

Identifying Rare Disease Research Priorities: Rare Disease Research Partnership (RAInDRoP)

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Background:

The European Commission on Public Health (2011) defines a rare disease as a life-threatening or chronically debilitating disease, mostly inherited. In EU countries, any disease affecting fewer than five people in 10,000 is considered rare, but most patients suffer from even rarer diseases affecting 1 person in 100,000 or more (EU Commission, 2019). Although each disease is rare but collectively these are common and affect many individuals. Rare Diseases translate into approximately 5,000-8,000 distinct rare diseases affect 6-8% of the EU population i.e. between 27 and 36 million (EU Commission, 2019).

The Rare Disease Research Partnership (RAInDRoP) was formally established in June 2018 to bring together the rich variety of diverse voices in the rare disease community in Ireland and form a research partnership. The establishment of formal networks of rare disease researchers including patient and public involvement (PPI) is aligned with recommendation by the Ireland's National Plan for Rare Diseases (HSE, 2014). Rare Disease Research Partnership (RAInDRoP) enabled clinicians, persons living with rare diseases and their family/carers, policy makers, researchers to work together to identify and prioritise uncertainties that could be answered by research. The RAINDRoP is a truly collaborative effort in which everybody has a voice, and it refocuses the research on the patients rather than the disease.

The objectives of RAINDRoP:

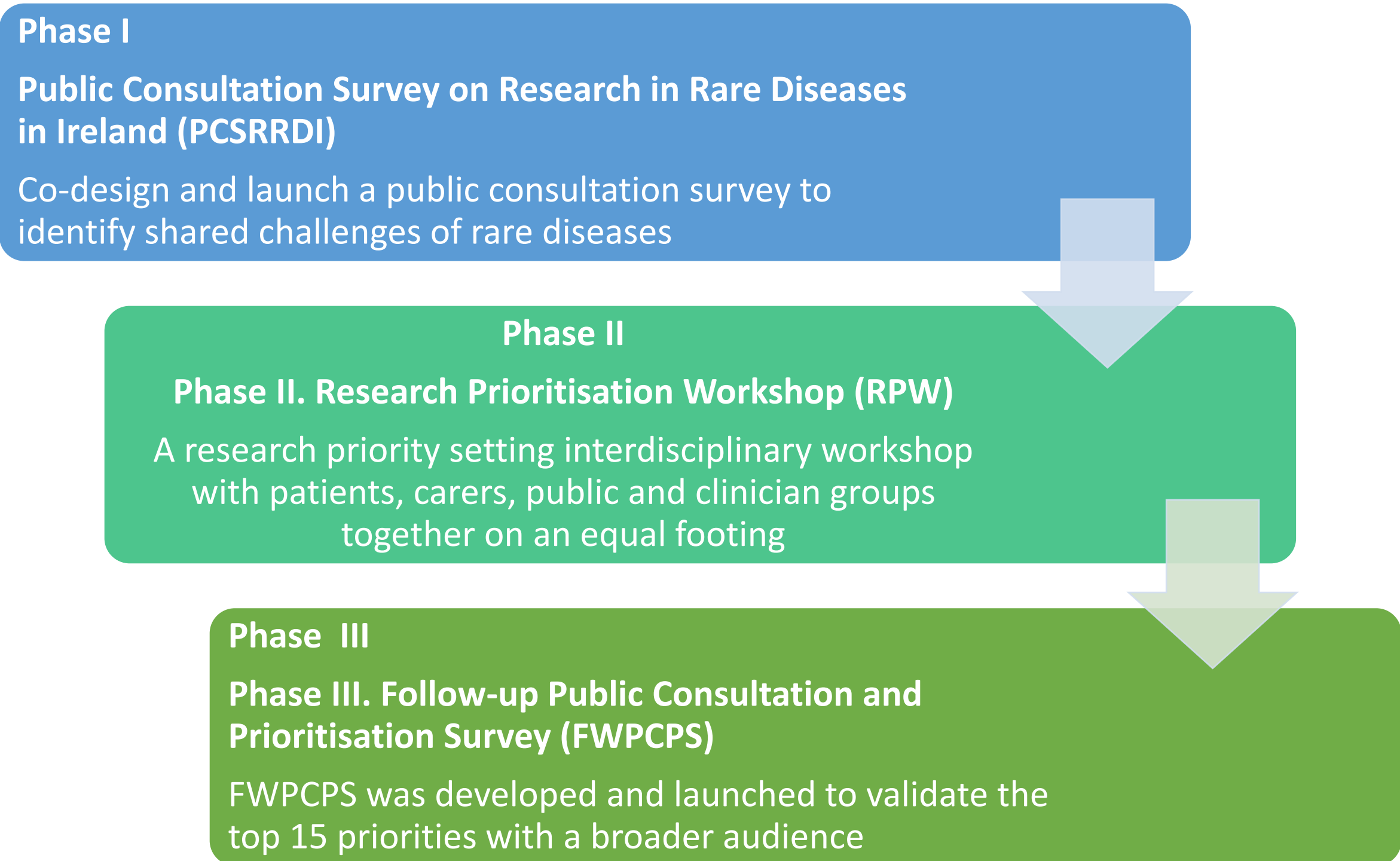
- To launch a platform for the Rare Disease Research network in Ireland
- To support the integration of rare disease research within relevant forthcoming
- Government research policy and legislation as per the National Plan for Rare Diseases (DOH, 2014)
- To create a consensus statement in relation to rare disease research priorities
- To create a strategic approach to rare disease research by developing a rare disease research strategy.
- To develop an action plan to engage with research commissioning and funding bodies

Setting: Republic of Ireland

Participants: Anyone with experience of care for patients with rare diseases including patients, carers, clinician's, allied health and social care professionals, nurses and midwives, academics, researchers NGO workers, government agencies and policy makers.

Method:

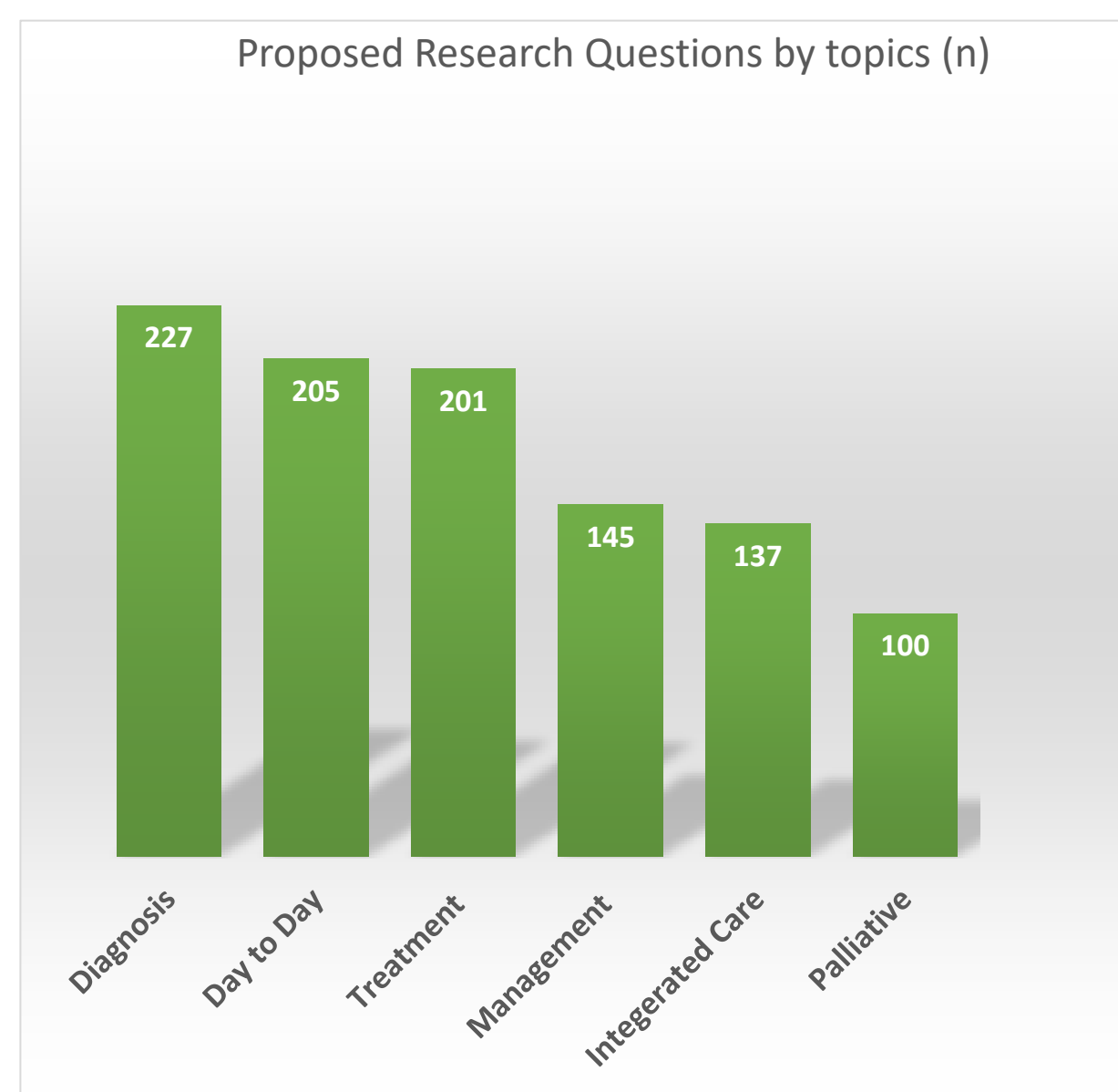
Participatory research method was utilised in this projects with a focus researching "on" to researching "with" participants via multiphase approaches.



Phase I: Public Consultation Survey on Research in Rare Diseases in Ireland (PCSRDI)

Topics identified by Expert Group

1. Diagnosis
2. Day-to-day life
3. Treatment
4. Self-management / overall management
5. Integrated / holistic care
6. Palliative care



High Quality Engagement and Screening

Following the survey, we conducted an interdisciplinary high -quality engagement meeting with rare disease research interest groups to categorise the results. We used a scoring system whereby anything above 50% was considered for shortlisting. From this analysis three themes emerged:

- I. Route to Diagnosis
- II. Living with rare disease
- III. Integrated care and palliative care

Phase II: A research priority setting interdisciplinary workshop (RPW)

The RAINDRoP workshop took place over one day and was attended by 62 (n=62) participants out of which 44(n=44) attended each café. Each group contained a cross-section of health care professionals, researchers and people living with a rare disease. Each participant took part in three cafés based on the three themes that emerged from the survey to discuss each topic. Participants then individually marked each topic as being high, medium or low importance and were asked to invest funding (€) in the research topics using 'fake' money they were provided with.



Café 1 Route to Diagnosis	IMPORTANCE		INVESTMENT	
	High importance assigned to each subtheme	Euro	High importance assigned to each subtheme	Euro
Facilitators: Dr Avril Keenan & Dr Emma Dorris				
Subthemes (n=10)	n	%	n	%
Support at the time of diagnosis	19	16	170	6
Role of infrastructure in diagnosis	15	13	525	18
Psychosocial impact of a diagnosis	14	12	570	20
Best way to deliver diagnosis	13	11	500	17
Diagnostic tests	12	10	150	5
Education and training	11	9	305	11
Timeframes to diagnosis	11	9	160	6
Understanding incidence and prevalence	9	8	295	10
Role of diagnosis in understanding impact of disease	9	8	190	7
Screening and risk	7	6	220	8
TOTAL	120	100	2865	100

Café 2 Living with RD	IMPORTANCE		INVESTMENT	
	High importance assigned to each subtheme	Euro	High importance assigned to each subtheme	Euro
Facilitators: Dr Aoife Brinkley & Dr Suja Somanadhan				
Subthemes (n=9)	n	%	n	%
Psychosocial impact	19	18	590	22
Transition services	16	15	425	16
Economic impact	16	15	245	9
Family experience	13	12	265	10
Patient Voice	12	11	245	9
Psychosocial services	11	10	400	15
Education and training	9	8	190	7
Info and awareness	6	6	210	8
Info for families and patients	5	5	105	4
TOTAL	107	100	2675	100

Café 3 Integrated and Palliative Care	IMPORTANCE		INVESTMENT	
	High importance assigned to each subtheme	Euro	High importance assigned to each subtheme	Euro
Facilitators: Prof Thilo Kroll & Dr Emma Nicholson				
Subthemes (n=9)	n	%	n	%
Data sharing and integration	24	21	945	44
Co-designing services	24	21	365	17
Community services, treatment, multimorbidity	15	13	230	11
Evidence-based models of integrated care	15	13	180	8
Palliative care at-home	13	11	20	1
Equitable and timely access, provision of palliative care	8	7	100	5
Palliative care education and training of HCPs	7	6	110	5
Prep for bereavement, acceptability of pall role	7	6	110	5
Advanced care planning	1	1	90	4
TOTAL	114	100	2150	100

Top 10 Priorities	Importance		Investment	
	n	%	n	%
1. Data sharing and integration (Café: Integrated and Palliative Care)	24	21	945	44
2. Co-designing services (Café: Integrated and Palliative Care)	24	21	365	17
3. Psychosocial impact (Café: Living with Rare Disease)	19	18	590	22
4. Support at the time of diagnosis (Café: Route to Diagnosis)	19	16	170	6
5. Transition services (Café: Living with Rare Disease)	16	15	425	16
6. Economic impact (Café: Living with Rare Disease)	16	15	245	9
7. Community based services, treatment, multi-morbidity (Café: Integrated and Palliative Care)	15	13	230	11
8. Evidence-based models of integrated care (Café: Integrated and Palliative Care)	15	13	180	8
9. Role of infrastructure in diagnosis (Café: Route to Diagnosis)	15	13	525	18
10. Psychosocial impact of a diagnosis (Café: Route to Diagnosis)	14	12	570	20

Phase III: Follow-up Public Consultation and Prioritisation Survey (FWPCPS)

A final consultation survey was successfully completed with (n=47) to prioritise top 15 research themes aimed at improving the health and wellbeing of people living with Rare Diseases. The response group was represented a cross-section of health care professionals (n=7), researchers (n=8) and people living with a rare disease (n=19) Carer/former carer of someone Rare Disease (n=6) Friend/family member of someone with Rare Disease (n=14) NGO workers (n=2).

Top 15 from survey	(n) respondents	%
Support at the time of a Rare Disease diagnosis	10	23%
Diagnostic tests for Rare Diseases(e.g. Use of genetics, Stratified medicine/ molecular targeted therapies, gene therapy etc.)	6	14%
Education and Training (e.g. health and social care professionals, school, GP and patient information and understanding of their illness and management)	6	14%
Patient Voice (eg: How to include the child's voice in relation to their care)	4	9%
Data sharing and integration of services for Rare Diseases	4	9%
Economical impact of living with Rare Diseases (e.g. healthcare costs, transportation costs, education costs, loss of earnings, etc.)	3	7%
Psychosocial impact of living with Rare Diseases (e.g. physical functioning, psychological, social and mental health and quality of life etc.)	2	5%
Community based services and treatment for Rare Diseases	2	5%
Evidence-based models of integrated care for Rare Diseases	2	5%
Family experience of living with Rare Diseases (e.g Parents, mother, father, siblings and grandparents experience of living and caring and life-course transitions)	1	2%
Transition services for Rare Diseases (e.g barriers and enablers for transitioning from paediatric to adults' services)	1	2%
Co-design of (research, services, information, dissemination) for Rare Diseases	1	2%
Psycho-social impact of a Rare Disease diagnosis	1	2%
Role of infrastructure in diagnosing a Rare Disease (e.g Registry/ERN Centres of excellence)	1	2%
Best way to deliver a Rare Disease diagnosis (e.g. mail, phone, in person (consultant, GP, Nurse, other)	0	0%
TOTAL	44	100%

Conclusion:

The RAINDRoP programme provided a high-quality engagement with patients, carers, clinician's, allied health and social care professional's, nurses and midwives, academics, researchers NGO workers, government agencies and policymakers together on an equal footing. This engagement was a two-way process to generate mutual benefit between the public and researchers and ultimately enhancing the quality or impact of research from the health consumers perspective. This proposed activity was in the line up with HRB specific objective to create opportunities for strategic national and international collaboration in patient-focused rare disease research partnership at all career stages. Overall the results of the RAINDRoP Research prioritisation is highly significant as they set research priorities for key national and European priority research areas including Rare Disease, Health Services Research and Interdisciplinary Research.