Supplementary material

Table S1: List of the MetabERN centres and participation to the survey

| | | Number of answers | | s | |
|---------|--|-------------------|-------|------|-------|
| Country | MetabERN centres | Clin. | Univ. | Lab. | Total |
| | CHU Liège | 1 | 3 | 2 | 6 |
| | Cliniques universitaires St Luc-Univ. catholique Louvain | 0 | 1 | 0 | 1 |
| BE | Ghent University hospital | 0 | 0 | 0 | 0 |
| | Metabolic Center ULB-VUB | 2 | 2 | 2 | 6 |
| | University Hospital of Antwerp UZA | 1 | 1 | 1 | 3 |
| | UZ Leuven | 2 | 0 | 1 | 3 |
| BU | University hospital "Alexandrovska"in Sofia | 0 | 0 | 0 | 0 |
| CR | University Hospital Center Zagreb | 0 | 0 | 1 | 1 |
| CZ | General University Hospital in Prague-GUH | 1 | 1 | 1 | 3 |
| DK | Copenhagen University Hospital, Rigshospitalet | 1 | 0 | 0 | 1 |
| | Hôpitaux Universitaires Paris IIe de France Ouest | 0 | 0 | 0 | 0 |
| | Hôpitaux Universitaires Paris Nord Beaujon | 1 | 2 | 0 | 3 |
| | Hôpitaux Universitaires Lyon | 0 | 0 | 0 | 0 |
| | Hôpitaux Universitaires Paris Nord Antoine-Béclère | 1 | 1 | 0 | 2 |
| FR | Hôpitaux Universitaires Paris Nord Louis-Mourier | 1 | 1 | 1 | 3 |
| | Hôpitaux Universitaires Paris Necker-Enfants Malades | 0 | 1 | 0 | 1 |
| | Hôpitaux Universitaires Paris Robert-Debré | 0 | 0 | 1 | 1 |
| | Hôpitaux Universitaires Lille | 1 | 1 | 1 | 3 |
| | Hôpitaux Universitaires Marseille | 0 | 0 | 0 | 0 |
| | Helios Dr. Horst Schmidt Kliniken | 0 | 0 | 1 | 1 |
| | Charité-Universitätsmedizin Berlin | 1 | 1 | 0 | 2 |
| DE | Hannover Medical School | 0 | 0 | 0 | 0 |
| | Medical Center University of Freiburg | 1 | 2 | 2 | 5 |
| | Muenster University Hospital | 0 | 0 | 1 | 1 |

| | Otto-von-Guericke-University Magdeburg | 0 | 0 | 0 | 0 |
|----|--|---|---|---|---|
| | Universitätskinderklinik Giessen | | 2 | 1 | 4 |
| | Universitätsklinikum Heidelberg | | 1 | 1 | 3 |
| | University Medical Center Hamburg-Eppendorf | 1 | 1 | 1 | 3 |
| | University Medical Center Mainz | 2 | 1 | 1 | 4 |
| HU | Hungary-University of Debrecen | 1 | 1 | 1 | 3 |
| | Azienda Sanitaria Universitaria Integrata di Udine | 2 | 1 | 1 | 4 |
| | Meyer Children's Hospital Firenze | 0 | 0 | 0 | 0 |
| | ASST Monza San Gerardo Hospital | 1 | 2 | 0 | 3 |
| | Azienda Ospedaliera Universitaria "Federico II", Napoli | 1 | 1 | 2 | 4 |
| | Azienda Ospedaliera Universitaria Integrata di Verona | 0 | 0 | 0 | 0 |
| IT | Ospedale Pediatrico Bambino Gesù, Roma | 0 | 0 | 0 | 0 |
| | Giannina Gaslini Institute, Genova | | 0 | 1 | 2 |
| | San Paolo Hospital, ASST Santi Paolo e Carlo, Milano | 1 | 1 | 1 | 3 |
| | University Hospital of Padova Azienda Ospedaliero-Universitaria Pisana | | 1 | 0 | 2 |
| | | | 1 | 0 | 1 |
| | Azienda Ospedaliera Universitaria Senese | | 2 | 2 | 6 |
| LI | Vilnius University Hospital Santariskiu klinikos | 0 | 0 | 0 | 0 |
| | Erasmus MC, University Medical Center, Rotterdam | 2 | 1 | 0 | 3 |
| | Maastricht University Medical Center | 0 | 0 | 1 | 1 |
| NL | University Medical Center of Groningen | 2 | 1 | 0 | 3 |
| | University Medical Center Utrecht (UMCU) | 1 | 1 | 1 | 3 |
| | Academic Medical Center Amsterdam | 1 | 0 | 1 | 2 |
| NO | Oslo University Hospital | 1 | 2 | 1 | 4 |
| | Helse Bergen HF, Haukeland University Hospital | 0 | 0 | 0 | 0 |
| PL | Krakow University Hospital | 1 | 1 | 4 | 6 |
| | Centro Hospitalar Lisboa Norte, EPE | 1 | 1 | 1 | 3 |
| PO | Centro Hospitalar do Porto, EPE (CHP) | 1 | 1 | 0 | 2 |
| | Centro Hospitalar do Sao Joao, E.P.E. | 1 | 1 | 2 | 4 |

| | Centro Hospitalar e Universitário de Coimbra, E.P.E. | 2 | 1 | 1 | 4 |
|----|---|----|----|----|-----|
| | Hospital Senhora da Oliveira – Guimarães | 0 | 0 | 0 | 0 |
| SO | University Medical Centre Ljubljana | 1 | 1 | 1 | 3 |
| | University Clinical Hospital of Santiago de Compostela. | 1 | 0 | 0 | 1 |
| | Hospital Sant Joan de Déu, Barcelona | 1 | 2 | 0 | 3 |
| SP | Hospital Universitario Cruces, Vizcaya | 1 | 1 | 1 | 3 |
| | Hospital Universitario Vall d'Hebron, Barcelona | 1 | 0 | 0 | 1 |
| | Hospital Universitario "12 de Octubre" Madrid | 1 | 2 | 1 | 4 |
| SW | Karolinska University Hospital, Solna | 2 | 1 | 0 | 3 |
| | Sahlgrenska University Hospital University of Gothenburg | 0 | 1 | 1 | 2 |
| | Cambridge University Hospitals | 0 | 0 | 0 | 0 |
| | Birmingham Children's Hospital | 1 | 2 | 1 | 4 |
| UK | University Hospitals Bristol, Bristol Children's Hospital | 0 | 0 | 0 | 0 |
| | Central Manchester University Hospitals | | 0 | 0 | 0 |
| | Great Ormond Street Hospital London | 0 | 0 | 0 | 0 |
| | University Hospital of Birmingham | 1 | 0 | 0 | 1 |
| | | 52 | 52 | 44 | 148 |



Survey MetabERN for Departments

| *1. Can you please type in your contact details | |
|--|---|
| Name | |
| Email Address | |
| Phone Number | |
| *2. Please select the name of your hospital /institute from the drop down menu | |
| | ▼ |
| | |
| | |

| 3. Please select what is applicable to your situation output male female |
|---|
| 4. Name of the higher education institution (in original language) |
| |
| 5. Faculty/ department/ institute (in English) |
| |
| 6. Name of the faculty/ department/ institute (in original language) |
| ▼ |
| 7. Type of institution Public university Private university |
| |

| Public college Private college Research institute |
|---|
| *8. What is the legal status of your institution Public Private mixed |
| *9. What is your position |
| C Associate Professor C Lecturer Senior Research Fellow Postdoc/Research Fellow Research Assistant Teaching Assistant Dean, head of department or other senior administration position Other (please specify) |

| *10. Please select the areas and fields in which your institution is undertaking research |
|---|
| activities, please select all the ones of interest: |
| Animal research |
| Basic research |
| Preclinical research |
| Clinical research |
| |
| y |

*11. Please indicate which are, the main research activities performed inside your Institution

| | No activity | Low activity | Rather low activity | Rather high activity | High activity |
|--|-------------|--------------|---------------------|----------------------|---------------|
| Health Systems Research | 0 | 0 | 0 | 0 | 0 |
| Human and Social Science Research | 0 | 0 | 0 | 0 | 0 |
| Epidemiology/Natural History of diseases | 0 | 0 | 0 | 0 | 0 |
| Research in Genetics and Pathophysiology | 0 | 0 | 0 | 0 | 0 |
| Pre-Clinical Therapeutic Research | 0 | 0 | 0 | | 0 |
| Therapeutic Research | 0 | 0 | 0 | C | 0 |
| Research Platforms/Infrastructures | 0 | 0 | 0 | 0 | 0 |
| Development and availability of tools needed to accelerate research on rare diseases | 0 | 0 | 0 | 0 | 0 |

*12. **Academic** Health Systems Research

| | No activity | Low activity | Rather low activity | Rather high activity | High activity |
|--|-------------|--------------|---------------------|----------------------|---------------|
| Research policy (e.g., guidelines development; national plans; health care resources) | c | c | c | c | 0 |
| Cost analysis (e.g., guidelines implementation) | 0 | 0 | 0 | 0 | 0 |
| New technologies (e.g., New diagnostic tests, population screening programmes, Innovative therapeutics) | 0 | С | С | 0 | c |
| Health-Technology assessment of Orphan Drugs | 0 | 0 | 0 | 0 | 0 |
| e-Health activities | C | С | С | C | С |

*13. Industrial Health Systems Research

| | No activity | Low activity | Rather low activity | Rather high activity | High activity |
|--|-------------|--------------|---------------------|----------------------|---------------|
| Research policy (e.g., guidelines development; national plans; health care resources) | С | С | С | С | 0 |
| Cost analysis (e.g., guidelines implementation) | 0 | 0 | 0 | 0 | 0 |
| New technologies (e.g., New diagnostic tests, population screening programmes, Innovative therapeutics) | 0 | 0 | 0 | 0 | 0 |
| Health-Technology assessment of Orphan Drugs | 0 | 0 | 0 | 0 | 0 |
| e-Health activities | 0 | 0 | 0 | 0 | 0 |

*14. Human and social science research

| | No activity | Low activity | Rather low activity | Rather high activity | High activity |
|---|-------------|--------------|---------------------|----------------------|---------------|
| Society and RD (e.g., social perception, accessibility to care,) | С | С | С | С | 0 |
| RD, Research and Innovation (e.g., public/private cooperation, role of patients org,) | 0 | 0 | 0 | 0 | О |
| Care practices (self- management, health education policy,) | 0 | 0 | 0 | 0 | С |
| Patients awareness (e.g., understanding genetic data, importance of animal models,) | 0 | 0 | 0 | 0 | С |
| Support for caregivers | 0 | 0 | 0 | 0 | 0 |
| Public Heath and RD (e.g., health care policies, prospective public policies,) | 0 | 0 | 0 | 0 | О |
| Methods for social support to improve quality of life | 0 | 0 | 0 | 0 | 0 |
| Services accessibility | 0 | 0 | 0 | 0 | 0 |
| Orphan Drugs availability | 0 | 0 | 0 | 0 | 0 |
| Methods to assess QoL | 0 | 0 | 0 | 0 | 0 |

^{*15.} Epidemiology & Natural History of diseases

| | No activity | Low activity | Rather low activity | Rather high activity | High activity |
|--|-------------|--------------|---------------------|----------------------|---------------|
| Registries | 0 | 0 | 0 | 0 | 0 |
| Databases/data management systems for epidemiology studies | 0 | 0 | 0 | 0 | 0 |
| Cohorts | 0 | 0 | 0 | 0 | 0 |
| Biomarkers to assess diagnosis, prognosis and severity of diseases | 0 | 0 | 0 | 0 | 0 |
| Diagnosis/prognosis methods (e.g., other than biomarkers) | 0 | 0 | 0 | 0 | 0 |
| Identification of new entities | 0 | 0 | 0 | 0 | 0 |
| Networks for ultra rare diseases | 0 | 0 | 0 | 0 | 0 |

*16. Research in genetics and physiopathology

| | No activity | Low activity | Rather low activity | Rather high activity | High activity |
|--|-------------|--------------|---------------------|----------------------|---------------|
| Gene identification and Molecular Mechanisms | 0 | 0 | 0 | 0 | 0 |
| New database method developments to be applied to omics research | О | c | c | 0 | 0 |
| Biobanking research | C | 0 | 0 | 0 | 0 |
| High throughput genomics/sequencing | C | 0 | 0 | 0 | 0 |
| New bioinformatic methods and developments | 0 | 0 | 0 | 0 | 0 |
| New technologies (e.g., atomic microscopy) | 0 | 0 | 0 | 0 | 0 |

| | No activity | Low activity | Rather low activity | Rather high activity | High activity |
|---|-------------|--------------|---------------------|----------------------|---------------|
| Omics technology (e.g., proteo, metabolo) | 0 | 0 | 0 | 0 | C |
| Development of new animal models | 0 | 0 | 0 | 0 | 0 |
| New Imaging/Microscopy methods | 0 | 0 | 0 | | 0 |
| Development of new therapeutic tools | 0 | 0 | 0 | 0 | 0 |

*17. Pre-clinical therapeutic research

| | No activity | Low activity | Rather low activity | Rather high activity | High activity |
|---|-------------|--------------|---------------------|----------------------|---------------|
| Proof-of-concept | 0 | 0 | 0 | 0 | 0 |
| Drug libraries | \circ | 0 | \circ | 0 | 0 |
| Search for active chemical compounds | 0 | 0 | 0 | 0 | 0 |
| High throughput drug screening | \circ | 0 | 0 | 0 | 0 |
| Therapeutic target identification/validation (e.g.,. stem cells, systems biology) | 0 | 0 | 0 | 0 | 0 |
| Cell therapy research (e.g., viral vectors) | \circ | 0 | 0 | 0 | 0 |
| Innovative biotechnological research (e.g., monoclonal antibodies, cell and gene therapy, enzyme therapy) | 0 | 0 | 0 | 0 | 0 |
| Drug toxicology/pharmacology | \circ | 0 | 0 | 0 | 0 |

*18. Therapeutic research

| | No activity | Low activity | Rather low activity | Rather high activity | High activity |
|--|-------------|--------------|---------------------|----------------------|---------------|
| Drug development | 0 | 0 | 0 | 0 | 0 |
| Phase I/II Clinical Trials | 0 | 0 | 0 | 0 | 0 |
| Bioproduction centres | 0 | 0 | 0 | 0 | 0 |
| Epidemiological/clinical/ therapeutical investigations | С | O | С | 0 | 0 |
| Medical devices | c | 0 | 0 | 0 | 0 |
| Assesment of surgery procedures | С | 0 | 0 | C | 0 |
| Transplants | c | C | 0 | C | 0 |
| Pharma/academic partnerships | 0 | 0 | 0 | 0 | 0 |

*19. Platforms / Infrastructures development

| | No activity | Low activity | Rather low activity | Rather high activity | High activity |
|--|-------------|--------------|---------------------|----------------------|---------------|
| Bioinformatics | 0 | 0 | C | 0 | С |
| Pharmacogenomic diagnostic assays | 0 | 0 | 0 | 0 | 0 |
| Population Registries | 0 | 0 | 0 | 0 | 0 |
| Genetically-modified animal model repositories | C | 0 | О | 0 | С |

| | No activity | Low activity | Rather low activity | Rather high activity | High activity |
|--------------------------------------|-------------|--------------|---------------------|----------------------|---------------|
| GMP platforms | 0 | 0 | 0 | 0 | 0 |
| Biobanking | 0 | 0 | 0 | 0 | 0 |
| Genomic and proteomic platforms | 0 | 0 | 0 | 0 | 0 |
| Long-term cohort of patients studies | 0 | 0 | 0 | 0 | 0 |
| High throughput drug screening | 0 | 0 | 0 | 0 | 0 |

*20. Human resources

| | No activity | Low activity | Rather low activity | Rather high activity | High activity |
|--------------------------------------|-------------|--------------|---------------------|----------------------|---------------|
| Rotational positions for clinicians | 0 | 0 | C | 0 | 0 |
| Rotational positions for researchers | 0 | 0 | 0 | 0 | 0 |
| Specific training in RD | c | 0 | С | 0 | 0 |
| Training in bioinformatics | c | О | 0 | О | О |

*21. Development and availability of tools needed to accelerate research on rare disease.

| | No activity | Low activity | Rather low activity | Rather high activity | High activity |
|--|-------------|--------------|---------------------|----------------------|---------------|
| Network development | 0 | 0 | 0 | 0 | 0 |
| Creation of transnational research consortiums | 0 | 0 | 0 | 0 | 0 |
| Transnational calls for projects funding | 0 | 0 | 0 | 0 | 0 |
| Opening of national research programmes to international collaboration | 0 | 0 | 0 | 0 | 0 |
| Transnational research infrastructures | 0 | 0 | 0 | 0 | 0 |
| Development of an inventory/directory of databases of experts | 0 | 0 | 0 | 0 | 0 |
| Transnational collection of funded RD research projects | О | 0 | 0 | C | 0 |

*22. What influences and governs research at your institution?

| | Strongly agree | Agree to some extent | Disagree to some extent | Strongly disagree | No information available | Not applicable |
|--|----------------|----------------------|-------------------------|-------------------|--------------------------------|----------------|
| Ministry/ national research agency has a strong say | 0 | 0 | 0 | 0 | 0 | 0 |
| Central university level mostly decides | 0 | 0 | 0 | 0 | 0 | 0 |
| Faculties/ departments/ institutes decide very much on what they do | 0 | 0 | 0 | 0 | 0 | 0 |
| Individual professors/ Researchers do very much what they want | 0 | 0 | 0 | 0 | 0 | С |
| In my own position, I can influence research agendas at my faculty/department/ institute | 0 | 0 | 0 | 0 | 0 | c |

*23. What is the main impact of the research activities undertaken at your faculty/department / institute?

| | high impact | impact to some extent | low to none impact | do not know | Not applicable |
|---|-------------|-----------------------|--------------------|-------------|----------------|
| Contribute to international research | 0 | | 0 | 0 | 0 |
| Contribute to national research | 0 | 0 | 0 | 0 | 0 |
| Support of local/ regional innovation of development | 0 | 0 | 0 | 0 | 0 |
| Informing teaching, to be sure that teaching is based on up-to- date research | 0 | С | 0 | C | С |
| Other | 0 | | 0 | 0 | 0 |

*24. What is the approximate number of staff in research and teaching at your faculty/department/ institute?

Full Professors

Full Professors

Full Professors total number menu

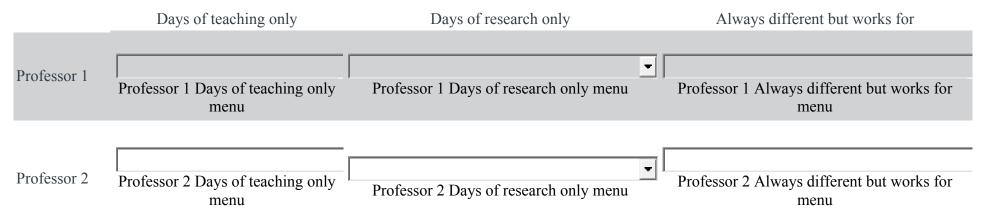
Nurses with responsibilities for research only

Nurses with responsibilities for research only total number menu

total number

| Physicians with responsibilities for research only | Physicians with responsibilities for research only total number menu |
|---|---|
| PhDs with responsibilities for research only | PhDs with responsibilities for research only total number menu |
| Nurses with responsibilities for teaching only | Nurses with responsibilities for teaching only total number menu |
| Physicians with responsibilities for teaching only | Physicians with responsibilities for teaching only total number menu |
| PhD with responsibilities for teaching only | PhD with responsibilities for teaching only total number menu |
| Nurses with responsibilities for teaching and researching | Nurses with responsibilities for teaching and researching total number menu |
| Physicians with responsibilities for teaching and researching | Physicians with responsibilities for teaching and researching total number menu |
| PhD with responsibilities for teaching and researching | PhD with responsibilities for teaching and researching total number menu |
| | |

25. What is the approximate total working days of your staff for teaching and/or research? (If you don't have Prof 2,3 etc and PhD 4,5, etc, you just fill in 0 in all columns, if you are able to fill in how many hours they teach and do research you can fill in 0 at the last column. If you are not able to specify between teaching and researching you fill in 0 in column 1 and 2 and you estimate the total working days in column 3)



| | Days of teaching only | Days of research only | Always different but works for |
|-------------|--|--|---|
| Professor 3 | Professor 3 Days of teaching only menu | Professor 3 Days of research only menu | Professor 3 Always different but works for menu |
| Professor 4 | Professor 4 Days of teaching only menu | Professor 4 Days of research only menu | Professor 4 Always different but works for menu |
| Professor 5 | Professor 5 Days of teaching only menu | Professor 5 Days of research only menu | Professor 5 Always different but works for menu |
| PhD 1 | PhD 1 Days of teaching only menu | PhD 1 Days of research only menu | PhD 1 Always different but works for menu |
| PhD 2 | PhD 2 Days of teaching only menu | PhD 2 Days of research only menu | PhD 2 Always different but works for menu |
| PhD 3 | PhD 3 Days of teaching only | PhD 3 Days of research only menu | PhD 3 Always different but works for menu |

| | Days of teaching only menu | Days of research only | Always different but works for |
|-------------|--|--|---|
| Phd 4 | Phd 4 Days of teaching only menu | Phd 4 Days of research only menu | Phd 4 Always different but works for menu |
| PhD 5 | PhD 5 Days of teaching only menu | PhD 5 Days of research only menu | PhD 5 Always different but works for menu |
| PhD 6 | PhD 6 Days of teaching only menu | PhD 6 Days of research only menu | PhD 6 Always different but works for menu |
| Physician 1 | Physician 1 Days of teaching only menu | Physician 1 Days of research only menu | Physician 1 Always different but works for menu |
| Physician 2 | Physician 2 Days of teaching only menu | Physician 2 Days of research only menu | Physician 2 Always different but works for menu |
| Physician 3 | Physician 3 Days of teaching only menu | Physician 3 Days of research only menu | Physician 3 Always different but works for menu |
| Physician 4 | Physician 4 Days of teaching only menu | Physician 4 Days of research only menu | Physician 4 Always different but works for menu |
| Physician 5 | Physician 5 Days of teaching only | Physician 5 Days of research only menu | Physician 5 Always different but works for menu |

| | Days of teaching only menu | Days of research only | Always different but works for |
|-------------|--|--|---|
| | | | _ |
| Physician 6 | Physician 6 Days of teaching only menu | Physician 6 Days of research only menu | Physician 6 Always different but works for menu |
| Physician 7 | Physician 7 Days of teaching only menu | Physician 7 Days of research only menu | Physician 7 Always different but works for menu |
| Nurse 1 | Nurse 1 Days of teaching only menu | Nurse 1 Days of research only menu | Nurse 1 Always different but works for menu |
| | | | |
| Nurse 2 | Nurse 2 Days of teaching only menu | Nurse 2 Days of research only menu | Nurse 2 Always different but works for menu |
| Nurse 3 | Nurse 3 Days of teaching only menu | Nurse 3 Days of research only menu | Nurse 3 Always different but works for menu |
| Nurse 4 | Nurse 4 Days of teaching only menu | Nurse 4 Days of research only menu | Nurse 4 Always different but works for menu |
| Nurse 5 | Nurse 5 Days of teaching only menu | Nurse 5 Days of research only menu | Nurse 5 Always different but works for menu |
| Nurse 6 | Nurse 6 Days of teaching only | Nurse 6 Days of research only menu | Nurse 6 Always different but works for menu |

| | International standard | Good national standard | Insufficient | Do not know | Not applicable |
|---|------------------------|------------------------|--------------|-------------|----------------|
| Research support services / unit | 0 | 0 | 0 | 0 | 0 |
| Research laboratories | 0 | 0 | 0 | 0 | 0 |
| Teaching labs (if different from the research Labs) | 0 | 0 | o | 0 | 0 |
| Large research equipment that requires technicians | 0 | 0 | 0 | 0 | 0 |
| Scientific databases (e.g. ISI Web of Knowledge) | 0 | 0 | 0 | 0 | 0 |

| | International standard | Good national standard | Insufficient | Do not know | Not applicable |
|---|------------------------|------------------------|--------------|-------------|----------------|
| Knowledge Transfer Office | 0 | 0 | 0 | 0 | 0 |
| Scientific Park | 0 | | 0 | 0 | C |
| Research ethics committee | 0 | 0 | 0 | 0 | 0 |
| Libraries | 0 | | 0 | | 0 |
| Research publication repositories | 0 | 0 | 0 | 0 | 0 |
| Computer equipment | 0 | | 0 | | 0 |
| IT Services | 0 | 0 | 0 | 0 | 0 |
| Accommodation for visiting fellows/researchers | 0 | 0 | 0 | 0 | 0 |
| Buildings/premi ses in general | 0 | 0 | 0 | 0 | 0 |
| Communication infrastructure (phone, internet etc.) | c | 0 | О | 0 | С |
| Data Sharing Platform | 0 | 0 | 0 | 0 | 0 |

*27. Do your faculty/ department/ institute members engage in the following activities?

| | Common practice | Possible but rarely used/done | Not practiced | Not possible, due to regulations/ laws | Do not know | Not applicable |
|---|-----------------|-------------------------------|---------------|---|-------------|----------------|
| Collaborative research with international companies | 0 | 0 | 0 | c | 0 | 0 |

| | Common practice | Possible but rarely used/done | Not practiced | Not possible, due to regulations/ laws | Do not know | Not applicable |
|--|-----------------|-------------------------------|---------------|---|-------------|----------------|
| Contract research for international companies | 0 | 0 | 0 | 0 | 0 | 0 |
| Collaborative research with domestic companies | 0 | 0 | 0 | 0 | С | 0 |
| Contract research for domestic companies | 0 | 0 | 0 | 0 | 0 | 0 |
| Working in industry (contract or part-time) | 0 | 0 | 0 | 0 | 0 | 0 |
| Spin-off creation | 0 | 0 | 0 | 0 | 0 | 0 |
| Patent activity | 0 | 0 | 0 | 0 | 0 | 0 |

*28. How would you rate the quality of the following at your faculty/ department/institute?

| | International standard, or close to it | Good national standard | Insufficient | Do not know | Not applicable |
|--------------------------------------|--|------------------------|--------------|-------------|----------------|
| Research Excellence | 0 | 0 | 0 | 0 | 0 |
| Attractive institutional environment | 0 | С | 0 | О | 0 |
| Industry contacts and collaboration | 0 | 0 | 0 | 0 | 0 |
| International networking | 0 | 0 | 0 | 0 | 0 |

| | International standard, or close to it | Good national standard | Insufficient | Do not know | Not applicable |
|-------------------------------|--|------------------------|--------------|-------------|----------------|
| Quality Assurance | 0 | 0 | О | 0 | О |
| *29. Do you award O Yes O No | doctorates | | | | |
| *30. How frequently | y are the follo | wing? | | | |

| | Common practice | Possibly but rarely used | Not practiced | Not possible due to regulations/laws |
|--|-----------------|--------------------------|---------------|--------------------------------------|
| Doctoral thesis can be written in English | 0 | 0 | 0 | 0 |
| PhD courses are taught in English | 0 | 0 | 0 | 0 |
| Transferable skills training is provided | 0 | 0 | 0 | 0 |
| Interdisciplinary research is encouraged | 0 | 0 | 0 | О |
| International mobility for doctoral candidates (to attend conferences, access libraries and labs abroad) | С | С | c | С |
| International exchange of doctoral candidates | 0 | 0 | 0 | 0 |
| International joint doctoral Programmes | C | 0 | 0 | C |
| Programmes in collaboration with industry | 0 | 0 | 0 | 0 |

| | Common practice | Possibly but rarely used | Not practiced | Not possible due to regulations/laws | | | |
|--|-----------------|--------------------------|---------------|--------------------------------------|--|--|--|
| Support units for doctoral studies | 0 | 0 | 0 | 0 | | | |
| Research study rooms for candidates | 0 | 0 | 0 | 0 | | | |
| Provision of accommodation for doctoral candidates | 0 | 0 | 0 | 0 | | | |
| Other (please specify) | | | | | | | |
| *31. Have you participated in any clinical trials before? Organical trials before? Organical trials before? | | | | | | | |
| *32. What was your role in the latest clinical trial that you have participated in? Not participated in clinical trial Principal investigator Co-investigator Sub-investigator | | | | | | | |
| *33. What do you think are the benefits of conducting clinical trials? | | | | | | | |

| | Strongly agree | Agree | Agree to some extent | Disagree |
|--|----------------|-------|----------------------|----------|
| Doctors may help patients with new treatments | 0 | 0 | 0 | 0 |
| Doctors can contribute to medical progress | 0 | 0 | 0 | 0 |
| Doctors can obtain a wider and deeper understanding of the disease | 0 | 0 | 0 | С |
| Doctors can obtain research grants or other rewards | 0 | C | 0 | 0 |
| Doctors can enhance their reputation in society or in the hospital | 0 | 0 | 0 | 0 |
| Doctors can obtain credits to be a board certified doctor | 0 | С | С | С |
| Doctors can write papers about the clinical trials | C | 0 | C | 0 |
| The clinical trial will have benefits for patients | 0 | 0 | 0 | 0 |
| Conducting clinical trial is just a waste of time and energy | 0 | 0 | 0 | 0 |

*34. What are the major problems in conducting clinical trials?

| | A serieus problem | A problem | A small problem | No problem |
|---|-------------------|-----------|-----------------|------------|
| Lack of time for clinicians | 0 | 0 | 0 | 0 |
| Shortage of clinical research coordinators/ | 0 | 0 | 0 | 0 |
| research nurses | 0 | 0 | 0 | 0 |
| Difficulties with data management and/or | 0 | 0 | 0 | 0 |

| | A serieus problem | n A problem | A small problem | No problem | | | |
|---|-------------------|-------------|-----------------|------------|--|--|--|
| statistical analysis | 0 | 0 | 0 | 0 | | | |
| Insufficiency of infrastructure | 0 | 0 | 0 | 0 | | | |
| Lack of systemic support from the hospital | 0 | 0 | 0 | 0 | | | |
| Difficulties in communication (e.g | 0 | 0 | 0 | 0 | | | |
| ethical committee, | 0 | 0 | | 0 | | | |
| Enrollment of trial participants | 0 | 0 | \circ | 0 | | | |
| Obtaining of informed consent | 0 | 0 | 0 | 0 | | | |
| Lack of communication with trial sponsor | 0 | 0 | 0 | 0 | | | |
| Funding | 0 | C | 0 | 0 | | | |
| Signing of the contract between the company and the hospital administration | 0 | o | 0 | 0 | | | |
| Other difficulties to be listed *35. Have you ever participated in a multinational clinical trial? Other difficulties to be listed *35. Have you ever participated in a multinational clinical trial? Other difficulties to be listed | | | | | | | |

clinical trial?[CB1]

[CB1]The following two questions will be linked to the answer to question 7 (yes or no) and we could separately analyse the answers form the yes and no group

| | A serieus problem | A problem | A small problem | No problem |
|-------------------------------------|-------------------|-----------|-----------------|------------|
| Investigator qualifications | 0 | О | 0 | 0 |
| Quality of clinical trials | 0 | 0 | 0 | 0 |
| Infrastructure of clinical trials | 0 | 0 | 0 | 0 |
| Difference of the medical system | 0 | 0 | 0 | 0 |
| Regulation process and legal system | О | С | О | 0 |
| Language barriers | 0 | 0 | 0 | 0 |
| Cost-effectiveness | 0 | 0 | 0 | C |
| | | | | |

Other difficulties to be listed

*37. What do you think is important for overcoming the obstacles for the development of clinical trials?

| | Very important | Important | Partially important | Not important |
|---|----------------|-----------|---------------------|---------------|
| Reform of the system and regulatory rules | 0 | C | 0 | О |
| Financial support to improve the quality of clinical trials | 0 | 0 | 0 | С |
| Training specialized people in clinical trials | 0 | C | 0 | 0 |

| | Very importa | ant Importan | t Partially importan | nt Not important | | | |
|--|---------------------------------------|---------------------------|-----------------------|------------------|--|--|--|
| Establishment of clinical trial centers | 0 | 0 | 0 | 0 | | | |
| Good training programs for clinical trials | 0 | c | О | C | | | |
| Public relations to citizens | 0 | 0 | С | 0 | | | |
| *38. Are you interested in conducting clinical trials? Yes No *39. Who decides on how this budget is allocated to the faculties/ departments? Institutional leadership (Dean, president, director) | | | | | | | |
| A central body of the institution (academic senate or similar) Ministry/ national agency *40. How would you generally describe facilities and infrastructure at your institution | | | | | | | |
| gene | International standard or close to it | Good national standard | Insufficient Do not k | | | | |
| Research support services / unit | 0 | C C | C | С | | | |

| | International standard or close to it | Good national standard | Insufficient | Do not know | Not applicable |
|---|---------------------------------------|------------------------|--------------|-------------|----------------|
| Research laboratories | 0 | 0 | 0 | 0 | 0 |
| Teaching labs (if different from research Labs) | 0 | 0 | 0 | 0 | 0 |
| Large research equipment that requires technicians | 0 | С | 0 | 0 | 0 |
| Scientific databases (e.g. ISI Web of Knowledge) | 0 | 0 | 0 | 0 | c |
| Knowledge Transfer Office | 0 | 0 | 0 | \circ | 0 |
| Science Park | 0 | 0 | 0 | 0 | 0 |
| Research ethics committee | 0 | 0 | 0 | \circ | 0 |
| Libraries | 0 | C | 0 | <u> </u> | 0 |
| Research publication repositories | 0 | 0 | 0 | 0 | 0 |
| Computer equipment | 0 | 0 | 0 | 0 | 0 |
| IT Services | 0 | 0 | 0 | 0 | 0 |
| Accommodation for visiting scholars | 0 | 0 | 0 | 0 | 0 |
| Buildings/premi ses in general | 0 | 0 | 0 | 0 | 0 |
| Power supply/ electricity | C | 0 | 0 | 0 | 0 |
| Communication infrastructure (phone, internet etc.) | О | 0 | 0 | 0 | 0 |
| | | | | | |

*41. Does your institution have, or engages in, any of the following

| | Yes very strong | Yes occasionally / some | No | Do not know | Not applicable |
|--|-----------------|-------------------------|----|-------------|----------------|
| Spin-offs | 0 | 0 | 0 | 0 | 0 |
| Research collaboration with industry | 0 | 0 | 0 | 0 | 0 |
| International research collaboration | 0 | 0 | 0 | 0 | 0 |
| Research collaboration with public/ state offices and agencies | 0 | 0 | 0 | 0 | 0 |
| Patents | 0 | 0 | 0 | 0 | 0 |

*42. Has your institution been involved in applications for research projects (regardless whether successful or not)?

| | Frequently as an applicant | Frequently as a partner | Occasionally as an applicant | Occasionally as partner | No involvement of application |
|---|----------------------------|-------------------------|------------------------------|-------------------------|-------------------------------|
| National, involving colleagues from another institution | С | 0 | 0 | C | 0 |
| Horizon 2020 / FP7 | 0 | 0 | 0 | 0 | 0 |
| Other European project | 0 | 0 | 0 | 0 | 0 |
| Other International project | 0 | 0 | 0 | 0 | 0 |
| | | ANY SHAPPINE | | | |

*43. When have you been last research active as a health professional? (Research activity is defined here as being involved in the design or implementation of a research project either as a researcher or a supervisor)

Within the last 2 years

O More than 2 years ago

Never been research active as a health professional

*44. Have you published any of the following over the last 3 years 2014-2017?

| | Yes mostly with single authorship | Yes mostly co- authored | Only supervising | No |
|---|-----------------------------------|----------------------------|------------------|----|
| ISI journal articles | 0 | C | 0 | С |
| Other journal articles (non-ISI) | 0 | 0 | 0 | 0 |
| Papers presented at international conferences | 0 | 0 | 0 | 0 |
| Papers presented at national conferences | С | 0 | 0 | 0 |
| PhD. Thesis | С | 0 | 0 | 0 |
| Books | 0 | 0 | 0 | 0 |
| Book chapters | 0 | 0 | 0 | 0 |
| Reports | O | C | 0 | 0 |

*45. If co-authored, who do you write with more often?

| | Colleagues from my institution | Colleagues from other institutions | - | Intercontinental Colleagues | No co-authoring |
|---|--------------------------------|------------------------------------|---|-----------------------------|-----------------|
| ISI journal articles | 0 | 0 | 0 | 0 | 0 |
| Other journal articles (non-ISI) | 0 | 0 | 0 | 0 | 0 |
| Papers presented at international conferences | О | 0 | 0 | 0 | С |
| Papers presented at national conferences | О | 0 | 0 | 0 | 0 |
| PhD. Thesis | 0 | 0 | 0 | 0 | 0 |
| Books | 0 | 0 | 0 | 0 | 0 |
| Book chapters | 0 | 0 | 0 | 0 | 0 |
| Reports | 0 | 0 | 0 | 0 | 0 |

*46. What factors had motivated you to be research active?

| | Did not motivate me | Yes some motivation | very strong motivation | N/A |
|--------------------------------|---------------------|---------------------|------------------------|-----|
| Personal motivation', ' | 0 | 0 | 0 | C |
| Completing an academic degree' | o | 0 | 0 | 0 |
| Professional development | 0 | C | 0 | 0 |
| Career advancement | 0 | 0 | 0 | C |

| | Did not motivate me | Yes some motivation | very strong motivation | N/A |
|--|--|---------------------|------------------------|---|
| Part of the professional role | 0 | 0 | С | С |
| For publication | 0 | 0 | 0 | 0 |
| To gain more expertise | С | c | 0 | С |
| *47. If not active in If active in research | | | | ch engagement: ts or discourages you |
| from engaging in re | search. | | • | |
| Not enough time available Clinical workload pressure Lack of support/encourag Lack of funding Lack of available resource Difficulties gaining ethical Research not valued in or Weaknesses in research Personal circumstances (Lack of research opportur Lack of supervision/mente Difficulties collecting or ga There is no reason why p | es ement es for research approval rganisation skills e.g., family commitment nities orship aining access to data | | | |

*48. What are the main problems that researchers face at your institution

| | Strongly agree | Agree to some extent | Disagree to some extend | 0 3 | Do not know | N/A |
|---|----------------|----------------------|-------------------------|-----|-------------|---------|
| There is no or insufficient research funding | 0 | 0 | 0 | 0 | 0 | 0 |
| There are no or insufficient research facilities | 0 | 0 | 0 | 0 | 0 | 0 |
| Some research facilities can only be accessed by some researchers | 0 | 0 | 0 | 0 | 0 | 0 |
| There is no or insufficient administrative capacity to prepare research proposals, manage research projects etc.) | 0 | 0 | 0 | 0 | 0 | 0 |
| Most researchers have no international research contacts | 0 | ं | 0 | 0 | 0 | 0 |
| Most researchers are not internationally mobile | 0 | 0 | 0 | 0 | 0 | \circ |
| It is not possible to hire international researchers | 0 | 0 | 0 | 0 | 0 | 0 |
| Most researchers have to teach, and cannot put enough focus on their research | 0 | 0 | 0 | 0 | 0 | 0 |
| Most researchers have other jobs on the side and cannot put enough focus on their research | O | O | c | С | 0 | 0 |

^{*49.} What do you think are the reasons why people in your Institution are not, or limited engaged in research?

| Not enough time available to engage in research | | | | | | | | |
|---|---|-------------------|----------------|--------------------|----------|----------------------|----------------|--|
| Clinica | Clinical workload pressures | | | | | | | |
| Lack o | Lack of support/encouragement | | | | | | | |
| Lack o | Lack of funding | | | | | | | |
| Lack o | Lack of available resources for research | | | | | | | |
| Difficul | Difficulties gaining ethical approval | | | | | | | |
| Research not valued in organisation | | | | | | | | |
| Weaknesses in research skills | | | | | | | | |
| Personal circumstances (e.g., family commitments limiting time for research) | | | | | | | | |
| Lack o | Lack of research opportunities | | | | | | | |
| Lack o | Lack of supervision/mentorship | | | | | | | |
| Difficul | Difficulties collecting or gaining access to data | | | | | | | |
| There is no reason why people should be limited or not engaged in research | | | | | | | | |
| | | | | | | | | |
| | | | | | | | | |
| *50. What do you see as the main problems for research in your country ? | | | | | | | | |
| 30. What do you see as the main problems for research in your country ? | | | | | | | | |
| | | | agree to | neither | | . 1 | . | |
| | | strongly agree | some extent | agree nor disagree | disagree | strongly disagree | Do not know | |
| Research is | not given sufficiently high priority by the state | 0 | 0 | C | 0 | 0 | 0 | |
| | | \mathbf{v} | V | U | U | v | V | |
| There is no national strategy, which would provide a clear framework | | 0 | 0 | 0 | 0 | 0 | 0 | |

0

There is no or insufficient research funding

| | strongly agree | agree to some extent | neither agree nor disagree | disagree | strongly disagree | Do not know |
|--|-------------------|----------------------|----------------------------------|----------|----------------------|-------------|
| There are no or insufficient research facilities | 0 | | 0 | 0 | 0 | 0 |
| Research is too much concentrated in a few institutions | 0 | 0 | 0 | 0 | 0 | 0 |
| Higher education institutions are focusing too much on teaching, and neglect research | 0 | 0 | 0 | 0 | 0 | 0 |
| Research is not sufficiently applied, and ignores the needs of the industry and society | 0 | 0 | 0 | 0 | 0 | 0 |
| There is too much focus on applied research | 0 | 0 | \circ | 0 | 0 | 0 |
| Interdisciplinary research is not sufficiently encouraged | 0 | 0 | 0 | 0 | 0 | 0 |
| The best researchers are leaving the country, which is a problem | 0 | 0 | 0 | 0 | | 0 |
| The best researchers are leaving the country, but they still continue to Contribute to national research | 0 | 0 | c | 0 | 0 | 0 |
| Young researchers are leaving the country | 0 | 0 | \circ | \circ | \circ | 0 |
| Young talented people look for careers outside of the research and higher Education sectors | С | c | C | С | c | C |

^{*51.} Of the changes listed in the table below, indicate how effective you believe they would be for increasing the level of research activity in your Institution

| Increased protected working time for research Higher levels of ongoing research training Recruiters in the health service giving more value to research experience when selecting candidates for clinical positions A more simplified and efficient ethics application process Greater levels of mentorship and support when conducting research More opportunities to advance one's research qualifications (e.g. post-doctoral fellowships) | | Would have no effect | Would be somewhat effective | Would be greatly effective |
|--|---|----------------------|-----------------------------|----------------------------|
| Recruiters in the health service giving more value to research experience when selecting candidates for clinical positions A more simplified and efficient ethics application process Greater levels of mentorship and support when conducting research More opportunities to advance one's research qualifications (e.g. post-doctoral fellowships) | Increased protected working time for research | 0 | 0 | C |
| experience when selecting candidates for clinical positions A more simplified and efficient ethics application process Greater levels of mentorship and support when conducting research More opportunities to advance one's research qualifications (e.g. post-doctoral fellowships) | Higher levels of ongoing research training | 0 | 0 | 0 |
| Greater levels of mentorship and support when conducting research More opportunities to advance one's research qualifications (e.g. post-doctoral fellowships) | | 0 | 0 | 0 |
| More opportunities to advance one's research qualifications (e.g. post-doctoral fellowships) | A more simplified and efficient ethics application process | 0 | 0 | 0 |
| post-doctoral fellowships) | Greater levels of mentorship and support when conducting research | 0 | 0 | 0 |
| More funded research posts | | 0 | 0 | 0 |
| Triole failed research posts | More funded research posts | 0 | 0 | 0 |
| More opportunities for collaboration with other clinical or academic researchers | * * | 0 | 0 | О |

*52. About research Infrastructure, what is applicable for you?

| | Don't know it | Know it | Collaborate with it |
|--|---------------|---------|---------------------|
| BBMRI Biobanking and Biomolecular Resources Research Infrastructure | 0 | С | С |
| C-ERIC Central-European Research Infrastructures Consortium | 0 | 0 | 0 |
| CLARIN Common Language Resources and Technology Infrastructure | 0 | 0 | 0 |
| EATRIS European Advanced Translational Research Infrastructure in Medicine | С | О | 0 |
| ECRIN European Clinical Research Infrastructure Network | 0 | 0 | C |

| | Don't know it | Know it | Collaborate with it |
|---|---------------|---------|---------------------|
| ELIXIR The European Life-Science Infrastructure for Biological Information | О | 0 | 0 |
| ERA European Research Area | 0 | C | 0 |
| ERIC European Research Infrastructure Consortium | C | 0 | С |
| ESFRI European Strategy Forum on Research Infrastructure | 0 | 0 | C |
| EU-OPENSCREEN European Infrastructure of Open Screening Platforms for Chemical Biology | 0 | 0 | 0 |
| Euro-BioImaging European Research Infrastructure for Imaging Technologies in Biological and Biomedical Sciences | o | 0 | 0 |
| INFRAFRONTIER European Research Infrastructure for the generation, phenotyping, archiving and distribution of mouse | С | 0 | 0 |
| ISBE Infrastructure for Systems Biology Europe | 0 | 0 | C |
| MIRRI Microbial Resource Research Infrastructure disease models | C | 0 | С |
| NRRI Research Infrastructure Roadmap | 0 | 0 | C |
| PRACE Partnership for Advanced Computing in Europe | 0 | 0 | 0 |
| RI Research infrastructure(s) | 0 | C | 0 |

*53. About biobanks, what is applicable for you?

| | Don't know it | Know it | Collaborate with it |
|--|---------------|---------|---------------------|
| Basque Biobank | C | 0 | C |
| Biobanco.pt (Portuguese National Consortium of Biobanks) | 0 | 0 | 0 |

| | Don't know it | Know it | Collaborate with it |
|---|---------------|---------|---------------------|
| Biobank Inherited Metabolic Disorders (blood/urine/liquor/fibroblasts) | 0 | 0 | 0 |
| Biobank of the Estonian genome center | 0 | 0 | 0 |
| BioNER: Biobank of the Institute of Rare Diseases Research/Institute of Health Carlos III | 0 | c | С |
| Brain-net – brain tissue bank Aachen | 0 | 0 | 0 |
| Brain-Net – brain tissue bank Munster (pediatric neuropathology) | 0 | C | 0 |
| Brain-Net Germany: National network of brain banks in Germany | 0 | 0 | C |
| Cell line and DNA Biobank from patients affected by genetic diseases | 0 | 0 | 0 |
| CIBERER Biobank (Biobank of rare diseases) | 0 | 0 | C |
| DNA biobank of the Department of Molecular Genetics and Diagnostics, National Institute of Environmental Health (NIEH) (EuroBioBank partner) | 0 | 0 | С |
| Genetic alliance Registry and Biobank | 0 | 0 | 0 |
| EuroBioBank European Network of DNA, cell and tissue banks for Rare Diseases | 0 | 0 | 0 |
| Human DNA and cell biobank of Genethon – genetic diseases, mainly neuromuscular diseases – part of the EuroBioBank network | 0 | 0 | С |
| Human tissue biobank of Neuromuscular diseases (EuroBioBank partner) | 0 | 0 | 0 |
| Neuromuscular Tissue Bank (NMTB) (EuroBioBank partner) | 0 | 0 | C |
| Telethon Network of Genetic Biobanks | 0 | 0 | 0 |
| UK Biobank | 0 | О | 0 |

^{*54.} About registry and database, what is applicable for you?

| | Don't know it | Know it | Collaborate with it |
|---|---------------|---------|---------------------|
| (NCARDRS) National Congenital Anomaly and Rare Disease Registration Service | C | С | О |
| Austrian Country Node of the Human Variome Project (HVP) | 0 | 0 | С |
| Austrian registry for inborn errors of metabolism | 0 | 0 | С |
| Belgian Neuromuscular Disease Registry | \circ | 0 | C |
| Belgian patient database for Wilson disease – contributes to the EuroWilson registry (terminated) | С | 0 | 0 |
| Biobank Inherited Metabolic Disorders (blood/urine/liquor/fibroblasts) | \circ | 0 | 0 |
| Canadian Neuromuscular Disease Registry – part of the TREAT-NMD network | <u> </u> | 0 | C |
| DDRMD – Dutch Diagnosis Registration Metabolic Diseases | \circ | 0 | 0 |
| Dutch Neuromuscular Database CRAMP: Computer Registry of All Myopathies and Polyneuropathies | 0 | 0 | 0 |
| Dutch patient registry for Fabry disease | \circ | 0 | C |
| Dutch patient registry for Gaucher disease | <u> </u> | 0 | 0 |
| Dutch patient registry for Niemann-Pick Disease Type B | 0 | 0 | О |
| Dutch patient registry for Niemann-Pick Disease Type C | 0 | 0 | 0 |
| ECARUCA: cytogenetic and clinical database on rare chromosomal disorders | 0 | 0 | 0 |
| E-HOD European Network and Registry for Homocystinurias and Methylation Defects | 0 | 0 | 0 |
| EIMD: European registry and network for intoxication type metabolic diseases | 0 | 0 | 0 |
| English alpha-1 antitrypsin deficiency registry – contributes to the Alpha One International Registry (AIR) | 0 | 0 | С |
| English cystinosis registry | 0 | 0 | 0 |

| | Don't know it | Know it | Collaborate with it |
|--|---------------|---------|---------------------|
| English hyperoxaluria registry | 0 | 0 | 0 |
| English phenylketonuria registry | 0 | 0 | 0 |
| EPNET: European porphyria Network | 0 | 0 | C |
| E-RARE Consortium | 0 | 0 | О |
| ESNEK – Rare paediatric neurological disease registry Germany | 0 | 0 | c |
| EUNEFRON: registry of the European network for the study of orphan nephropathies | 0 | 0 | 0 |
| EUROCAT European Surveillance on congenital anomalies | 0 | 0 | 0 |
| EUROGLYCANET – International patient registry and cohort for congenital disorders of glycosylation | 0 | 0 | 0 |
| European cystinosis cohort | 0 | 0 | c |
| European Porphyria Registry (EPR) | 0 | \circ | 0 |
| Fabry Disease Registry –DE | 0 | 0 | <u> </u> |
| FOS : Fabry Outcome Survey | 0 | 0 | С |
| French certified registry of glycogen storage disease type 2 | 0 | 0 | C |
| French certified registry of patients affected by Gaucher disease | 0 | \circ | C |
| French cystinosis registry | 0 | 0 | c |
| French registry of neuromuscular diseases from reference centres | 0 | \circ | 0 |
| Galactosemia Patient Registry – part of the Galactosemia Network | 0 | 0 | C |
| Genetic alliance Registry and Biobank | 0 | 0 | 0 |
| German mucopolysaccharidosis patient registry | 0 | 0 | c |
| HOS: Hunter Outcome Survey | 0 | О | С |

| | Don't know it | Know it | Collaborate with it |
|--|---------------|---------|---------------------|
| iNTD Network – International Working Group on Neurotransmitter Related Disorders | 0 | С | С |
| International Collaborative Gaucher Group (ICGG) Gaucher Registry – DE | 0 | 0 | 0 |
| International Morquio A registry | 0 | 0 | C |
| International Niemann-Pick Disease Registry | 0 | 0 | C |
| LOVD: GAA gene (Glucosidase, alpha, acid) variant database | 0 | 0 | C |
| Mendelian cytogenetics network online database | 0 | 0 | 0 |
| MITOCON: National registry mitochondrial disease | 0 | 0 | 0 |
| Mucopolysaccharidosis type 1 (MPS I) Registry – | 0 | 0 | 0 |
| NCL-Registry: International neuronal ceroid lipofuscinoses patient registry | 0 | 0 | 0 |
| NCL-Registry: International neuronal ceroid lipofuscinoses patient registry | 0 | 0 | 0 |
| Neuro-Heart-Registry: Registry for patients with heart involment in neurovascular diseases | 0 | 0 | 0 |
| NMDcy: Cypriot Neuromuscular Diseases – contributes to TREAT-NMD | 0 | 0 | 0 |
| NMD-ES: Spanish Registry of Neuromuscular Diseases | 0 | 0 | 0 |
| Norvegian Porphyria Registry | 0 | 0 | 0 |
| Pompe Disease Registry – DE | 0 | 0 | 0 |
| Portuguese Fabry patient registry – contributing to the international Fabry registry | 0 | C | С |
| RAMEDIS: Rare Metabolic Diseases Database | 0 | 0 | C |
| RAMEDIS: Rare Metabolic Diseases mutation Database | 0 | 0 | 0 |
| Registry for Patients with Niemann-Pick Type C Disease | 0 | 0 | 0 |
| Registry for Wolfram syndrome, Alström syndrome, Bardet-Biedl syndrome | 0 | 0 | 0 |

| | Don't know i | t Know it | Collaborate with it |
|--|--------------|-----------|---------------------|
| and other rare diabetes syndromes - contributing to EURO-WABB | | | |
| Registry_Biobank Name | \circ | 0 | 0 |
| The International Collaborative Gaucher Group (ICGG) Gaucher registry | 0 | 0 | 0 |
| The international Fabry registry | <u> </u> | 0 | 0 |
| The international Mps I registry | 0 | 0 | 0 |
| The international Pompe registry | <u> </u> | 0 | 0 |
| TREAT-NMD: Accelerating Treatments for Neuromuscular Diseases (registries) | C | 0 | 0 |
| UK10K RARE NEUROMUSCULAR – Genetic neuromuscular diseases variant database | c | 0 | o |
| X-ALD: X-linked adrenoleukodystrophy database | \circ | 0 | 0 |

*55. About scientific organisation, what is applicable for you?

| | Don't know it | Know it | Collaborate with it |
|---|---------------|---------|---------------------|
| AECOM – Asociación Española para el Estudio de los Errores Congénitos del Metabolismo | 0 | 0 | 0 |
| APS – Arbeitsgemeinschaft für Pädiatrische Stoffwechselstörungen (Working group for paediatric metabolic disturbances, Germany) | 0 | c | 0 |
| B4B – Brains for Brain | 0 | | 0 |
| BIMDG – British Inherited Metabolic Disease Group | 0 | 0 | 0 |
| CLIMB – Children Living with Inherited Metabolic Diseases, UK | 0 | 0 | 0 |

| | Don't know it | Know it | Collaborate with it |
|--|---------------|---------|---------------------|
| EBC – European Brain Council | 0 | 0 | 0 |
| ERNDIM Quality assurance in laboratory testing for IEM | 0 | 0 | 0 |
| ESGCT – European Society of Gene and Cell Therapy | 0 | 0 | 0 |
| EURORDIS – European Organisation for Rare Disorders | 0 | 0 | C |
| Fondazione Telethon | 0 | \circ | C |
| IBBS – International Brain Barriers Society | 0 | 0 | C |
| IRDIRC | 0 | 0 | 0 |
| ISNS – International Society for Neonatal Screening | 0 | 0 | 0 |
| SEEIM – Sociedad Española de Errores Innatos del Metabolismo | 0 | 0 | 0 |
| SFEIM – Societé Française pour l'Étude des Erreurs Innées du Métabolisme | c | 0 | С |
| SIMD – Society for Inherited Metabolic Disorders | 0 | \circ | 0 |
| SISMME – Societa' Italiana per lo Studio delle Malattie Metaboliche Ereditairie | c | 0 | О |
| SPDM – Sociedade Portuguesa de Doenças Metabólicas | 0 | \circ | C |
| SSIEM – The Society for Study of Inborn Errors of Metabolism | 0 | C | С |

^{*56.} How many ongoing research do you participate in at your department at the moment.

| 57. How many of these are clinical trials? | |
|---|----------|
| | |
| 58. How many of the clinical trials are in phase I? | • |
| 59. How many of the clinical trials are in phase II? | • |
| 60. How many of the clinical trials are in phase III? | V |
| 61. How many of the clinical trials are in phase IV? | • |
| | |



4. Type of institution

Survey MetabERN for Universities

| *1. Can you please type in your contact details | |
|--|---|
| Name | |
| Email Address | |
| Phone Number | |
| *2. Please select the name of your hospital /institute from the drop down menu | |
| | ▼ |
| 3. Please select what is applicable to your situation | |
| | |
| ^C male | |

| | Public university Private university Public college Private college Research institute |
|-----------|--|
| * 0 0 0 | 5. What is the legal status of your institution Public Private mixed |
| * 0000000 | 6. What is your position Professor Associate Professor Lecturer Senior Research Fellow Postdoc/Research Fellow Research Assistant Teaching Assistant Dean, head of department or other senior administration position |
| 0 | Dean, head of department or other senior administration position Other (please specify) |

| *7 | . Please select | the areas a | nd fields in | which you | ır institution | is undertaking | g research | activities |
|-----|-------------------|---------------|--------------|-----------|----------------|----------------|------------|------------|
| ple | ase select all th | ne ones of in | nterest: | | | | | |

- Animal research
- Basic research
- Preclinical research
- Clinical research

Questions to inquire the level of activity in the research thematic area.

*8. Please indicate which are, the main research activities performed inside your Institution

| | No activity | Low activity | Rather low activity | Rather high activity | High activity |
|--|-------------|--------------|---------------------|----------------------|---------------|
| Health Systems Research | 0 | 0 | 0 | 0 | 0 |
| Human and Social Science Research | 0 | 0 | 0 | 0 | C |
| Epidemiology/Natural History of diseases | 0 | 0 | 0 | 0 | C |
| Research in Genetics and Pathophysiology | 0 | 0 | 0 | 0 | C |
| Pre-Clinical Therapeutic Research | 0 | 0 | 0 | 0 | C |
| Therapeutic Research | 0 | 0 | 0 | 0 | C |
| Research Platforms/Infrastructures | 0 | 0 | 0 | 0 | C |

| Development and availability of tools needed to accelerate research on rare diseases | • | Low activity | Rather low activity | Rather high activity | High activi |
|--|-------------|--------------|---------------------|----------------------|-------------|
| *9. Academic Health Systems Research | | | | | |
| | No activity | Low activity | Rather low activity | Rather high activity | High activ |
| Research policy (e.g., guidelines development; national plans; health care resources) | 0 | С | 0 | С | С |
| Cost analysis (e.g., guidelines implementation) | 0 | 0 | О | 0 | 0 |
| New technologies (e.g., New diagnostic tests, population screening programmes, Innovative therapeutics) | С | С | О | С | 0 |
| Health-Technology assessment of Orphan Drugs | 0 | 0 | C | C | 0 |
| e-Health activities | С | С | С | О | С |
| *10. Industrial Health Systems Research | h | | | | |
| | No activity | Low activity | Rather low activity | Rather high activity | High acti |
| | | | | | |

| | No activity | Low activity | Rather low activity | Rather high activity | High activity |
|--|-------------|--------------|---------------------|----------------------|---------------|
| health care resources) | | | | | |
| Cost analysis (e.g., guidelines implementation) | 0 | 0 | 0 | 0 | 0 |
| New technologies (e.g., New diagnostic tests, population screening programmes, Innovative therapeutics) | c | 0 | 0 | C | С |
| Health-Technology assessment of Orphan Drugs | C | 0 | C | 0 | 0 |
| e-Health activities | C | С | 0 | С | C |

*11. Human and social science research

| | No activity | Low activity | Rather low activity | Rather high activity | High activity |
|---|-------------|--------------|---------------------|----------------------|---------------|
| Society and RD (e.g., social perception, accessibility to care,) | О | О | c | 0 | С |
| RD, Research and Innovation (e.g., public/private cooperation, role of patients org,) | С | С | 0 | С | 0 |
| Care practices (self- management, health education policy,) | С | С | c | С | c |
| Patients awareness (e.g., understanding genetic data, importance of animal models,) | С | С | 0 | С | C |
| Support for caregivers | C | C | 0 | C | 0 |
| Public Heath and RD (e.g., health care policies, prospective public policies,) | 0 | 0 | c | 0 | 0 |

| | No activity | Low activity | Rather low activity | Rather high activity | High activity |
|---|-------------|--------------|---------------------|----------------------|---------------|
| Methods for social support to improve quality of life | 0 | 0 | 0 | 0 | 0 |
| Services accessibility | 0 | 0 | 0 | 0 | 0 |
| Orphan Drugs availability | 0 | 0 | 0 | 0 | 0 |
| Methods to assess QoL | 0 | 0 | 0 | 0 | 0 |

*12. Epidemiology & Natural History of diseases

| | No activity | Low activity | Rather low activity | Rather high activity | High activity |
|--|-------------|--------------|---------------------|----------------------|---------------|
| Registries | C | C | 0 | 0 | 0 |
| Databases/data management systems for epidemiology studies | 0 | О | O | О | О |
| Cohorts | 0 | 0 | 0 | 0 | 0 |
| Biomarkers to assess diagnosis, prognosis and severity of diseases | 0 | 0 | 0 | O | 0 |
| Diagnosis/prognosis methods (e.g., other than biomarkers) | o | С | c | С | C |
| Identification of new entities | С | 0 | 0 | C | 0 |
| Networks for ultra rare diseases | С | 0 | 0 | C | 0 |

*13. Research in genetics and physiopathology

| | No activity | Low activity | Rather low activity | Rather high activity | High activity |
|--|-------------|--------------|---------------------|----------------------|---------------|
| Gene identification and Molecular Mechanisms | С | 0 | 0 | 0 | 0 |
| New database method developments to be applied to omics research | С | С | С | С | С |
| Biobanking research | C | 0 | 0 | 0 | C |
| High throughput genomics/sequencing | С | C | C | C | C |
| New bioinformatic methods and developments | 0 | C | C | C | C |
| New technologies (e.g., atomic microscopy) | 0 | 0 | 0 | 0 | 0 |
| Omics technology (e.g., proteo, metabolo) | 0 | C | C | C | C |
| Development of new animal models | 0 | 0 | 0 | 0 | 0 |
| New Imaging/Microscopy methods | 0 | 0 | 0 | 0 | 0 |
| Development of new therapeutic tools | С | 0 | C | C | 0 |

*14. Pre-clinical therapeutic research

| | No activity | Low activity | | Rather high activity | High activity |
|------------------|-------------|--------------|---|----------------------|---------------|
| Proof-of-concept | 0 | 0 | 0 | 0 | 0 |
| Drug libraries | 0 | 0 | 0 | 0 | 0 |

| | No activity | Low activity | Rather low activity | Rather high activity | High activity |
|---|-------------|--------------|---------------------|----------------------|---------------|
| Search for active chemical compounds | 0 | 0 | 0 | 0 | 0 |
| High throughput drug screening | 0 | C | 0 | 0 | 0 |
| Therapeutic target identification/validation (e.g.,. stem cells, systems biology) | С | 0 | О | С | О |
| Cell therapy research (e.g., viral vectors) | C | 0 | C | 0 | 0 |
| Innovative biotechnological research (e.g., monoclonal antibodies, cell and gene therapy, enzyme therapy) | С | 0 | 0 | С | 0 |
| Drug toxicology/pharmacology | 0 | C | 0 | 0 | 0 |

*15. Therapeutic research

| | No activity | Low activity | Rather low activity | Rather high activity | High activity |
|--|-------------|--------------|---------------------|----------------------|---------------|
| Drug development | 0 | 0 | 0 | 0 | c |
| Phase I/II Clinical Trials | 0 | 0 | С | 0 | C |
| Bioproduction centres | 0 | C | 0 | 0 | c |
| Epidemiological/clinical/ therapeutical investigations | С | С | O | O | О |
| Medical devices | 0 | C | 0 | 0 | c |
| Assesment of surgery procedures | 0 | 0 | 0 | 0 | c |
| Transplants | 0 | C | 0 | 0 | c |

| | No activity | Low activity | Rather low activity | Rather high activity | High activity |
|------------------------------|-------------|--------------|---------------------|----------------------|---------------|
| Pharma/academic partnerships | C | C | C | 0 | 0 |

*16. Platforms / Infrastructures development

| | No activity | Low activity | Rather low activity | Rather high activity | High activity |
|--|-------------|--------------|---------------------|----------------------|---------------|
| Bioinformatics | С | С | С | 0 | c |
| Pharmacogenomic diagnostic assays | C | 0 | 0 | 0 | O |
| Population Registries | c | C | c | C | C |
| Genetically-modified animal model repositories | 0 | О | С | С | 0 |
| GMP platforms | c | 0 | 0 | 0 | c |
| Biobanking | 0 | 0 | c | 0 | 0 |
| Genomic and proteomic platforms | c | 0 | 0 | C | c |
| Long-term cohort of patients studies | C | 0 | 0 | 0 | 0 |
| High throughput drug screening | 0 | С | 0 | С | 0 |

*17. Human resources

| | No activity | Low activity | Rather low activity | Rather high activity | High activity |
|--------------------------------------|-------------|--------------|---------------------|----------------------|---------------|
| Rotational positions for clinicians | | 0 | 0 | 0 | 0 |
| Rotational positions for researchers | | 0 | 0 | 0 | 0 |
| Specific training in RD | | C | С | C | 0 |
| Training in bioinformatics | , | 0 | 0 | O | 0 |

*18. Development and availability of tools needed to accelerate research on rare disease.

| | No activity | Low activity | Rather low activity | Rather high activity | High activity |
|--|-------------|--------------|---------------------|----------------------|---------------|
| Network development | 0 | 0 | 0 | 0 | 0 |
| Creation of transnational research consortiums | 0 | 0 | 0 | 0 | 0 |
| Transnational calls for projects funding | 0 | 0 | 0 | 0 | 0 |
| Opening of national research programmes to international collaboration | 0 | O | 0 | C | 0 |
| Transnational research infrastructures | 0 | 0 | 0 | 0 | 0 |
| Development of an inventory/directory of databases of experts | 0 | О | О | С | 0 |
| Transnational collection of funded RD research projects | С | 0 | С | С | c |

*19. What influences and governs research at your institution?

| | Strongly agree | Agree to some extent | Disagree to some extent | Strongly disagree | No information available | Not applicable |
|--|----------------|----------------------|-------------------------|-------------------|--------------------------|----------------|
| Ministry/ national research agency has a strong say | C | 0 | 0 | C | 0 | C |
| Central university level mostly decides | 0 | C | C | C | О | 0 |
| Faculties/ departments/ institutes decide very much on what they do | 0 | С | С | C | c | 0 |
| Individual professors/ Researchers do very much what they want | 0 | C | 0 | 0 | 0 | 0 |
| In my own position, I can influence research agendas at my faculty/department/ institute | 0 | С | С | c | c | 0 |

*20. Please rate the importance of funding sources for research

| | Very important | Of some importance | Not important | Do not know | Not applicable |
|-------------------------------------|----------------|--------------------|---------------|-------------|----------------|
| Institutional funding by government | 0 | 0 | 0 | С | C |
| Research grants (EU) | С | С | С | 0 | О |
| Other international research grants | С | c | c | 0 | С |
| Contracts from industry | C | С | С | 0 | С |
| Endowment income, donations | c | c | c | c | С |
| Research Board | 0 | 0 | 0 | 0 | О |

| | Very important | Of some importance | Not important | Do not know | Not applicable |
|---|----------------|--------------------|---------------|-------------|----------------|
| Charity | 0 | 0 | C | 0 | 0 |
| Private company | 0 | C | 0 | 0 | С |
| Scientific Society/PatientsOrganization | C | C | c | 0 | C |
| Local Istitutional Funding by Single Univeristies, | 0 | 0 | O | C | 0 |
| Local and/or Regional authorities, | c | C | c | 0 | С |
| Funding by Health Authorities | 0 | C | 0 | 0 | С |
| Funding by no-profit, Private Agencies (e.g. Telethon), | С | c | 0 | С | 0 |

*21. Who decides on how this budget is allocated to the faculties/ departments?

C Institutional leadership (Dean, president, director)

A central body of the institution (academic senate or similar)

Ministry/ national agency

*22. How would you generally describe facilities and infrastructure at your institution

International standard or close Good national to it standard

Insufficient Do not know

Not applicable

| | International standard or close to it | Good national standard | Insufficient | Do not know | Not applicable |
|---|---------------------------------------|------------------------|--------------|-------------|----------------|
| Research support services / unit | С | 0 | С | 0 | C |
| Research laboratories | 0 | 0 | 0 | 0 | 0 |
| Teaching labs (if different from research Labs) | 0 | 0 | 0 | 0 | С |
| Large research equipment that requires technicians | 0 | 0 | 0 | O | 0 |
| Scientific databases (e.g. ISI Web of Knowledge) | o | c | 0 | c | С |
| Knowledge Transfer Office | O | О | О | C | С |
| Science Park | 0 | 0 | C | C | C |
| Research ethics committee | 0 | C | 0 | C | 0 |
| Libraries | C | 0 | C | 0 | 0 |
| Research publication repositories | 0 | 0 | 0 | 0 | 0 |
| Computer equipment | o _C | С | C | 0 | C |
| IT Services | C | 0 | C | 0 | 0 |
| Accommodation for visiting scholars | C | 0 | 0 | 0 | C |
| Buildings/premi ses in general | 0 | C | C | 0 | 0 |
| Power supply/ electricity | C | C | С | 0 | C |
| Communication infrastructure (phone, internet etc.) | С | С | c | С | О |

*23. Does your institution have, or engages in, any of the following

| | Yes very strong | Yes occasionally / some | No | Do not know | Not applicable |
|--|-----------------|-------------------------|----|-------------|----------------|
| Spin-offs | О | С | 0 | 0 | С |
| Research collaboration with industry | 0 | 0 | 0 | 0 | 0 |
| International research collaboration | 0 | 0 | 0 | 0 | 0 |
| Research collaboration with public/ state offices and agencies | 0 | С | 0 | c | 0 |
| Patents | 0 | 0 | C | C | C |

*24. Has your institution been involved in applications for research projects (regardless whether successful or not)?

| | Frequently as an applicant | Frequently as a partner | Occasionally as an applicant | Occasionally as partner | No involvement of application |
|---|----------------------------|-------------------------|------------------------------|-------------------------|-------------------------------|
| National, involving colleagues from another institution | c | c | c | С | C |
| Horizon 2020 / FP7 | C | C | 0 | 0 | C |
| Other European project | C | 0 | 0 | 0 | 0 |
| Other International project | 0 | 0 | 0 | 0 | C |

*25. When have you been last research active as a health professional?

(Research activity is defined here as being involved in the design or implementation of a research project either as a researcher or a supervisor)

Within the last 2 years

More than 2 years ago

Never been research active as a health professional

*26. Have you published any of the following over the last 3 years 2014-2017?

| | Yes mostly with single authorship | Yes mostly co- authored | Only supervising | No |
|---|-----------------------------------|----------------------------|------------------|----|
| ISI journal articles | 0 | 0 | C | С |
| Other journal articles (non-ISI) | 0 | C | C | С |
| Papers presented at international conferences | С | 0 | 0 | 0 |
| Papers presented at national conferences | 0 | С | 0 | 0 |
| PhD. Thesis | 0 | С | C | С |
| Books | 0 | С | C | С |
| Book chapters | 0 | 0 | 0 | C |
| Reports | 0 | C | 0 | 0 |

*27. If co-authored, who do you write with more often?

| | Colleagues from my institution | Colleagues from other institutions | | Intercontinental Colleagues | No co-authoring |
|---|--------------------------------|------------------------------------|---|-----------------------------|-----------------|
| ISI journal articles | С | C | 0 | c | С |
| Other journal articles (non-ISI) | C | С | 0 | c | С |
| Papers presented at international conferences | С | 0 | С | o | С |
| Papers presented at national conferences | С | 0 | O | 0 | 0 |
| PhD. Thesis | c | c | 0 | c | C |
| Books | C | C | 0 | c | С |
| Book chapters | c | С | 0 | c | С |
| Reports | 0 | 0 | 0 | 0 | 0 |

*28. What factors had motivated you to be research active?

| | Did not motivate me | Yes some motivation | very strong motivation | N/A |
|--------------------------------|---------------------|---------------------|------------------------|-----|
| Personal motivation', ' | c | С | С | С |
| Completing an academic degree' | 0 | C | 0 | C |

| | Did not motivate me | Yes some motivation | very strong motivation | N/A |
|-------------------------------|---------------------|---------------------|------------------------|-----|
| Professional development | 0 | C | 0 | C |
| Career advancement | 0 | 0 | 0 | c |
| Part of the professional role | 0 | 0 | 0 | C |
| For publication | 0 | 0 | 0 | 0 |
| To gain more expertise | 0 | С | C | 0 |

*29. If not active in research define the potential barriers to research engagement:

If active in research: Is there anything you can point out that prevents or discourages you from engaging in research.

| Not enough time available to engage in research |
|--|
| Clinical workload pressures |
| Lack of support/encouragement |
| Lack of funding |
| Lack of available resources for research |
| Difficulties gaining ethical approval |
| Research not valued in organisation |
| Weaknesses in research skills |
| Personal circumstances (e.g., family commitments limiting time for research) |
| Lack of research opportunities |
| Lack of supervision/mentorship |

| Difficulties collecting or gaining access to data |
|--|
| There is no reason why people should be limited or not engaged in research |

*30. What are the main problems that researchers face at your institution

| | Strongly agree | Agree to some extent | Disagree to some extend | 0 2 | Do not know | N/A |
|---|----------------|----------------------|-------------------------|-----|-------------|-----|
| There is no or insufficient research funding | 0 | 0 | 0 | 0 | 0 | 0 |
| There are no or insufficient research facilities | 0 | 0 | 0 | 0 | 0 | 0 |
| Some research facilities can only be accessed by some researchers | С | 0 | С | С | c | 0 |
| There is no or insufficient administrative capacity to prepare research proposals, manage research projects etc.) | С | 0 | С | С | 0 | 0 |
| Most researchers have no international research contacts | 0 | 0 | 0 | 0 | 0 | 0 |
| Most researchers are not internationally mobile | 0 | 0 | 0 | 0 | 0 | 0 |
| It is not possible to hire international researchers | 0 | 0 | 0 | 0 | 0 | 0 |
| Most researchers have to teach, and cannot put enough focus on their research | 0 | 0 | 0 | c | 0 | 0 |
| Most researchers have other jobs on the side and cannot put enough focus on their research | C | 0 | C | С | c | c |

^{*31.} What do you think are the reasons why people in your Institution are not, or limited engaged in research?

| Not enough time available to engage in research |
|--|
| Clinical workload pressures |
| Lack of support/encouragement |
| Lack of funding |
| Lack of available resources for research |
| Difficulties gaining ethical approval |
| Research not valued in organisation |
| Weaknesses in research skills |
| Personal circumstances (e.g., family commitments limiting time for research) |
| Lack of research opportunities |
| Lack of supervision/mentorship |
| Difficulties collecting or gaining access to data |
| There is no reason why people should be limited or not engaged in research |
| |

*32. What do you see as the main problems for research in your **country**?

| | strongly agree | agree to some extent | neither agree nor disagree | disagree | strongly disagree | Do not know |
|--|-------------------|----------------------|----------------------------------|----------|----------------------|-------------|
| Research is not given sufficiently high priority by the state | 0 | 0 | 0 | 0 | 0 | 0 |
| There is no national strategy, which would provide a clear framework | 0 | 0 | 0 | 0 | 0 | 0 |
| There is no or insufficient research funding | C | C | C | С | C | C |

| | strongly agree | agree to some extent | neither agree nor disagree | disagree | strongly disagree | Do not know |
|--|-------------------|----------------------|----------------------------------|----------|----------------------|-------------|
| There are no or insufficient research facilities | 0 | | 0 | 0 | 0 | 0 |
| Research is too much concentrated in a few institutions | 0 | 0 | 0 | C | 0 | C |
| Higher education institutions are focusing too much on teaching, and neglect research | О | О | С | c | С | C |
| Research is not sufficiently applied, and ignores the needs of the industry and society | c | C | 0 | С | 0 | О |
| There is too much focus on applied research | 0 | C | 0 | 0 | 0 | C |
| Interdisciplinary research is not sufficiently encouraged | 0 | 0 | | 0 | 0 | 0 |
| The best researchers are leaving the country, which is a problem | 0 | 0 | 0 | 0 | | 0 |
| The best researchers are leaving the country, but they still continue to Contribute to national research | 0 | c | С | С | С | С |
| Young researchers are leaving the country | 0 | 0 | 0 | 0 | 0 | 0 |
| Young talented people look for careers outside of the research and higher Education sectors | 0 | 0 | c | С | C | C |

*33. Of the changes listed in the table below, indicate how effective you believe they would for increasing the level of research activity in your Institution

Would have no effect

Would be somewhat effective

Would be greatly effective

| | Would have no effect | Would be somewhat effective | Would be greatly effective |
|--|----------------------|-----------------------------|----------------------------|
| Increased protected working time for research | C | 0 | C |
| Higher levels of ongoing research training | 0 | 0 | 0 |
| Recruiters in the health service giving more value to research experience when selecting candidates for clinical positions | 0 | С | С |
| A more simplified and efficient ethics application process | 0 | 0 | 0 |
| Greater levels of mentorship and support when conducting research | C | C | С |
| More opportunities to advance one's research qualifications (e.g. post-doctoral fellowships) | О | С | С |
| More funded research posts | C | C | C |
| More opportunities for collaboration with other clinical or academic researchers | c | С | С |

*34. What is applicable for you in the following list about research infrastructure

| | Don't know it | Know it | Collaborate with it |
|--|---------------|---------|---------------------|
| BBMRI Biobanking and Biomolecular Resources Research Infrastructure | С | О | С |
| C-ERIC Central-European Research Infrastructures Consortium | C | 0 | С |
| CLARIN Common Language Resources and Technology Infrastructure | 0 | 0 | О |
| EATRIS European Advanced Translational Research Infrastructure in Medicine | О | c | 0 |

| | Don't know it | Know it | Collaborate with it |
|---|---------------|---------|---------------------|
| ECRIN European Clinical Research Infrastructure Network | C | C | 0 |
| ELIXIR The European Life-Science Infrastructure for Biological Information | С | 0 | С |
| ERA European Research Area | 0 | C | C |
| ERIC European Research Infrastructure Consortium | C | 0 | 0 |
| ESFRI European Strategy Forum on Research Infrastructure | 0 | C | О |
| EU-OPENSCREEN European Infrastructure of Open Screening Platforms for Chemical Biology | С | О | С |
| Euro-BioImaging European Research Infrastructure for Imaging Technologies in Biological and Biomedical Sciences | С | c | О |
| INFRAFRONTIER European Research Infrastructure for the generation, phenotyping, archiving and distribution of mouse | С | 0 | o |
| ISBE Infrastructure for Systems Biology Europe | C | c | 0 |
| MIRRI Microbial Resource Research Infrastructure disease models | C | 0 | 0 |
| NRRI Research Infrastructure Roadmap | C | c | С |
| PRACE Partnership for Advanced Computing in Europe | C | c | C |
| RI Research infrastructure(s) | О | c | 0 |

*35. What is applicable for you in the following list about biobanks

| | Don't know it | Know it | Collaborate with it |
|----------------|---------------|---------|---------------------|
| Basque Biobank | C | C | С |

| | Don't know it | Know it | Collaborate with it |
|---|---------------|---------|---------------------|
| Biobanco.pt (Portuguese National Consortium of Biobanks) | 0 | 0 | 0 |
| Biobank Inherited Metabolic Disorders (blood/urine/liquor/fibroblasts) | 0 | 0 | C |
| Biobank of the Estonian genome center | 0 | 0 | 0 |
| BioNER: Biobank of the Institute of Rare Diseases Research/Institute of Health Carlos III | 0 | С | 0 |
| Brain-net – brain tissue bank Aachen | О | 0 | 0 |
| Brain-Net – brain tissue bank Munster (pediatric neuropathology) | 0 | 0 | 0 |
| Brain-Net Germany: National network of brain banks in Germany | O | 0 | 0 |
| Cell line and DNA Biobank from patients affected by genetic diseases | 0 | 0 | C |
| CIBERER Biobank (Biobank of rare diseases) | 0 | 0 | 0 |
| DNA biobank of the Department of Molecular Genetics and Diagnostics, National Institute of Environmental Health (NIEH) (EuroBioBank partner) | c | С | О |
| Genetic alliance Registry and Biobank | О | 0 | 0 |
| EuroBioBank European Network of DNA, cell and tissue banks for Rare Diseases | 0 | С | О |
| Human DNA and cell biobank of Genethon – genetic diseases, mainly neuromuscular diseases – part of the EuroBioBank network | 0 | С | c |
| Human tissue biobank of Neuromuscular diseases (EuroBioBank partner) | 0 | 0 | 0 |
| Neuromuscular Tissue Bank (NMTB) (EuroBioBank partner) | 0 | 0 | 0 |
| Telethon Network of Genetic Biobanks | 0 | 0 | C |
| UK Biobank | 0 | 0 | 0 |

*36. What is applicable for you in the following list about registry and database

| | Don't know it | Know it | Collaborate with it |
|---|---------------|---------|---------------------|
| (NCARDRS) National Congenital Anomaly and Rare Disease Registration Service | 0 | С | О |
| Austrian Country Node of the Human Variome Project (HVP) | 0 | 0 | C |
| Austrian registry for inborn errors of metabolism | 0 | C | C |
| Belgian Neuromuscular Disease Registry | 0 | 0 | C |
| Belgian patient database for Wilson disease – contributes to the EuroWilson registry (terminated) | О | С | С |
| Biobank Inherited Metabolic Disorders (blood/urine/liquor/fibroblasts) | 0 | 0 | C |
| Canadian Neuromuscular Disease Registry – part of the TREAT-NMD network | 0 | C | C |
| DDRMD – Dutch Diagnosis Registration Metabolic Diseases | 0 | C | C |
| Dutch Neuromuscular Database CRAMP: Computer Registry of All Myopathies and Polyneuropathies | 0 | С | С |
| Dutch patient registry for Fabry disease | 0 | 0 | C |
| Dutch patient registry for Gaucher disease | 0 | C | C |
| Dutch patient registry for Niemann-Pick Disease Type B | 0 | 0 | 0 |
| Dutch patient registry for Niemann-Pick Disease Type C | 0 | C | 0 |
| ECARUCA: cytogenetic and clinical database on rare chromosomal disorders | 0 | 0 | 0 |
| E-HOD European Network and Registry for Homocystinurias and Methylation Defects | 0 | c | О |
| EIMD: European registry and network for intoxication type metabolic diseases | 0 | C | 0 |

| | Don't know it | Know it | Collaborate with it |
|---|---------------|---------|---------------------|
| English alpha-1 antitrypsin deficiency registry – contributes to the Alpha One International Registry (AIR) | c | c | С |
| English cystinosis registry | C | 0 | 0 |
| English hyperoxaluria registry | С | 0 | C |
| English phenylketonuria registry | 0 | C | С |
| EPNET: European porphyria Network | 0 | С | С |
| E-RARE Consortium | 0 | 0 | 0 |
| ESNEK – Rare paediatric neurological disease registry Germany | 0 | 0 | C |
| EUNEFRON: registry of the European network for the study of orphan nephropathies | c | c | С |
| EUROCAT European Surveillance on congenital anomalies | 0 | c | C |
| EUROGLYCANET – International patient registry and cohort for congenital disorders of glycosylation | c | С | С |
| European cystinosis cohort | C | 0 | 0 |
| European Porphyria Registry (EPR) | 0 | 0 | C |
| Fabry Disease Registry –DE | 0 | c | C |
| FOS : Fabry Outcome Survey | 0 | С | C |
| French certified registry of glycogen storage disease type 2 | C | 0 | C |
| French certified registry of patients affected by Gaucher disease | 0 | 0 | 0 |
| French cystinosis registry | 0 | 0 | С |
| French registry of neuromuscular diseases from reference centres | 0 | C | C |
| Galactosemia Patient Registry – part of the Galactosemia Network | C | C | 0 |

| | Don't l | know it Know it | Collaborate with it |
|--|---------|-----------------|---------------------|
| Genetic alliance Registry and Biobank | \circ | 0 | 0 |
| German mucopolysaccharidosis patient registry | 0 | c | С |
| HOS: Hunter Outcome Survey | 0 | 0 | 0 |
| iNTD Network – International Working Group on Neurotransmitter Related Disorders | 0 | 0 | c |
| International Collaborative Gaucher Group (ICGG) Gaucher Registry – DE | 0 | 0 | 0 |
| International Morquio A registry | 0 | c | C |
| International Niemann-Pick Disease Registry | 0 | 0 | 0 |
| LOVD: GAA gene (Glucosidase, alpha, acid) variant database | 0 | c | C |
| Mendelian cytogenetics network online database | 0 | 0 | 0 |
| MITOCON: National registry mitochondrial disease | 0 | c | C |
| Mucopolysaccharidosis type 1 (MPS I) Registry – | 0 | 0 | 0 |
| NCL-Registry: International neuronal ceroid lipofuscinoses patient registry | 0 | c | C |
| NCL-Registry: International neuronal ceroid lipofuscinoses patient registry | 0 | 0 | 0 |
| Neuro-Heart-Registry: Registry for patients with heart involment in neurovascular diseases | 0 | С | О |
| NMDcy: Cypriot Neuromuscular Diseases – contributes to TREAT-NMD | 0 | О | 0 |
| NMD-ES: Spanish Registry of Neuromuscular Diseases | 0 | c | 0 |
| Norvegian Porphyria Registry | 0 | 0 | 0 |
| Pompe Disease Registry – DE | | င | 0 |
| Portuguese Fabry patient registry – contributing to the international Fabry registry | 0 | С | С |

| | Don't know it | Know it | Collaborate with it |
|--|---------------|---------|---------------------|
| RAMEDIS : Rare Metabolic Diseases Database | 0 | 0 | 0 |
| RAMEDIS: Rare Metabolic Diseases mutation Database | 0 | 0 | 0 |
| Registry for Patients with Niemann-Pick Type C Disease | 0 | 0 | C |
| Registry for Wolfram syndrome, Alström syndrome, Bardet-Biedl syndrome and other rare diabetes syndromes – contributing to EURO-WABB | 0 | 0 | С |
| Registry_Biobank Name | 0 | 0 | C |
| The International Collaborative Gaucher Group (ICGG) Gaucher registry | 0 | 0 | 0 |
| The international Fabry registry | 0 | 0 | 0 |
| The international Mps I registry | 0 | 0 | 0 |
| The international Pompe registry | 0 | 0 | 0 |
| TREAT-NMD: Accelerating Treatments for Neuromuscular Diseases (registries) | С | С | С |
| UK10K RARE NEUROMUSCULAR – Genetic neuromuscular diseases variant database | С | С | С |
| X-ALD: X-linked adrenoleukodystrophy database | 0 | 0 | 0 |

*37. What is applicable for you in the following list about scientific organisation

| | Don't know it | Know it | Collaborate with it |
|---|---------------|---------|---------------------|
| AECOM – Asociación Española para el Estudio de los Errores Congénitos del Metabolismo | c | С | С |

| | Don't know it | Know it | Collaborate with it |
|---|---------------|---------|---------------------|
| APS – Arbeitsgemeinschaft für Pädiatrische Stoffwechselstörungen (Working group for paediatric metabolic disturbances, Germany) | С | С | С |
| B4B – Brains for Brain | 0 | 0 | С |
| BIMDG – British Inherited Metabolic Disease Group | 0 | C | С |
| CLIMB – Children Living with Inherited Metabolic Diseases, UK | 0 | C | 0 |
| EBC – European Brain Council | C | C | С |
| ERNDIM Quality assurance in laboratory testing for IEM | 0 | C | C |
| ESGCT – European Society of Gene and Cell Therapy | 0 | 0 | 0 |
| EURORDIS – European Organisation for Rare Disorders | 0 | 0 | 0 |
| Fondazione Telethon | 0 | 0 | 0 |
| IBBS – International Brain Barriers Society | 0 | 0 | 0 |
| IRDIRC | 0 | 0 | 0 |
| ISNS – International Society for Neonatal Screening | 0 | 0 | C |
| SEEIM – Sociedad Española de Errores Innatos del Metabolismo | 0 | 0 | 0 |
| SFEIM – Societé Française pour l'Étude des Erreurs Innées du Métabolisme | 0 | 0 | 0 |
| SIMD – Society for Inherited Metabolic Disorders | C | 0 | 0 |
| SISMME – Societa' Italiana per lo Studio delle Malattie Metaboliche Ereditairie | 0 | 0 | 0 |
| SPDM – Sociedade Portuguesa de Doenças Metabólicas | 0 | 0 | 0 |
| SSIEM – The Society for Study of Inborn Errors of Metabolism | 0 | 0 | 0 |



Survey MetabERN for Laboratories

*1. Can you please type in your contact details

Name

Email Address

Phone Number

*2. Please select the name of your hospital /institute from the drop down menu

| 3. Please select what is ap orange male female | plicable to your situation | |
|---|---|--|
| 4. Academic degrees (if y | ou have several, just the once that | are most important for your researcher |
| career) | | |
| | Year of award | Country of award |
| Undergraduate degree (or equivalent) | Undergraduate degree (or equivalent) Year of award menu | Undergraduate degree (or equivalent) Country of award menu |
| Master degree (or equivalent) | Master degree (or equivalent) Year of award menu | Master degree (or equivalent) Country of award menu |
| Doctorate | Doctorate Year of award menu | Doctorate Country of award menu |
| Specialization Degree | Specialization Degree Year of award menu | Specialization Degree Country of award menu |
| 5. Type of institution | | |

| | Public university Private university Public college Private college Research institute |
|-------|---|
| * | 6. What is your position |
| 00000 | Professor Associate Professor Lecturer Senior Research Fellow Postdoc/Research Fellow Research Assistant Teaching Assistant Dean, head of department or other senior administration position Other (please specify) |
| | What type of research do you perform? Animal research Basic research Preclinical research |

| Clinical research | | | | | | | |
|--|-------------|-----------------|---------------------|----------------------|---------------|--|--|
| *8. Please indicate which are, the main | research a | activities p | erformed is | nside your | Institution | | |
| | No activity | Low activity | Rather low activity | Rather high activity | High activity | | |
| Health Systems Research | 0 | 0 | C | 0 | 0 | | |
| Human and Social Science Research | 0 | C | 0 | 0 | С | | |
| Epidemiology/Natural History of diseases | 0 | C | С | 0 | C | | |
| Research in Genetics and Pathophysiology | С | С | C | C | 0 | | |
| Pre-Clinical Therapeutic Research | 0 | C | 0 | 0 | 0 | | |
| Therapeutic Research | О | C | C | C | C | | |
| Research Platforms/Infrastructures | 0 | C | 0 | 0 | C | | |
| Development and availability of tools needed to accelerate research on rare diseases | С | С | 0 | c | С | | |
| *9. Academic Health Systems Research | | | | | | | |
| | No activit | ty Low activity | Rather low activity | Rather high activity | High activity | | |
| Research policy (e.g., guidelines development; national plan health care resources) | ns; | С | С | c | С | | |
| Cost analysis (e.g., guidelines implementation) | О | С | О | О | С | | |

| | No activity | Low activity | Rather low activity | Rather high activity | High activity |
|--|-------------|--------------|---------------------|----------------------|---------------|
| New technologies (e.g., New diagnostic tests, population screening programmes, Innovative therapeutics) | О | 0 | 0 | 0 | С |
| Health-Technology assessment of Orphan Drugs | 0 | 0 | 0 | 0 | C |
| e-Health activities | С | C | C | C | С |

*10. **Industrial** Health Systems Research

| | No activity | Low activity | Rather low activity | Rather high activity | High activity |
|--|-------------|--------------|---------------------|----------------------|---------------|
| Research policy (e.g., guidelines development; national plans; health care resources) | С | С | С | С | С |
| Cost analysis (e.g., guidelines implementation) | 0 | 0 | 0 | 0 | 0 |
| New technologies (e.g., New diagnostic tests, population screening programmes, Innovative therapeutics) | c | c | С | С | С |
| Health-Technology assessment of Orphan Drugs | 0 | 0 | 0 | 0 | 0 |
| e-Health activities | 0 | 0 | 0 | 0 | 0 |

*11. Human and social science research

| | No activity | Low activity | Rather low activity | Rather high activity | High activity |
|---|-------------|--------------|---------------------|----------------------|---------------|
| Society and RD (e.g., social perception, accessibility to care,) | С | С | С | С | c |
| RD, Research and Innovation (e.g., public/private cooperation, role of patients org,) | C | O | C | C | С |
| Care practices (self- management, health education policy,) | c | o | c | c | С |
| Patients awareness (e.g., understanding genetic data, importance of animal models,) | C | С | С | С | С |
| Support for caregivers | 0 | C | 0 | 0 | C |
| Public Heath and RD (e.g., health care policies, prospective public policies,) | О | О | C | C | С |
| Methods for social support to improve quality of life | 0 | 0 | 0 | 0 | C |
| Services accessibility | 0 | 0 | 0 | 0 | 0 |
| Orphan Drugs availability | 0 | 0 | 0 | 0 | 0 |
| Methods to assess QoL | 0 | 0 | 0 | 0 | 0 |

*12. Epidemiology & Natural History of diseases

| | No activity | Low activity | Rather low activity | Rather high activity | High activity |
|---------------------------------------|-------------|--------------|---------------------|----------------------|---------------|
| Registries | C | C | 0 | 0 | C |
| Databases/data management systems for | 0 | 0 | 0 | 0 | 0 |

| | No activity | Low activity | Rather low activity | Rather high activity | High activity |
|--|-------------|--------------|---------------------|----------------------|---------------|
| epidemiology studies | | | | | |
| Cohorts | 0 | C | C | 0 | C |
| Biomarkers to assess diagnosis, prognosis and severity of diseases | С | С | С | С | С |
| Diagnosis/prognosis methods (e.g., other than biomarkers) | С | С | o | c | С |
| Identification of new entities | 0 | 0 | 0 | 0 | 0 |
| Networks for ultra rare diseases | C | 0 | 0 | 0 | 0 |

*13. Research in genetics and physiopathology

| | No activity | Low activity | Rather low activity | Rather high activity | High activity |
|--|-------------|--------------|---------------------|----------------------|---------------|
| Gene identification and Molecular Mechanisms | 0 | C | 0 | 0 | 0 |
| New database method developments to be applied to omics research | С | С | С | С | С |
| Biobanking research | 0 | 0 | 0 | 0 | 0 |
| High throughput genomics/sequencing | 0 | C | C | 0 | 0 |
| New bioinformatic methods and developments | 0 | C | 0 | 0 | 0 |
| New technologies (e.g., atomic microscopy) | 0 | 0 | 0 | 0 | 0 |
| Omics technology (e.g., proteo, metabolo) | 0 | C | C | C | 0 |

| | No activity | Low activity | Rather low activity | Rather high activity | High activity |
|--------------------------------------|-------------|--------------|---------------------|----------------------|---------------|
| Development of new animal models | 0 | 0 | 0 | 0 | 0 |
| New Imaging/Microscopy methods | C | 0 | 0 | 0 | 0 |
| Development of new therapeutic tools | 0 | 0 | 0 | 0 | 0 |

*14. Pre-clinical therapeutic research

| | No activity | Low activity | Rather low activity | Rather high activity | High activity |
|---|-------------|--------------|---------------------|----------------------|---------------|
| Proof-of-concept | 0 | 0 | 0 | 0 | 0 |
| Drug libraries | C | C | C | 0 | 0 |
| Search for active chemical compounds | 0 | C | 0 | 0 | 0 |
| High throughput drug screening | C | C | C | 0 | 0 |
| Therapeutic target identification/validation (e.g.,. stem cells, systems biology) | С | С | 0 | О | О |
| Cell therapy research (e.g., viral vectors) | C | C | C | C | 0 |
| Innovative biotechnological research (e.g., monoclonal antibodies, cell and gene therapy, enzyme therapy) | 0 | c | c | o | o |
| Drug toxicology/pharmacology | C | С | С | C | C |

*15. Therapeutic research

| | No activity | Low activity | Rather low activity | Rather high activity | High activity |
|--|-------------|--------------|---------------------|----------------------|---------------|
| Drug development | 0 | 0 | 0 | 0 | C |
| Phase I/II Clinical Trials | 0 | 0 | 0 | 0 | 0 |
| Bioproduction centres | 0 | 0 | 0 | 0 | 0 |
| Epidemiological/clinical/ therapeutical investigations | С | 0 | О | О | С |
| Medical devices | c | C | 0 | 0 | C |
| Assesment of surgery procedures | 0 | 0 | 0 | 0 | C |
| Transplants | 0 | 0 | 0 | 0 | C |
| Pharma/academic partnerships | 0 | 0 | 0 | 0 | 0 |

*16. Platforms / Infrastructures development

| | No activity | Low activity | Rather low activity | Rather high activity | High activity |
|-----------------------------------|-------------|--------------|---------------------|----------------------|---------------|
| Bioinformatics | 0 | c | 0 | C | 0 |
| Pharmacogenomic diagnostic assays | 0 | 0 | 0 | 0 | 0 |
| Population Registries | c | 0 | 0 | 0 | C |
| Genetically-modified animal model | 0 | 0 | 0 | 0 | 0 |

| | No activity | Low activity | Rather low activity | Rather high activity | High activity |
|--------------------------------------|-------------|--------------|---------------------|----------------------|---------------|
| repositories | | | | | |
| GMP platforms | C | 0 | C | C | C |
| Biobanking | 0 | C | 0 | C | C |
| Genomic and proteomic platforms | C | C | C | 0 | C |
| Long-term cohort of patients studies | 0 | C | O | 0 | C |
| High throughput drug screening | c | C | 0 | 0 | C |

*17. Human resources

| | No activity | Low activity | Rather low activity | Rather high activity | High activity |
|--------------------------------------|-------------|--------------|---------------------|----------------------|---------------|
| Rotational positions for clinicians | C | 0 | С | C | C |
| Rotational positions for researchers | С | С | С | c | 0 |
| Specific training in RD | c | C | С | 0 | c |
| Training in bioinformatics | o | c | o | o | 0 |

^{*18.} Development and availability of tools needed to accelerate research on rare disease.

| | No activity | Low activity | Rather low activity | Rather high activity | High activity |
|--|-------------|--------------|---------------------|----------------------|---------------|
| Network development | 0 | 0 | 0 | 0 | C |
| Creation of transnational research consortiums | 0 | 0 | 0 | 0 | С |
| Transnational calls for projects funding | 0 | 0 | 0 | C | С |
| Opening of national research programmes to international collaboration | О | С | С | С | О |
| Transnational research infrastructures | 0 | 0 | 0 | C | C |
| Development of an inventory/directory of databases of experts | О | О | O | С | О |
| Transnational collection of funded RD research projects | c | 0 | o | c | 0 |

*19. How do you usually conduct your research activities

| | Never | Seldom | Sometimes | Very commonly | / N/A |
|---|-------|--------|-----------|---------------|-------|
| Alone | 0 | 0 | 0 | 0 | С |
| Within a research group/team | 0 | 0 | C | C | C |
| Collaboration with colleagues from my institution | 0 | 0 | 0 | 0 | C |
| Collaboration with colleagues from other institutions in my country | O | О | O | O | С |
| Collaboration with international colleagues at EU level | 0 | 0 | 0 | o | С |
| Collaboration with intercontinental colleagues | 0 | 0 | C | C | C |
| Collaboration with domestic industry/Biotech Company | О | С | О | С | О |

| | Never | Seldom | Sometimes | Very commonly | N/A |
|--|-------|--------|-----------|---------------|-----|
| Collaboration with international industry/Biotech Company | 0 | 0 | 0 | 0 | c |
| Collaboration with local Patients Organizations | C | 0 | C | C | С |
| Collaboration with international Patients Organizations | 0 | С | 0 | 0 | С |
| Collaboration with local Scientific Organizations | C | 0 | C | C | С |
| Collaboration with international | 0 | 0 | 0 | 0 | 0 |
| Scientific Organizations | 0 | C | С | С | С |

*20. Do you have access to the following? If yes, how would you rate the quality?

| | International standard or close to it | Good national standard | Insufficient | Do not know | Not applicable |
|--|---------------------------------------|------------------------|--------------|-------------|----------------|
| Research support services / unit | 0 | 0 | C | 0 | 0 |
| Research laboratories | C | C | 0 | 0 | 0 |
| Teaching labs (if different from research Labs) | c | c | 0 | o | С |
| Large research equipment that requires technicians | С | 0 | О | О | С |
| Scientific databases (e.g. ISI Web of Knowledge) | 0 | С | c | c | С |
| Knowledge Transfer Office | 0 | 0 | 0 | 0 | 0 |

| | International standard or close to it | Good national standard | Insufficient | Do not know | Not applicable |
|---|---------------------------------------|------------------------|--------------|-------------|----------------|
| Science Park | 0 | C | 0 | C | C |
| Research ethics committee | 0 | С | C | С | С |
| Libraries | C | C | C | С | С |
| Research publication repositories | 0 | О | O | С | С |
| Computer equipment | 0 | 0 | 0 | C | С |
| IT Services | 0 | 0 | 0 | C | C |
| Accommodation for visiting scholars | 0 | 0 | 0 | C | C |
| Buildings/premi ses in general | 0 | O | 0 | 0 | 0 |
| Power supply/ electricity | 0 | 0 | 0 | C | C |
| Communication infrastructure (phone, internet etc.) | O | С | 0 | c | 0 |

*21. Which of the following describes your situation

| | Strongly agree | Agree to some extent | Disagree to some extend | Strongly disagree | Do not know | N/A |
|---|----------------|----------------------|-------------------------|-------------------|-------------|-----|
| I have access to the necessary research infrastructure (labs, | C | 0 | 0 | 0 | 0 | 0 |
| computers, library etc.) to do my research | 0 | 0 | 0 | 0 | 0 | 0 |
| I have the necessary staff support to do my research | 0 | 0 | 0 | 0 | 0 | C |

| | Strongly agree | Agree to some extent | Disagree to some extend | • • | Do not know | N/A |
|---|----------------|----------------------|-------------------------|-----|-------------|-----|
| I have the necessary funding to do my research | С | 0 | 0 | 0 | 0 | 0 |
| In my position, I have a high level of autonomy in developing my research | c | 0 | c | c | 0 | 0 |
| I am involved in interdisciplinary research collaboration within my institution | 0 | 0 | 0 | C | 0 | 0 |
| I am involved in interdisciplinary research collaboration within colleagues from other institutions | 0 | 0 | c | C | 0 | 0 |
| I am involved in spin-off activities, research incubators or similar | 0 | 0 | 0 | 0 | 0 | 0 |
| I am/have been involved in patents | 0 | 0 | 0 | 0 | 0 | 0 |
| I frequently attend international conferences | 0 | 0 | 0 | 0 | 0 | 0 |
| I am very dedicated to my work | 0 | 0 | C | C | 0 | 0 |
| I am satisfied with my career | 0 | 0 | 0 | 0 | 0 | 0 |
| I am satisfied with my salary | 0 | 0 | C | C | 0 | 0 |
| I am proud to work at my institution/organisation | О | 0 | 0 | 0 | 0 | 0 |
| My working situation is difficult, due to a high workload | 0 | 0 | 0 | C | 0 | 0 |
| My working situation is difficult, due to bureaucracy, administration | 0 | 0 | 0 | 0 | 0 | 0 |

^{*22.} Have you published any of the following over the last 3 years 2014-2017?

| | Yes mostly with single authorship | Yes mostly co- authored | Only supervising | No |
|---|-----------------------------------|----------------------------|------------------|----|
| ISI journal articles | 0 | 0 | 0 | C |
| Other journal articles (non-ISI) | C | C | 0 | 0 |
| Papers presented at international conferences | 0 | 0 | С | С |
| Papers presented at national conferences | 0 | С | 0 | 0 |
| PhD. Thesis | 0 | С | c | C |
| Books | C | С | С | О |
| Book chapters | О | С | c | О |
| Reports | 0 | C | 0 | 0 |

*23. If co-authored, who do you write with more often?

| | Colleagues from my institution | Colleagues from other institutions | European Colleagues | Intercontinental Colleagues | No co-authoring |
|---|--------------------------------|------------------------------------|------------------------|--------------------------------|-----------------|
| ISI journal articles | C | 0 | С | 0 | С |
| Other journal articles (non-ISI) | C | C | C | C | C |
| Papers presented at international conferences | 0 | С | c | c | 0 |
| Papers presented at national conferences | С | С | С | С | С |
| PhD. Thesis | C | 0 | 0 | 0 | C |

| Books C C C C C C C Book chapters C C C C C C C Reports C C C C C C *24. Have you been involved in applications for research projects (regardless wether success or not)? Frequently as an applicant partner anapplicant of application of applica | | Colleagues from my institution | Colleagues from other institutions | European Colleagues | Intercontinental Colleagues | No co-authoring | | | |
|--|-----------------------------|--------------------------------|------------------------------------|------------------------|-----------------------------|-----------------|--|--|--|
| *24. Have you been involved in applications for research projects (regardless wether success or not)? Frequently as an applicant partner of application of application National, involving colleagues from another institution National project | Books | o | c | 0 | _ | | | | |
| *24. Have you been involved in applications for research projects (regardless wether success or not)? Frequently as an applicant partner of application of | Book chapters | C | 0 | 0 | 0 | C | | | |
| Frequently as an applicant partner of application National, involving colleagues from another institution Horizon 2020 / FP7 Other European project Other International project Less than 1 year Frequently as an partner partner of application Occasionally as No involvement of application | Reports | 0 | 0 | c | 0 | 0 | | | |
| National, involving colleagues from another institution Horizon 2020 / FP7 C C C C C C C C C C C C C C C C C C | | | | | | | | | |
| #25. Have you ever worked as a researcher for the following period? Less than 1 year Between 1 and 2 years C C C C C C C C C C C C C C C C C C C | | applicant | partner | _ | | of application | | | |
| Other European project C C C C C C C C C C C C C C C C C C | | О | О | О | c | О | | | |
| Other International project *25. Have you ever worked as a researcher for the following period? Less than 1 year Between 1 and 2 years More than 2 years | Horizon 2020 / FP7 | c | 0 | 0 | 0 | c | | | |
| *25. Have you ever worked as a researcher for the following period? Less than 1 year Between 1 and 2 years More than 2 years | Other European project | c | 0 | 0 | 0 | c | | | |
| Less than 1 year Between 1 and 2 years More than 2 years | Other International project | 0 | 0 | 0 | 0 | c | | | |
| | | | | | | | | | |
| III JOHI VOMINI, | In your country | | С | | О | , | | | |

| | Less than 1 year | Between 1 and 2 years | More than 2 years |
|----------------------------------|------------------|-----------------------|-------------------|
| In the EU (outside your country) | 0 | c | c |
| In the US | С | С | С |
| both in the EU and the US | 0 | О | С |
| neither in the EU nor in the US | 0 | С | С |

*26. Evaluate your research skills using the table below

| | very weak | weak | average strong | strong | very strong | don't know |
|---------------------------------|-----------|------|----------------|--------|-------------|------------|
| Handling routine data | C | 0 | 0 | 0 | 0 | С |
| Generating a research idea | 0 | 0 | 0 | 0 | 0 | 0 |
| Conducting a literature review | 0 | 0 | 0 | 0 | 0 | 0 |
| Applying for ethics approval | C | 0 | О | C | С | 0 |
| Applying for funding | 0 | 0 | 0 | 0 | 0 | 0 |
| Designing quantitative research | C | 0 | О | С | С | О |
| Designing qualitative research | 0 | 0 | 0 | 0 | O | C |
| Recruiting participants | C | 0 | О | С | С | О |
| Collecting data | 0 | 0 | O | 0 | O | C |
| Quantitative data analysis | 0 | C | О | С | С | 0 |
| Qualitative data analysis | 0 | 0 | 0 | 0 | 0 | 0 |

| | very weak | weak | average strong | strong | very strong | don't know |
|--|-----------|------|-------------------|--------|-------------|------------|
| Presenting the results | 0 | 0 | 0 | 0 | 0 | \circ |
| Critical appraisal of research | 0 | 0 | 0 | 0 | 0 | 0 |
| Orally presenting research | C | C | 0 | 0 | 0 | 0 |
| Publishing research | 0 | 0 | | 0 | 0 | 0 |
| Teaching colleagues and/or students | C | 0 | 0 | 0 | 0 | 0 |
| Working as a member of a team doing research | C | C | C | 0 | C | 0 |
| Accessing resources to undertake research (e.g, money, information, equipment) | 0 | 0 | C | С | 0 | 0 |
| Undertaking administrative activities | 0 | 0 | 0 | C | 0 | C |

*27. How do you rate the following in relation to the motivation to do research abroad?

| | not important | quite important | important | very important | extremely important |
|--|---------------|-----------------|-----------|----------------|---------------------|
| Learn different Language | 0 | 0 | C | 0 | C |
| Career progression goals | 0 | 0 | C | 0 | C |
| Access to knowledge not | C | 0 | C | 0 | c |
| available in your country | 0 | 0 | 0 | О | С |
| Getting access to the facilities / equipment necessary | С | С | С | c | 0 |
| to your research | C | C | C | C | С |
| Opportunities to collaborate | 0 | 0 | C | 0 | c |

| | not important | quite important | important | very important | extremely important |
|--|---------------|-----------------|-----------|----------------|---------------------|
| with top-class researchers | 0 | 0 | C | 0 | 0 |
| More attractive remuneration | 0 | 0 | 0 | 0 | 0 |
| schemes | 0 | 0 | 0 | C | C |
| More opportunities to work in the | 0 | 0 | C | 0 | 0 |
| industry | 0 | 0 | C | 0 | 0 |
| Obtaining funding for own research | 0 | 0 | C | 0 | 0 |
| Availability of adequate schools you're your children | c | c | 0 | 0 | 0 |
| Work opportunities for your partner (and other family members) | С | С | С | c | С |
| Quality and cost of accommodation | C | C | 0 | | C |
| Personal interest in the culture of foreign country | С | С | С | c | С |

*28. Please rank the effect of the international mobility on your research career

| | remained unchanged | a little bit improved | improved | strongly improved | N/A |
|--|--------------------|-----------------------|----------|-------------------|-----|
| Publication output | C | C | 0 | 0 | C |
| Patient output | 0 | 0 | 0 | 0 | 0 |
| Ability to work in the industrial sector | C | С | С | C | 0 |

| | remained unchanged | a little bit improved | improved | strongly improved | N/A |
|--|--------------------|-----------------------|----------|-------------------|-----|
| Access to infrastructure and know-how | 0 | 0 | 0 | 0 | 0 |
| Access to an international network of professionals active in your field | c | 0 | 0 | c | С |
| Professional experience as researcher | C | C | 0 | С | 0 |
| Future job opportunities in the country where you have previously worked/studied | C | c | C | c | С |
| "Recognition" in the research community as a researcher | С | c | 0 | С | С |

*29. What are the main problems that researchers face at your **institution**?

| | Strongly agree | Agree to some extent | Disagree to some extend | | Do not know | N/A |
|---|----------------|----------------------|-------------------------|---|----------------|-----|
| There is no or insufficient research funding | 0 | 0 | 0 | 0 | 0 | C |
| There are no or insufficient research facilities | 0 | 0 | 0 | O | 0 | 0 |
| Some research facilities can only be accessed by some researchers | С | 0 | 0 | c | 0 | c |
| There is no or insufficient administrative capacity to prepare research proposals, manage research projects etc.) | С | 0 | С | С | С | О |
| Most researchers have no international research contacts | 0 | 0 | 0 | 0 | C | 0 |
| Most researchers are not internationally mobile | 0 | 0 | 0 | 0 | C | 0 |
| It is not possible to hire international researchers | C | C | 0 | C | C | C |

| | Strongly agree | Agree to some extent | Disagree to some extend | Strongly disagree | Do not know | N/A |
|--|----------------|----------------------|-------------------------|-------------------|----------------|-----|
| Most researchers have to teach, and cannot put enough focus on their research | C | О | С | С | О | О |
| Most researchers have other jobs on the side and cannot put enough focus on their research | 0 | c | c | 0 | 0 | 0 |

*30. What do you see as the main problems for research in your **country**?

| | strongly agree | agree to some extent | neither agree nor disagree | disagree | strongly disagree | Do not know |
|---|-------------------|----------------------|----------------------------------|----------|----------------------|----------------|
| Research is not given sufficiently high priority by the state | 0 | 0 | 0 | 0 | 0 | C |
| There is no national strategy, which would provide a clear framework | 0 | 0 | 0 | 0 | 0 | 0 |
| There is no or insufficient research funding | 0 | 0 | 0 | 0 | 0 | 0 |
| There are no or insufficient research facilities | 0 | | 0 | 0 | 0 | 0 |
| Research is too much concentrated in a few institutions | 0 | 0 | 0 | 0 | 0 | C |
| Higher education institutions are focusing too much on teaching, and neglect research | С | 0 | С | С | С | С |
| Research is not sufficiently applied, and ignores the needs of the industry and society | С | 0 | c | c | С | С |
| There is too much focus on applied research | \circ | 0 | 0 | 0 | 0 | 0 |
| Interdisciplinary research is not sufficiently encouraged | | 0 | 0 | 0 | 0 | C |

| | strongly agree | agree to some extent | neither agree nor disagree | disagree | strongly disagree | Do not know |
|--|-------------------|----------------------|----------------------------------|-----------|----------------------|-------------|
| The best researchers are leaving the country, which is a problem | С | 0 | О | C | | О |
| The best researchers are leaving the country, but they still continue to Contribute to national research | 0 | 0 | 0 | 0 | 0 | 0 |
| Young researchers are leaving the country | 0 | 0 | 0 | 0 | 0 | 0 |
| Young talented people look for careers outside of the research and higher Education sectors | С | C | С | C | 0 | С |
| *31. What do you think are the reason in research? Not enough time available to engage in research Clinical workload pressures Lack of support/encouragement Lack of funding Lack of available resources for research Difficulties gaining ethical approval Research not valued in organisation Weaknesses in research skills | s why por | | | actor are | 1101, 01 11 | |

| Difficulties collecting or gaining access to data |
|--|
| There is no reason why people should be limited or not engaged in research |
| |

*32. What do you see as the main problems for research in your country?

| | strongly agree | agree to some extent | neither agree nor disagree | disagree | strongly disagree | Do not know |
|--|-------------------|----------------------|----------------------------------|----------|----------------------|-------------|
| Research is not given sufficiently high priority by the state | 0 | 0 | 0 | 0 | 0 | C |
| There is no national strategy, which would provide a clear framework | 0 | 0 | О | 0 | О | 0 |
| There is no or insufficient research funding | 0 | 0 | 0 | 0 | 0 | 0 |
| There are no or insufficient research facilities | 0 | | C | 0 | 0 | 0 |
| Research is too much concentrated in a few institutions | 0 | 0 | 0 | 0 | 0 | 0 |
| Higher education institutions are focusing too much on teaching, and neglect research | О | 0 | 0 | 0 | 0 | О |
| Research is not sufficiently applied, and ignores the needs of the industry and society | c | 0 | 0 | c | 0 | С |
| There is too much focus on applied research | 0 | 0 | 0 | 0 | 0 | 0 |
| Interdisciplinary research is not sufficiently encouraged | 0 | 0 | 0 | 0 | 0 | 0 |
| The best researchers are leaving the country, which is a problem | 0 | 0 | 0 | 0 | | 0 |
| The best researchers are leaving the country, but they still continue to Contribute to national research | 0 | 0 | 0 | 0 | 0 | С |

| | strongly agree | agree to some extent | neither agree nor disagree | disagree | strongly disagree | Do not know |
|---|-------------------|----------------------|----------------------------------|----------|----------------------|----------------|
| Young researchers are leaving the country | 0 | 0 | 0 | 0 | C | 0 |
| Young talented people look for careers outside of the research and higher Education sectors | c | c | С | o | С | С |

*33. Of the changes listed in the table below, indicate how effective you believe they would for increasing the level of research activity in your Institution

| | Would have no effect | Would be somewhat effective | Would be greatly effective |
|--|----------------------|-----------------------------|----------------------------|
| Increased protected working time for research | C | C | C |
| Higher levels of ongoing research training | 0 | С | С |
| Recruiters in the health service giving more value to research experience when selecting candidates for clinical positions | О | С | С |
| A more simplified and efficient ethics application process | C | C | C |
| Greater levels of mentorship and support when conducting research | 0 | C | 0 |
| More opportunities to advance one's research qualifications (e.g. post-doctoral fellowships) | О | О | О |
| More funded research posts | 0 | 0 | 0 |
| More opportunities for collaboration with other clinical or academic researchers | 0 | O | O |

*34. What is applicable for you in the following list about **Research Infrastructure**.

| | Don't know it | Know it | Collaborate with it |
|---|---------------|---------|---------------------|
| BBMRI Biobanking and Biomolecular Resources Research Infrastructure | С | С | С |
| C-ERIC Central-European Research Infrastructures Consortium | C | C | 0 |
| CLARIN Common Language Resources and Technology Infrastructure | С | С | О |
| EATRIS European Advanced Translational Research Infrastructure in Medicine | С | С | c |
| ECRIN European Clinical Research Infrastructure Network | c | C | 0 |
| ELIXIR The European Life-Science Infrastructure for Biological Information | С | С | С |
| ERA European Research Area | С | C | C |
| ERIC European Research Infrastructure Consortium | C | C | 0 |
| ESFRI European Strategy Forum on Research Infrastructure | C | C | 0 |
| EU-OPENSCREEN European Infrastructure of Open Screening Platforms for Chemical Biology | С | С | О |
| Euro-BioImaging European Research Infrastructure for Imaging Technologies in Biological and Biomedical Sciences | С | С | С |
| INFRAFRONTIER European Research Infrastructure for the generation, phenotyping, archiving and distribution of mouse | С | С | С |
| ISBE Infrastructure for Systems Biology Europe | 0 | C | 0 |
| MIRRI Microbial Resource Research Infrastructure disease models | C | C | 0 |
| NRRI Research Infrastructure Roadmap | C | C | c |

| | Don't know it | Know it | Collaborate with it |
|--|---------------|---------|---------------------|
| PRACE Partnership for Advanced Computing in Europe | 0 | 0 | 0 |
| RI Research infrastructure(s) | 0 | 0 | 0 |

*35. What is applicable for you in the following list about **Biobanks**.

| | Don't know it | Know it | Collaborate with it |
|---|---------------|---------|---------------------|
| Basque Biobank | 0 | C | C |
| Biobanco.pt (Portuguese National Consortium of Biobanks) | 0 | С | C |
| Biobank Inherited Metabolic Disorders (blood/urine/liquor/fibroblasts) | 0 | С | C |
| Biobank of the Estonian genome center | 0 | C | O |
| BioNER: Biobank of the Institute of Rare Diseases Research/Institute of Health Carlos III | С | С | С |
| Brain-net – brain tissue bank Aachen | C | C | 0 |
| Brain-Net – brain tissue bank Munster (pediatric neuropathology) | C | С | C |
| Brain-Net Germany: National network of brain banks in Germany | 0 | С | C |
| Cell line and DNA Biobank from patients affected by genetic diseases | 0 | С | C |
| CIBERER Biobank (Biobank of rare diseases) | 0 | C | C |
| DNA biobank of the Department of Molecular Genetics and Diagnostics, National Institute of Environmental Health (NIEH) (EuroBioBank partner) | С | С | С |
| Genetic alliance Registry and Biobank | 0 | 0 | 0 |

| | Don't know it | Know it | Collaborate with it |
|--|---------------|---------|---------------------|
| EuroBioBank European Network of DNA, cell and tissue banks for Rare Diseases | 0 | o | 0 |
| Human DNA and cell biobank of Genethon – genetic diseases, mainly neuromuscular diseases – part of the EuroBioBank network | 0 | С | О |
| Human tissue biobank of Neuromuscular diseases (EuroBioBank partner) | 0 | c | С |
| Neuromuscular Tissue Bank (NMTB) (EuroBioBank partner) | 0 | 0 | С |
| Telethon Network of Genetic Biobanks | 0 | С | С |
| UK Biobank | 0 | 0 | 0 |

*36. What is applicable for you in the following list about **Registry and database**.

| | Don't know it | Know it | Collaborate with it |
|---|---------------|---------|---------------------|
| (NCARDRS) National Congenital Anomaly and Rare Disease Registration Service | 0 | 0 | С |
| Austrian Country Node of the Human Variome Project (HVP) | C | C | C |
| Austrian registry for inborn errors of metabolism | 0 | C | C |
| Belgian Neuromuscular Disease Registry | О | C | 0 |
| Belgian patient database for Wilson disease – contributes to the EuroWilson registry (terminated) | С | С | С |
| Biobank Inherited Metabolic Disorders (blood/urine/liquor/fibroblasts) | О | C | 0 |
| Canadian Neuromuscular Disease Registry – part of the TREAT-NMD network | 0 | С | C |

| | Don't know it | Know it | Collaborate with it |
|---|---------------|---------|---------------------|
| DDRMD – Dutch Diagnosis Registration Metabolic Diseases | 0 | 0 | 0 |
| Dutch Neuromuscular Database CRAMP: Computer Registry of All Myopathies and Polyneuropathies | c | 0 | 0 |
| Dutch patient registry for Fabry disease | C | 0 | С |
| Dutch patient registry for Gaucher disease | О | C | C |
| Dutch patient registry for Niemann-Pick Disease Type B | C | 0 | С |
| Dutch patient registry for Niemann-Pick Disease Type C | 0 | 0 | c |
| ECARUCA: cytogenetic and clinical database on rare chromosomal disorders | 0 | 0 | С |
| E-HOD European Network and Registry for Homocystinurias and Methylation Defects | c | О | o |
| EIMD: European registry and network for intoxication type metabolic diseases | C | 0 | С |
| English alpha-1 antitrypsin deficiency registry – contributes to the Alpha One International Registry (AIR) | 0 | С | o |
| English cystinosis registry | 0 | 0 | 0 |
| English hyperoxaluria registry | 0 | C | c |
| English phenylketonuria registry | 0 | 0 | С |
| EPNET: European porphyria Network | 0 | C | c |
| E-RARE Consortium | 0 | C | С |
| ESNEK – Rare paediatric neurological disease registry Germany | 0 | C | С |
| EUNEFRON: registry of the European network for the study of orphan nephropathies | 0 | 0 | 0 |
| EUROCAT European Surveillance on congenital anomalies | 0 | 0 | С |

| | Don't know it | Know it | Collaborate with it |
|--|---------------|---------|---------------------|
| EUROGLYCANET – International patient registry and cohort for congenital disorders of glycosylation | c | 0 | c |
| European cystinosis cohort | 0 | c | 0 |
| European Porphyria Registry (EPR) | C | 0 | 0 |
| Fabry Disease Registry –DE | C | C | C |
| FOS : Fabry Outcome Survey | C | 0 | O |
| French certified registry of glycogen storage disease type 2 | 0 | c | 0 |
| French certified registry of patients affected by Gaucher disease | 0 | 0 | 0 |
| French cystinosis registry | 0 | c | 0 |
| French registry of neuromuscular diseases from reference centres | 0 | 0 | 0 |
| Galactosemia Patient Registry – part of the Galactosemia Network | C | c | 0 |
| Genetic alliance Registry and Biobank | 0 | 0 | 0 |
| German mucopolysaccharidosis patient registry | C | c | 0 |
| HOS: Hunter Outcome Survey | 0 | 0 | O |
| iNTD Network – International Working Group on Neurotransmitter Related Disorders | С | С | С |
| International Collaborative Gaucher Group (ICGG) Gaucher Registry – DE | 0 | C | 0 |
| International Morquio A registry | C | 0 | C |
| International Niemann-Pick Disease Registry | С | 0 | 0 |
| LOVD: GAA gene (Glucosidase, alpha, acid) variant database | С | 0 | 0 |
| Mendelian cytogenetics network online database | О | 0 | С |

| | Don't know it | Know it | Collaborate with it |
|--|---------------|---------|---------------------|
| MITOCON: National registry mitochondrial disease | 0 | 0 | C |
| Mucopolysaccharidosis type 1 (MPS I) Registry – | 0 | 0 | 0 |
| NCL-Registry: International neuronal ceroid lipofuscinoses patient registry | 0 | 0 | 0 |
| NCL-Registry: International neuronal ceroid lipofuscinoses patient registry | 0 | 0 | 0 |
| Neuro-Heart-Registry: Registry for patients with heart involment in neurovascular diseases | c | c | С |
| NMDcy: Cypriot Neuromuscular Diseases – contributes to TREAT-NMD | 0 | 0 | 0 |
| NMD-ES: Spanish Registry of Neuromuscular Diseases | 0 | 0 | C |
| Norvegian Porphyria Registry | 0 | 0 | 0 |
| Pompe Disease Registry – DE | C | C | 0 |
| Portuguese Fabry patient registry – contributing to the international Fabry registry | С | С | С |
| RAMEDIS : Rare Metabolic Diseases Database | 0 | 0 | C |
| RAMEDIS: Rare Metabolic Diseases mutation Database | 0 | C | 0 |
| Registry for Patients with Niemann-Pick Type C Disease | C | C | C |
| Registry for Wolfram syndrome, Alström syndrome, Bardet-Biedl syndrome and other rare diabetes syndromes – contributing to EURO-WABB | С | О | С |
| Registry_Biobank Name | 0 | C | С |
| The International Collaborative Gaucher Group (ICGG) Gaucher registry | C | 0 | C |
| The international Fabry registry | 0 | c | C |
| The international Mps I registry | 0 | C | О |
| The international Pompe registry | 0 | C | C |

| | Don't know it | Know it | Collaborate with it |
|--|---------------|---------|---------------------|
| TREAT-NMD: Accelerating Treatments for Neuromuscular Diseases (registries) | 0 | o | 0 |
| UK10K RARE NEUROMUSCULAR – Genetic neuromuscular diseases variant database | О | С | 0 |
| X-ALD: X-linked adrenoleukodystrophy database | 0 | 0 | C |

*37. What is applicable for you in the following list about **Scientific Organization.**

| | Don't know it | Know it | Collaborate with it |
|---|---------------|---------|---------------------|
| AECOM – Asociación Española para el Estudio de los Errores Congénitos del Metabolismo | С | C | С |
| APS – Arbeitsgemeinschaft für Pädiatrische Stoffwechselstörungen (Working group for paediatric metabolic disturbances, Germany) | С | С | c |
| B4B – Brains for Brain | c | C | С |
| BIMDG – British Inherited Metabolic Disease Group | 0 | 0 | С |
| CLIMB – Children Living with Inherited Metabolic Diseases, UK | C | 0 | С |
| EBC – European Brain Council | 0 | 0 | c |
| ERNDIM Quality assurance in laboratory testing for IEM | C | 0 | О |
| ESGCT – European Society of Gene and Cell Therapy | 0 | 0 | 0 |
| EURORDIS – European Organisation for Rare Disorders | C | C | С |
| Fondazione Telethon | 0 | C | C |

| | Don't know it | Know it | Collaborate with it |
|--|---------------|---------|---------------------|
| IBBS – International Brain Barriers Society | 0 | c | 0 |
| IRDIRC | 0 | C | 0 |
| ISNS – International Society for Neonatal Screening | 0 | c | 0 |
| SEEIM – Sociedad Española de Errores Innatos del Metabolismo | 0 | C | 0 |
| SFEIM – Societé Française pour l'Étude des Erreurs Innées du Métabolisme | o | 0 | С |
| SIMD – Society for Inherited Metabolic Disorders | 0 | C | 0 |
| SISMME – Societa' Italiana per lo Studio delle Malattie Metaboliche Ereditairie | c | 0 | С |
| SPDM – Sociedade Portuguesa de Doenças Metabólicas | C | C | 0 |
| SSIEM – The Society for Study of Inborn Errors of Metabolism | С | С | C |