The Polka Delphi Study

‘Policy recommendations for Rare Disease Centres of Expertise’

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The Polka Project

Main Goals

Selection of "hot topics" in the field of Rare Diseases

Deliberative sessions for the empowerment of patients and their representatives

Patients' opinion better considered in national plans for Rare Diseases, in Centres of Expertise and Reference Networks



The Polka Project

Consensus on Preferred Policies for Rare Diseases

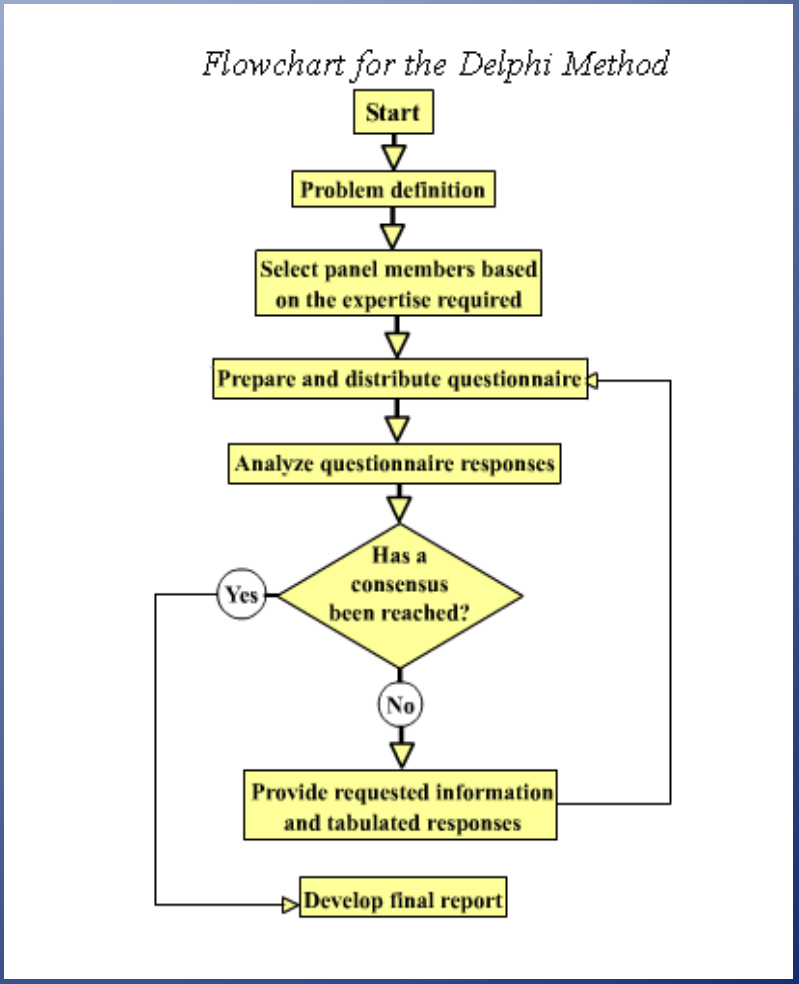
- Gather opinions on:
  - Stem cell research
  - Cost of orphan drugs
  - Newborn screening
  - Diagnosis, information and genetic counselling
  - Cross-border care
  - Pre-implantation Genetic Diagnosis
  - Centres of expertise for rare diseases
- Communicate opinions to policy makers (National and EU)



The Polka Delphi Study

Methodology

- A structured communication process
- Useful when there is little knowledge or uncertainty surrounding a complex area being investigated
- Relies on a panel of experts for opinions



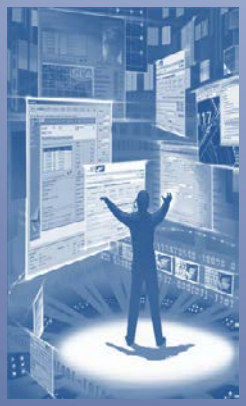
The Polka Delphi Process

1. Selection of participants: Patients and Healthcare professionals

8 HCPs and 12 patients/ patient representatives and carers)

Centres
UK=7, DK= 1, FR=??

2. First round: Questionnaires to assess status based on EurordisCare3 survey [13 Policy Areas]



Total responses (134)
HCP: UK=45,DK=5
Patients: UK=71, DK=13

Survey analysis

3. Second round: Face to Face meetings, feedback & consensus on issues

Total participants (67)
HCP: UK=30,DK=5
Patients: UK=22, DK=10

Analysis of results from Round 1 & 2

4. Final report: Policy Recommendations

5. Third round: Individuals' comments on the report (Total=100)

6. Recommendations to EC and MS

Participating Centres

- England

Centre of Expertise (CoE)	Diseases
Birmingham Children's Hospital NHS Trust (Birmingham)	Epidermolysis bullosa, Lysosomal Storage Disorders
St John's Institute of Dermatology, Guy's and St Thomas' NHS Trust (London)	Epidermolysis bullosa
Department of Genetic Medicine, Central Manchester University Hospitals (Manchester)	Neurofibromatosis type one and two
Birmingham Children's Hospital NHS Trust & Torbay Hospital, South Devon Healthcare NHS Foundation Trust (Birmingham & Torbay)	Alstrom syndrome adults and transition adolescents
Guys and St Thomas NHS Foundation trust (London)	Xeroderma pigmentosum
Mitochondrial Research Group, Newcastle University (Newcastle)	Mitochondrial Diseases
Birmingham Children's Hospital NHS Trust (Birmingham)	Developmental/Genetic disorders



Participating Centres

- Denmark

Centre of Expertise (CoE)	Diseases
Centre for rare disease (Center for sjældne sygdomme), Aarhus University Hospital (Aarhus)	Rare, often hereditary diseases with onset in childhood and symptoms from several organ systems Neurofibromatosis, Marfan syndrome , Ehlers Danlos Syndrome, Prader Willi Syndrome, Spielmeyer-Vogt syndrome, skeletal dysplasia, von Hippel-Lindau, Klippel-Trenaunay, Angelman, tuberous sclerosis, 22q11ds, Rubinstein-Taybi, Osteogenesis Imperfecta craniofacial disorders, tuberous sclerosis, Angelman syndrome, Moebius syndrome Moya-moya, Cri-du-chat, Williams syndrome, Beckwith-Wiedemann syndrome etc.



Policy areas explored

1. Patients and care environment at the centre
2. Access to centres
3. Coordination and cooperation within and outside the centre
4. Transition of care from childhood to adulthood
5. Patient autonomy
6. Patient representatives and support groups at CoEs
7. Provision of information to patients
8. Awareness and training amongst healthcare and non-healthcare professionals
9. Patient orientated and multidisciplinary care at the centre
10. Social care
11. Facilities for diagnosis of rare diseases
12. Performance management of the centre
13. Research at the centre



Policy Recommendations

Environment at CoEs

- ***The essential functions of a CoE should include provision of diagnosis, treatment and care in rare diseases it specialises in.***
- ***CoEs should be set up as separate units with dedicated staff providing multidisciplinary care for patients.***
- ***They should not be "over-medicalised" and should be designed such that they takes into account the needs of patients.***



Policy Recommendations

Access to CoEs

- ***Outreach clinics headed by CoEs should be established to minimise travel for patients and ensure continuity of care.***
- ***Cross-border agreements between European member states should be set-up to allow European patients with no access to care to allow treatment at other European CoE where can is available.***



Policy Recommendations

Coordination and cooperation within and outside CoEs

- ***Systems need to be established to ensure efficient exchange of patient information in the healthcare system (such as between GPs and specialists treating patients within & outside the CoE) to ensure early and accurate diagnosis and provision of efficient care.***
- ***Systems need to be set up in order to ensure patients are identified and moved appropriately through emergency services, such as immediate availability of important information about their condition and access to experts at CoEs during an emergency.***

Policy Recommendations

Transition of care from childhood to adulthood

The transition process should:

- ***Should ensure smooth transition of patients from paediatric care to adult care and patient records.***
- ***Consider any future needs of patients that may arise due to the condition.***
- ***Be overseen by a transition coordinator.***



Policy Recommendations

Patient representatives and support groups at CoEs

- ***Every CoE should be linked to a patient-led organisation for the condition(s) the CoE specialises in.***



Policy Recommendations

Provision of information to patients

- ***Patients and their families should be regularly provided with information about their condition using appropriate channels and formats adapted for the target audience taking into account factors such as age, ethnicity, education level etc.***

Policy Recommendations

Raising awareness and training of rare disease

- *Training for HCPs such as GPs and specialists, who are likely to come across rare disease patients.*
- *Training should be made mandatory (or offered as CPD or CME) to help ensure early detection, diagnosis and continuous appropriate management of rare diseases.*
- *HCPs (doctors, nurses and allied HCPs) should receive diversity training.*
- *Non-HCPs who come in contact with rare disease patients (such as school teachers, carers, employers, etc.) should be offered appropriate training.*

Policy Recommendations

Social care assistance at CoEs

- ***A dedicated social support worker/welfare officer with knowledge of the condition should be available at CoE to help patients navigate the social care system.***
- ***CoEs should have access to psychologists to provide counselling to patients, their carers and families.***



Policy Recommendations

Performance management of CoEs

- ***Systems should be set up to performance manage CoEs with representation on committees from patients or patient organisations.***



Policy Recommendations

Research at CoEs

CoEs should:

- ***Establish themselves as research facilities, include patient organisations in the design and execution of studies.***
- ***Ensure appropriate dissemination of research results to all stakeholders.***



Future Use

- Can this process be repeated?
- Re-evaluation every three years?

- More countries can plug in
- European database?
- Translation issues?
- Going from recommendations to reality



Questions?


EURORDIS
Rare Diseases Europe


Rare Disorders Denmark


NHS


Fundació
Doctor Robert
UAB CASA CONVALESCÈNCIA