satellite event?

No

5. Was your event co-funded by another funding source?

Yes

6. Please estimate the number of participants in your event:

100-250

7. Have you applied for an EJP RD Networking Grant in the past?

No

8. Are the meeting participants part of an existing EJP RD funded or E-Rare funded consortium?

No

9. What is the acronym of your EJP RD or E-Rare funded project?

No acronym

10. Within the applicants to the current networking grant, how many collaborated in the past?

2 out of 7

11. List of applicants

	Name	Gender	Country	Type of partner	Early Career Scientist	
					Yes	No
Principal Applicant	Smeets, Bert	Male	The Netherlands	Academic+partner		X
Co-applicant 1	Verbrugge, Bram	Male	The Netherlands	Patient organisation		X
Co-applicant 2	Rüegg, Markus A.	Male	Switzerland	Academic+partner		X
Co-applicant 3	Durbeej, Madeleine	Female	Sweden	Academic+partner		X
Co-applicant 4	Stępniewski, Jacek	Male	Poland	Academic+partner		X
Co-applicant 5	Topaglu, Haluk	Male	Turkey	Clinical+department		X
Co-applicant 6	Sarkozy, Anna	Female	United Kingdom	Clinical+department		X
Co-applicant 7	Allamand, Valérie	Female	France	Academic+partner		X
Co-applicant 8						
Co-applicant 9						

12. How would you describe the main added value of your networking event?

Integration into a European network
Bringing together multidisciplinary academic expertise
Formation of new collaborations
Links to clinical partnerships
Links to patient representatives/organization partnerships

3. Project Outcomes: Economic and Health Impact

13. What was the main focus/aim of your meeting?

Results of therapeutic research
Technologies (cellular models, animal models, biomarkers, -omics, etc.)
Initiating/improving/using patient registries
Results of diagnostic research (including genetic, epigenetic and patho-physiological studies)

14. Were participants from industry attending this meeting?

No

15. Did your event involve a patient representative/organization?

Yes

16. Please estimate how many patients/patient representatives participated in the meeting

150

17. What was the role(s) of the patient representative(s)/organization(s)?

Co-applicant
Active involvement in the discussions during the meeting
Serving as members of the meeting advisory or steering committee
Commenting and developing patient information leaflets or other (research) materials
Raising awareness of the event through media such as television programmes, newspapers and social media
Dissemination to patient organizations and the patient community on the outcomes of the event

18. How much did the involvement/engagement of patients/ patient organizations contribute to the meeting?

7

19. Did your event involve an Early Career Scientist (ECS)?

Yes

20. Please estimate how many ECS participated in the meeting

15

21. What was the role(s) of the ECS(s)?

Active involvement in the discussions during the meeting
Other (please specify): Organisation of the event
Raising awareness of the event through media such as television programmes, newspapers and social media
Dissemination on the outcomes of the event

22. How much did the involvement/engagement of ECS contribute to the meeting?

6

23. Did your event involve an stakeholder(s) from underrepresented countries (URCs)?

Yes

24. Please estimate how many participants from URCs participated in the meeting

20

25. What was the role(s) of these participants?

Co-applicant
Active involvement in the discussions during the meeting
Serving as members of the meeting advisory or steering committee
Raising awareness of the event through media such as television programmes, newspapers and social media
Dissemination on the outcomes of the event

26. How much did the involvement/engagement of participants from URCs contribute to the meeting?

6

4. Communication and Dissemination of the Event and its Results

27. Was the event publicized by yourself?

Yes

28. By what means was the event advertized?

Website
Social media
E-Mailing list

29. Were outcomes of the event publicized?

Yes

30. By what means were the outcomes of the event made public?

Website
Social media
Newsletter(s)
E-Mailing list
Other:: scientific report (to be finalised)

5. Collaborations, Sustainability and Ethics

31. Were new collaborations established during the event?

Yes

32. Please provide details

At the final day of the meeting, 3 groups were formed, one focusing on natural history/trial readiness, one on therapy development and one on patient networks. All these groups defined an action plan with concrete objectives to achieve on these topics. The principal applicant will take the lead in organising this by planning regular meetings and by facilitating exchange of research material and data.

33. Are you planning a follow-up meeting/teleconference with your network following the event?

Yes

34. Was the meeting organized in compliance with the ethical requirements, such as personal data management rules?

Yes

6. Narrative Report

35. Public Summary of the meeting

Title:

LAMA2 Muscular Dystrophy: Paving the road to therapy

General objectives:

LAMA2-MD is an ultra-rare, clinically heterogeneous disease, of which the diagnosis is difficult and many patients still need to be identified. The clinical history is only partially known and, despite intense research, a cure is not available. Also, existing treatments to preserve quality of life do not reach all patients and clinicians. The objective of the networking event is to address these issues and make further steps on the road to therapy for LAMA2-MD by: 1. Aligning patient registries and natural history studies and define biomarkers and guidelines which can be used in patient care; 2. Defining diagnostic criteria and guidelines for alleviating symptoms and maintaining quality of life with a dissemination plan for Europe; 3. Listing available patient samples for research, sharing confidential research progress and establishing a LAMA2-MD therapy task-force.

Main outcomes:

Objective 1: Ongoing and planned natural history studies from 9 European and other countries were presented. The necessity of conducting these studies in LAMA2-MD was emphasised, as these are required to establish trial readiness. To collect the necessary information, the importance of developing standardised registries was acknowledged. A clinical work group was formed, which will address this and is going to meet online regularly with patient representatives to allow a smooth transfer to the patients and regular care. A European initiative, for which funding will be requested, will further align and standardise these studies .
Objective 2: Patient organisations and patient representatives from 9 countries actively participated in the meeting, with many more from other countries online. A European LAMA2 community was established, which will be the umbrella organisation for the national organisation and for patients without national organisations. Concrete plans were made for facilitating potential trials within Europe, supporting and co-funding international research, and, primarily, informing patients and families of the latest research progress and clinical guidelines.
Objective 3: The key groups working on therapy development presented their work and exciting progress. Approaches were largely complementary, allowing open collaborations and stimulating joint initiatives. A therapy working group is formed, which will have regular research meetings. In addition, a scientific resource is being established, allowing a rapid exchange of biological samples, biomaterials, experimental models and protocols to shorten the road to therapy.

7. Feedback

36. Please indicate your degree of satisfaction from the EJP RD networking scheme

Very Satisfied

37. Would you recommend the EJP RD Networking Support Scheme to colleagues?

Yes

38. Will you consider applying to the Networking Support Scheme again?

Yes

39. To what extent would you agree with the following statements about key factors that may have affected the organization of your event under the EJP RD Networking Support Scheme?

	Strongly agree	Agree	Disagree	Strongly disagree
The administrative burden from the EJP RD Networking Support Scheme was **not** excessive		X		
The resources available through this scheme were adequate			X	
There was good quality interactions with the other co-applicants	X			

40. What have been the main outcomes of the meeting?

	Major outcome	Moderate outcome	Minor outcome	Not applicable
Formation of new collaborations	X			
Increased research capacity	X			
Development of new method, data or technology		X		
Development of new/improved product or service				X
Development of a new technical process		X		
Better access to international network/markets	X			
Better understanding of other European cultures/issues		X		
Enhanced research network to compete for future European project funding		X		
Better collaboration with PAO(s)	X			
Better collaborations with underrepresented countries		X		

41. Please share concerns or problems that you may have encountered

Not applicable

42. Do you have suggestions for improving the EJP RD Networking Support Scheme?

Increasing the budget for these meeting. We were not able to cover all the costs of the participants. For professional participants, this is usually not a big issue, but for patient representatives, especially from underrepresented countries, it is. For the patient representatives these were, luckily, covered by the patient organizations. A budget of 50-60k€ should allow covering all costs.