

## **Underlying Data File:2**

### **Initial Grouping of Themes into Questions from PCSRRDI**

#### **Café 1: Diagnosis**

##### **1. The best way to deliver a diagnosis**

- What is the best way to communicate a diagnosis?
- What is the role of genetic counselling in relation to diagnosis?

##### **2. The psychosocial impact of a diagnosis**

- What is the impact of a diagnosis on a patient?
- What is the impact of a diagnosis on the patient's family?

##### **3. Support at the time of diagnosis**

- What information about the disease should be provided at diagnosis?
- What resources should be provided to carers at the time of a diagnosis?
- What support should be provided to the parents in the case of a diagnosis before birth?

##### **4. Diagnostic tests**

- How to we make diagnostic tests more specific?
- What is the role of genetic testing in diagnosis?
- How can diagnosis help us identify sub-groups of patients that might need a different approach or respond differently to treatment?
- What is the role of a multi-disciplinary team in diagnosis?

##### **5. The role of diagnosis in understanding the impact of disease**

- How does a diagnosis help with understanding the likely path of a rare disease?
- How can diagnosis help us measure the impact of the disease, in terms of cost and quality of life?
- Can diagnosis provide information on the risk of additional conditions occurring, in addition to the initial condition?

##### **6. Timeframes to diagnosis**

- What are the average timeframes from diagnostic tests to results
- How do we minimise delays in diagnosis?

##### **7. The role of infrastructure in diagnosis**

- How can European Reference Networks help with diagnosis?
- How can centres of excellence help with diagnosis?
- How can patient registries help with diagnosis?

##### **8. Screening and risk**

- What advice on risk should be provided to family members of someone who has received a diagnosis?
- What screening options should be provided to family members of someone who has received a diagnosis?
- What can a diagnosis tell us about causes or risk factors for rare diseases?
- What role does diagnosis have in disease prevention?

#### **9. Understanding incidence and prevalence**

- How can diagnosis help us understand more about the number of people living with a rare disease and the number of new cases every year?

#### **10. Education and training**

- What training should be provided to healthcare professionals in relation to diagnosis?
- What training should be provided to carers in relation to diagnosis?
- What training should be provided to those in community support roles following at diagnosis (e.g. schools, HR staff)?

## **Café 2: Living with Rare Diseases**

### **1: Psychosocial Impact of Living with a Rare Disease**

- How does living with a rare disease affect the quality of life of patients and their families?
- How does living with a rare disease impact on education, housing, employment and other life domains?
- What is the role of positive and intimate friendships in the lives of people living with rare diseases?
- How does rare disease impact school life?

### **2: Psychosocial Services**

- What would effective psychosocial services for people and families living with a rare disease look like?
- How does input from AHP/MDT impact physical functioning, psychological wellbeing and quality of life

### **3: Education and Training for Health Care Professionals**

- What education and training programmes do we need to develop for health professionals?
- What are the benefits of medical guidelines or standards?

### **4: Family Experience**

- How do we capture family experience data?
- What are the mitigating and exacerbating factors of caregiver stress?

#### **5: Patient voice**

- How can we improve patient involvement in research? i.e listening to the patient in rare disease research (6/8)
- How to include the child's voice in relation to their care

#### **6: Information and awareness of RD**

- What information do patients need to be able to manage day to day medical care?
- What information do General Practitioner need to be able to provide day to day medical care (6/9)
- What is the awareness of managing rare diseases in General Practice?
- How does patient information and understanding of their illness impact their daily lives?

#### **7: Transition services**

- How do we support transitioning to adult services in light of limited awareness of rare disease in some adult services?
- What are the barriers and enablers for transitioning from paediatric to adults' services?

#### **8: Economic impact**

- What is the extent of economic burden on families/carers of people with a rare disease?

#### **9: Information for families and patients**

- What sources of information do families and carers use in relation to rare disease?

### **Café 3: Integrated and Palliative Care**

#### **1: Community-Based Services and Treatments and Multi-morbidity**

- How do we develop effective community-based services and treatments?
- How can we develop effective treatments and services for individuals who have to manage more than one condition?
- What is the best approach or model to support self-management of rare-diseases in the community?

#### **2: Co-Designing Services**

- How can we meaningfully involve patients and families in the Co-Design of Care Systems and in the Delivery of Care?
- What is the best approach for designing and delivering services to support management of Rare Diseases?

### **3: Data sharing and integration**

- What would an effective and integrated patient data management system look like?
- What could we do to link data across different data sources?
- How can we make progress towards digitalising medical records?

### **5: Evidence-based Models of Integrated Care**

- What is the best international evidence from other countries for integrated care models and how can this evidence be used to inform services in the Irish health system?
- How can we develop effective, integrated care networks?
- What is the relative contribution of the publicly funded sector and the charity sector?
- Can improved anticipatory care reduce the need to access hospital services? Can primary care play a role in this?
- Are peer support meetings helpful for patients, families, carers?

### **6: Palliative Care At-Home**

- How can palliative care be safely and appropriately delivered at home outside a hospital or hospice setting?

### **7: Equitable and timely Access to and Provision of Palliative Care**

- Do patients with rare diseases have the same access and do they receive the same level and quality of palliative care than those with a cancer diagnosis?

### **8: Advanced Care Planning**

- How can Advanced Care Planning be introduced?

### **9: Palliative Care Education and Training of Health Care Professionals**

- What specific training can be developed and provided for health professionals to provide support to patients with rare diseases and their families at the end of life?

### **10: Preparing for Bereavement and Acceptability of Palliative Role in End-of-Life Management**

- How can professionals and families best be prepared for eventual bereavement?
- What needs to be done to enhance the acceptability of the palliative role among health professionals?

## Underlying Data File:3

### All café priority-based data on high importance and investment

	IMPORTANCE		INVESTMENT	
<b>Café 1 Route to Diagnosis</b> Facilitators: Dr Avril Keenan & Dr Emma Dorris	High importance assigned to each theme		Euro	
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 a 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 the impact of disease	9	8	190	7
Screening and risk	7	6	220	8
<b>TOTAL</b>	<b>120</b>	<b>100</b>	<b>2865</b>	<b>100</b>
	IMPORTANCE		INVESTMENT	
<b>Café 2 Living with RD</b> Facilitators: Dr Aoife Brinkley & Dr Suja Somanadhan	High importance assigned to each theme		Euro	
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
<b>TOTAL</b>	<b>107</b>	<b>100</b>	<b>2675</b>	<b>100</b>
	IMPORTANCE		INVESTMENT	
<b>Café 3 Integrated and Palliative Care</b> Facilitators: Prof Thilo Kroll and Dr Emma Nicholson	High importance assigned to each theme		Euro	
Subthemes (n=9)	n	%	n	%
Data sharing and integration	24	21	945	44
Co-designing services	24	21	365	17
Community based services, treatment, multi-morbidity	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 the pall role in end-of-life management	7	6	110	5
Advanced care planning	1	1	90	4
<b>TOTAL</b>	<b>114</b>	100	<b>2150</b>	100

## Underlying Data File: 4

### Priority ratings in terms of 'importance' by café group in phase II

HCP, healthcare professional; PwRD, People living with rare disease; Others, academics, researchers, academic policymakers and research managers.

<b>Café 1. Route to Diagnosis</b>	<b>High importance</b>			
<b>Subthemes (n=10)</b>	<b>HCPs</b>	<b>Others</b>	<b>PwRD</b>	<b>Totals</b>
Best way to deliver a diagnosis	3	0	10	13
Psychosocial impact of a diagnosis	3	5	6	14
Support at the time of diagnosis	7	3	9	19
Diagnostic tests	6	6	0	12
Role of diagnosis in understanding the impact of disease	5	2	2	9
Timeframes to diagnosis	6	0	5	11
Role of infrastructure in diagnosis	4	5	6	15
Screening and risk	5	2	0	7
Understanding incidence and prevalence	3	2	4	9
Education and training	3	4	4	11
<b>TOTAL</b>	<b>45</b>	<b>29</b>	<b>46</b>	<b>120</b>
<b>Café 2 Living with Rare Disease</b>	<b>High importance</b>			
<b>Subthemes (n=9)</b>	<b>HCPs</b>	<b>Others</b>	<b>PwRD</b>	<b>Totals</b>
Psychosocial impact	6	5	8	19
Transition services	8	4	4	16
Economic impact	7	4	5	16
Family experience	4	5	4	13
Patient Voice	1	4	7	12
Psychosocial services	6	0	5	11
Education and training	3	2	4	9
Information and awareness	0	1	5	6
Information for families and patients	2	1	2	5
<b>TOTAL</b>	<b>37</b>	<b>26</b>	<b>44</b>	<b>107</b>

<b>Café 3. Integrated and Palliative Care</b>	<b>High importance</b>			
<b>Subthemes (n=9)</b>	<b>HCPs</b>	<b>Others</b>	<b>PwRD</b>	<b>Totals</b>
Co-designing services	7	7	10	24
Data sharing and integration	7	7	10	24
Community-based services, treatment, multi-morbidity	4	4	7	15
Palliative Care at-home	8	2	5	15
Evidence-based models of integrated care	2	4	7	13
Equitable and timely access, provision of palliative care	3	3	2	8
Palliative Care education and training of HCPs	2	3	2	7
Prep for bereavement, acceptability of the palliative role in end-of-life Management	1	2	4	7
Advanced care planning	0	0	1	1
<b>TOTAL</b>	<b>34</b>	<b>32</b>	<b>48</b>	<b>114</b>



## Underlying Data file:5

### Priorities ranked in the first position by respondents in FWPCPS

Q: Which research question or area would you like to see prioritised for Rare Diseases? <i>Use the drop-down to rank in order of your preference</i>		
Priorities ranked in the first position by respondents	(n) respondents	%
1. <b>Support at the time of a Rare Disease diagnosis</b>	10	23%
2. <b>Diagnostic tests for Rare Diseases</b> (e.g. use of genetics, stratified medicine/ molecular targeted therapies, gene therapy etc.)	6	14%
3. <b>Education and Training</b> (e.g. health and social care professionals, school, GP and patient information and understanding of their illness and management)	6	14%
4. <b>Patient Voice</b> (e.g. How to include the child's voice about their care )	4	9%
5. <b>Data sharing and integration of services for Rare Diseases</b>	4	9%
6. <b>The economic impact of living with Rare Diseases</b> (e.g. healthcare costs, transportation costs, education costs, loss of earnings etc)	3	7%
7. <b>Psychosocial impact of living with Rare Diseases</b> (e.g. physical functioning, psychological, social and mental health and quality of life etc.)	2	5%
8. <b>Community-based services and treatment for Rare Diseases</b>	2	5%
9. <b>Evidence-based models of integrated care for Rare Diseases</b>	2	5%
10. <b>Family experience of living with Rare Diseases</b> (e.g. Parents, mother, father, siblings and grandparents experience of living and caring and life-course transitions )	1	2%
11. <b>Transition services for Rare Diseases</b> (e.g. barriers and enablers for transitioning from paediatric to adults' services)	1	2%
12. <b>Co-design</b> of (research, services, information, dissemination) for Rare Diseases	1	2%
13. <b>Psychosocial impact</b> of a Rare Disease <b>diagnosis</b>	1	2%
14. <b>Role of infrastructure in diagnosing</b> a Rare Disease (e.g. Registry/ERN Centres of excellence)	1	2%
15. <b>Best way to deliver</b> a Rare Disease diagnosis (e.g. mail. phone, in person (consultant, GP, Nurse, other)	0	0%
<b>TOTAL</b>	<b>44</b>	<b>100%</b>