Supplementary File :1 PCSRRDI Participant Information Sheet

RAinDRoP

Rare Disease Research Partnership

Tell Us Please

What questions would you like to see answered by Rare Disease research?

We are asking people living with Rare Diseases and their families, clinicians, health and social care professionals to identify the top 10 research questions.

This will help us (RAinDRop) to guide Rare Disease research to focus on the most urgent needs of people living with Rare Diseases.

Should you require any assistance or if you have additional questions,
please Contact raredisease@ucd.ie

HRB Health Research Board

IPPOSI

Rare Diseases Ireland

HSE

medical research charities group
Unity is our Strength

UCD DUBLIN

What is this Survey About?

The purpose of the Survey is to identify shared challenges of Rare Diseases.

Who Has Developed This Survey?

A team of researchers has developed the Survey at UCD in collaboration with partners from the National Clinical Care Programmes, Rare Diseases Ireland and Rare Disease Taskforce.

Why am I asked to complete the Survey?

You are being asked to complete the survey as part of a consultation exercise. We value your experience and opinion, either as a patient, family member, carer, health professional, researcher or a member of the public.

How will the survey be carried out?

This survey questionnaire will be promoted widely via social media. Recruitment will commence in February 2019, and the report of the findings will be completed in June 2019. Sample size has not been determined for this survey, as the aim is to reach as many eligible clinicians, patients and carers to work together to identify and prioritise uncertainties that could be answered by rare disease research. Should you decide to participate, you may use the link to the at the end of this information leaflet which will direct you to the survey. Hard copies of the study are also Version 5. 26.02.19 will be available for participants wishing to complete by hand. Completion of the survey will be deemed as consent to participate.

What will happen to me if I agree to take part?

If you agree to participate, you will be asked to complete the survey once, either electronically online or using a hard copy. Completion of the survey should take no longer than 10 minutes and does not involve any interaction with the investigator. Further information relating to data protection can be found below.

What are the benefits and risks of taking part in this survey?

There are no individual benefits anticipated in the short term; however, for clinicians, patients and carers that were previously unaware of the RAinDRoP, this survey provides an opportunity to network and avail of the resources and support of this group. No risks have been identified in relation to participation in this survey.

The survey results will feed into a workshop discussion and prioritisation exercise by the Rare Disease Research (RAinDRoP) partnership which will seek to influence future research priorities in Rare Disease. The Health Research Board Ireland (HRB) will fund the workshop, which is organised by University College Dublin (UCD) in collaboration with National Clinical Care Programmes, Rare Diseases Ireland, Rare Disease Taskforce, The Irish Platform for Patient Organisations, Science and Industry (IPPOSI), and the Medical Research Charities Group (MRCG).

Is the survey confidential

All data received will be treated with full respect for confidentiality and individual data will not be discussed with any third parties. Completed survey forms will be stored securely and destroyed as soon as data has been entered into the 'Survey' database and the researchers are satisfied that there are no transcription errors. Once the data is entered in the 'Survey' database, it is anonymous and cannot be linked to the participant. Data entered into the 'Survey' database may be viewed by statisticians providing support in analysing and interpreting the data. As re-identification will not be possible, you will not be able to request that your information is withdrawn once it is entered in the 'Survey' database.

Data Protection

The purpose or reason for processing your personal data.
Personal identifiable data (e.g. name, email address or other contact details) will be used to initiate contact with you by RAinDRoP. A survey announcement and 'Consent for Contact' form has been developed for this purpose. This consent for contact form will be used for initial survey contact only and not saved for inclusion in the proposed RAinDRop database. The core survey dataset will not include personally identifiable data.

The legal basis under which we are processing your data.
The legal basis for processing your data within the context of this survey is consent. Consent for inclusion in the study is implied by your completion and submission of the survey form.

Who are the recipients of the data?
The Principle Investigators (PI) and Co-Investigators will have access to data provided by each participant in the survey. Survey responses will be exported to a survey database for analysis and interpretation. The survey database may be viewed by statisticians where necessary.

How long will the data be stored for?
Hard copies of the survey will be checked for completeness prior to manually entering into the survey database. A further quality check will be made, after which scanned copies will be made and the hard copies destroyed. SurveyMonkey® responses will be exported to the survey database. The scanned copies and SurveyMonkey® responses will be destroyed once all data accuracy has been verified. This is expected to be no later than July 2019. The survey database will be deleted when the researchers are satisfied that all data analysis has been performed and no further interrogation of the data is necessary – not later than December 2019.

Risks and/or implications that might arise as a result of the processing of your data
Hard copies of the survey will be scanned and then destroyed following input into the database. Electronic data will be located on a password protected project specific folder and stored on the organisation drive. No identifiable or sensitive data is being collected for this survey.

Withdrawal of consent.
There is no obligation to take part in the survey. However, if you do decide to complete the survey, the responses are anonymous and cannot be linked to participants, therefore any request for information to be withdrawn once it is submitted will not be possible.

Consent to Future Uses.
The data collected in the survey will be used for this project only and will not be retained for future use. The survey results will feed into a workshop discussion and prioritisation exercise of the Rare Disease Research (RAinDRoP) partnership which will seek to influence future research priorities in Rare Disease.

How long will it take to complete the Survey?
It will take approximately 10 minutes to complete all questions.
Should you require any assistance or if you have additional questions, please contact raredisease@ucd.ie

Supplementary File :2 Phase I Survey

Rare Disease Research Priority Setting (RAinDRoP) survey

What is this Survey About?

The Survey contains questions about the future direction of research in rare diseases.

Who Has Developed This Survey?

A team of researchers has developed the Survey at UCD in collaboration with partners from the National Clinical Care Programmes, Rare Disease Ireland and Rare Disease Taskforce.

Why am I asked to complete the Survey?

You are being asked to complete the survey as part of a consultation exercise. We value your experience and opinion, either as a health professional, researcher of a member of the public. How will the Survey Information Be Used? The survey results will feed into a workshop discussion and prioritisation exercise of the Rare Disease Research partnership (RAinDRop) which will seek to influence future research priorities in Rare Disease. The Health Research Board Ireland (HRB) funds the Workshop, which is organised by the University College Dublin (UCD) in collaboration with National Clinical Care Programmes, Rare Disease Ireland, Rare Disease Taskforce, The Irish Platform for Patient Organisations, Science and Industry (IPPOSI), and the Medical Research Charities Group (MRCG).

How long will it take to complete the Survey?

It will take approximately 5 minutes to complete all questions.

Should you require any assistance or if you have additional questions, please contact rarediseasesymposium@ucd.ie

Many thanks for your participation RAinDRoP Team

By completing this survey, I am confirming that I understand the purpose of the study and give my consent for my responses to be used anonymously for research.

Yes ◯

No ◯

2. By participating in this survey you are agreeing to allow us to anonymously publish the report as part of the Rare Disease Research Priority Partnership (RAinDRoP) Report.

Yes ◯

No ◯

Rare Disease Research Priority Setting (RAinDRoP) survey

3. Which of the following categories best describes you?

Patient with a rare disease ◯
Family caregiver ◯
Clinician Allied Health Care Professionals ◯
Academics ◯
Researcher ◯
Scientists ◯
Patient Organisations ◯
Policy Makers ◯
Funding Body ◯
Other Specify ◯

Section 4: Survey

To write your unanswered questions or areas important

What question(s) about dealing with the **diagnosis** of Rare Disease would you like to see answered by research?
1.
2.
3.
What question(s) about managing day-to-day life with Rare Disease would you like to see answered by research?
1.
2.
3.
What question(s) about the treatment of Rare Disease would you like to see answered by research?
1.
2.
3.

What question(s) about the **self-management/overall management** of Rare Disease would you like to see answered by research?
1.
2.
3.
What question(s) about the integrated care/holistic care of Rare Disease would you like to see answered by research?
1.
2.
3.
What question(s) about the palliative care service for advanced Rare Disease would you like to see answered by research?
1.
2.
3.

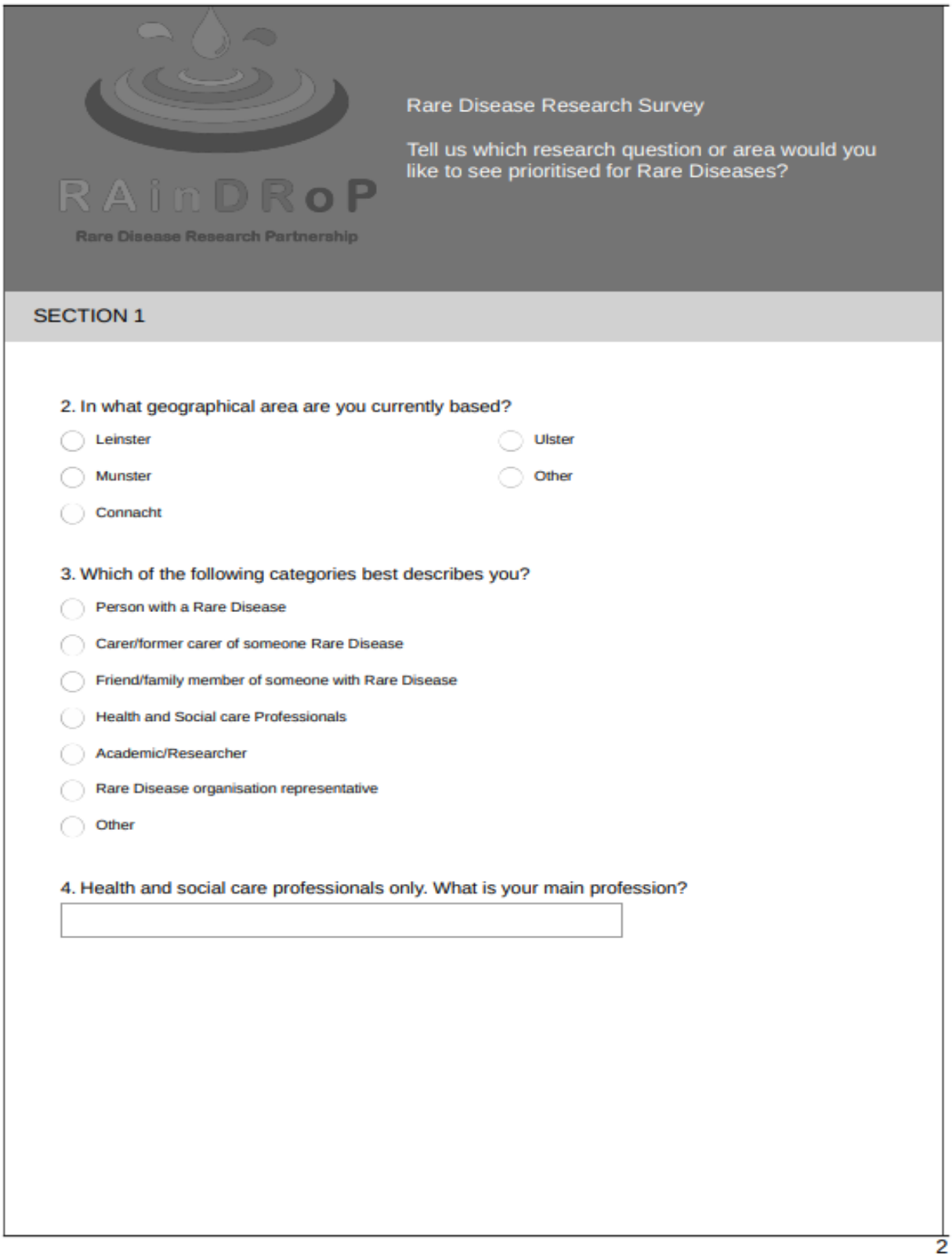
Do you have any other questions that you feel are important but do not fall into the areas above?

Supplementary File 3: RPW Workshop agenda

0830-0900	Registration & Networking	
0900-0905	Welcome and Chair: Prof Thilo Kroll, Professor of Health Systems Management, UCD School of Nursing, Midwifery and Health Systems	
0905-0915	Opening Address	Ms Avril Daly, CEO, Patient-led Global NGO, Retina International, Vice-President of EURORDIS
0915-0930	Overview of the workshop	Dr Suja Somanadhan, Assistant Professor in Children's Nursing, UCD School of Nursing Midwifery and Health Systems Dr Emma Dorris, Molecular Biologist, UCD Centre for Arthritis Research
0930-0950	Theme 1 Route to Diagnosis	Dr Sally Ann Lynch, Consultant Clinical Genetics, Our Lady's Children's Hospital Crumlin. Dr Sean Ennis, Director of UCD Academic Centre of Rare Diseases
0950-1020	Theme 2 Living with and caring for Rare Diseases	Mr Dermot Devlin, Patient's perspective Ms. Anne Lawlor, Mother's perspective
1020-1040	Theme 3 Integrated care and Palliative Care	Associate Prof Maria Brenner, Associate Professor in Children's Nursing, School of Nursing and Midwifery, Trinity College Dublin Dr Julie Ling, Chief Executive Officer, European Association for Palliative Care
1040-1115	Coffee Break	

1115-1315	RAinDRoP CAFÉ 1 (40 Minutes) Route to Diagnosis Lead: Dr Avril Kennan	RAinDRoP CAFÉ 2 (40 Minutes) Living with rare diseases Lead: Dr Aoife Brinkley	RAinDRoP CAFÉ 3 (40 Minutes) Integrated care Lead: Prof Thilo Kroll
	Co-lead: Dr Emma Dorris	Co-lead: Dr Suja Somanadhan	Co-lead: Dr Emma Nicholson
1315-1400	Lunch Break & Networking		
	Chair: Ms Vicky McGrath, CEO, Rare Diseases Ireland.		
1400-1430	Keynote Speaker Prof Alison Metcalfe: Pro Vice-Chancellor and Dean for the Faculty of Health and Wellbeing, Sheffield Hallam University.		
1430-1445	Prof Eileen Treacy, Clinical Lead, National Clinical Programme for Rare Diseases, Director, National Rare Diseases Office.		
1445-1500	Ms Eilish Hardiman, Chief Executive, Children's Health Ireland.		
1500-1515	Dr Anne Cody, Head of Pre-Award, Health Research Board (HRB) Ireland		
1515-1600	Rare Disease Research Top 10 Priority Settings report by workshop leads Dr Avril Kennan, Dr Aoife Brinkley, Prof Thilo Kroll		
1600-1645	Panel Conversation : The Future of Rare Disease Research in Ireland Chair: Dr Derick Mitchell, Chief Executive, IPPOSI Panel Members: Dr Anne Cody, Prof Eileen Treacy, Prof Aine Carroll, Ms Julie Power, Mr Dermot Devlin		
1645-1700	Key Learning and next steps : Dr Avril Kennan, CEO, Medical Research Charities Group		

Supplementary File 4:
Follow-up Public Consultation and Prioritisation Survey (FWPCPS)



5. Which research question or area would you like to see prioritised for Rare Diseases? Use the drop down to rank in order of your preference.

- Support at the time of a Rare Disease diagnosis
- Data sharing and integration of services for Rare Diseases
- Co-design of (research, services, information, dissemination) for Rare Diseases
- Psycho-social impact of a Rare Disease diagnosis
- Role of infrastructure in diagnosing a Rare Disease (e.g Registry/ERN Centres of excellence)
- Diagnostic tests for Rare Diseases(e.g.Use of genetics, Stratified medicine/ molecular targeted therapies, gene therapy etc.)
- Best way to deliver a Rare Disease diagnosis (e.g. mail. phone, in person (consultant, GP, Nurse, other)
- Evidence-based models of integrated care for Rare Diseases
- Community based services and treatment for Rare Diseases
- Psychosocial impact of living with Rare Diseases (e.g. physical functioning, psychological, social and mental health and quality of life etc.)
- Economical Impact of living with Rare Diseases (e.g. healthcare costs, transportation costs, education costs, loss of earnings, etc.)
- Transition Services for Rare Diseases (e.g barriers and enablers for transitioning from paediatric to adults' services)
- Family Experience of living with Rare Diereses (e.g Parents, mother, father, siblings and grandparents experience of living with and caring)
- Education and Training (e.g. health and social care professionals, school, GP and patient information and understanding of their illness and management)
- Patient Voice (eg: How to include the child's voice in relation to their care)

Supplementary file 5: RAinDRoP Project Timelines

Timelines	
May 2018	Networks established at the UCD Rare Disease symposium on the 03rd of May 2018 The inaugural Rare Disease Symposium took place in University College Dublin – a first step in forming a collaborative partnership between researchers, people living with rare disease, researchers and health care practitioners
6th& & 11th June 2018	Group 1 &2 steering members: Engaged partners in consultations, pre-planning to identify a process for identifying the top priorities of rare disease research
30th Oct 2018, 21st November 21st January 2019	Co-designed developed a public online survey with the steering committee
Feb-March 2019	Launched, Processed and collated survey responses
4th March 2019	High-quality engagement and screening to cluster results into three themes (emailed all the clinicians (who couldn't be there at the meeting) to hear their final opinion and feedback
4th of April, 2019	Priority setting workshop to prioritise rare disease research from a selection of 28 areas (presented under three themes)
21st May 2019	A final public survey is developed and launched to validate 15 priorities with a wider audience