Guidelines for organisations providing information on rare diseases

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Preamble

All people, irrespective of race, creed or nationality should be entitled to a high standard of relevant health care. The highest standards and ethics in the practice of information service should be promoted.
Access to information is a fundamental right, whether the disease is common or rare. Information on rare diseases is one of the most important services that patient groups can provide. This is specifically because people with rare diseases feel isolated by the rarity of the condition affecting them or their family and the additional issues raised by the genetic cause of most rare diseases.
It is also because of barriers that exist in accessing information on rare diseases that is appropriate, validated and understandable. There is confusion because many rare diseases are complex syndromes with several definitions and synonyms, which isolates affected people even more.

Rare diseases in Europe represent[1]:

- 5 to 6% of EU population (27 000 000 people after enlargement)
- 6000 to 7000 or even more different diseases.
- A very heterogeneous population with most of patients' groups representing a few tens to a few hundreds of people.
- 80% are of genetic origin.
- They cause disability (44% with mobility impairment), disfiguring (37%) and source of discriminations and questions, not only for disabled children facing world and society, but also for disabled adults with unaffected children.
- They are chronic; they represent 10% of all diseases, a leading cause of death among young people (35% mortality prior to 1 year of age, 12% between 5 and 15 years of age).
- and no diagnosis exists for about half of the affected people.

[1] Orphanet 2003

Introduction

Patient driven groups are an unparalleled source of information on rare diseases. They have developed an expertise that is unique and which should be exploited to the full, so long as they adhere to the good practices which have been identified and practised by existing groups throughout Europe and which are enshrined within these guidelines.

It is always the responsibility of the information service to have high quality information adapted to the needs of the enquirer whatever their reason may be for contacting the service. In providing information, services should always act with respect and empathy.

These recommendations are intended as a guide to recognise good practice and make every effort to include that good practice within the information service being developed or reviewed. Information provided is based on the experience of patients. It is recognised that this will be dictated, to some extent, by available resources, and whether the service is delivered by volunteers or paid staff. However there are certain core values that should be practised regardless of size, maturity and resources.

They apply universally, for small groups as well are larger ones, and they constitute long term goals to be achieved. Their implementation should reflect the cultural, political, scope and resources background of each group. Finally information is just one of the many services patients' organisations can provide.

Organisational principles

- **PATIENTS' EXPERTISE:**
  Recognise the importance of patients and families as a source of information, expertise and empathy. Ensure that services will involve people with rare diseases at all levels of corporate governance and service development where possible.

- **INCLUSIVENESS:**
  Recognise the value of family and carers and include them where appropriate, as people affected with a rare disease may have physical limitations to move, or may suffer from neurological impairment. There should be no distinction among members whether patients or not.

- **ACCESSIBILITY:**
  Provide information services in settings that are accessible and ensure confidentiality. Severe disabilities are common features of rare diseases. Whenever possible, these services should be free of charge for people with rare diseases and their families.

- **SENSITIVITY:**
  Ensure appropriate and effective services by involving people who reflect the voices of users and carers, as experts in defining their own wishes and needs. Among wishes and needs, identify questions on genetic inheritance as a very frequent need, as a large majority of rare diseases are of genetic origin.

- **HUMAN RESOURCES OBLIGATIONS:**
  Deliver an information service by staff whether voluntary or salaried who are supported, resourceful and accountable.

- **ADVISORY EXPERT COMMITTEE:**
  As information on rare diseases is most often sparse and with limited sources, establish an advisory expert committee with experts to whom to refer social, medical or scientific questions (social workers, lawyers, clinicians and fundamental research scientists). Such a committee could include geneticists when appropriate.

Ethical principles

- **CONFIDENTIALITY AND USE OF INFORMATION:**
  Respect data confidentiality at all times and person anonymity unless directed otherwise in writing by the individual concerned. Ensure that collected data always has a purpose and is used and recorded accordingly. Any data for statistical research or evidence purposes should only be disseminated to a larger audience if anonymised, and following consent.

- **LOYALTY:**
  Ensure that the operator's primary loyalty is to the person to whom they are delivering information, and always in a manner that protects confidentiality.

- **ANTI-DISCRIMINATION RULES:**
  Consider any person without distinction and prevent from discriminating in terms of social situation, education, religion, gender, sexuality, ethnic or geographical origin. Services are accessible to people with rare diseases of all cultures, beliefs, ethnic and linguistic backgrounds.

- **CONFLICTS OF INTEREST:**
  Strive to be independent, autonomous and minimise conflicts of interest.

- **SIGN-POSTING:**
  Rare diseases have major impact in everyday life and not only on patient's health. Have in mind the organisation's limits and make reasonable efforts to offer multi-disciplinary approaches on medical and paramedical subjects, legal and political aspects, social law, ethics, finances… Know to whom enquiries should be referred.

- **OBJECTIVITY:**
  Ensure that advice remains objective and non judgemental.

Procedural principles

Goals : *to provide validated, up-to-date and understandable information on all aspects of the disease to those affected, their families, and the professionals working with them. How to get there:*

- **FIELD OF EXPERTISE AND DIAGNOSIS CHECK:**
  Define the particular area of expertise unique to the information service. Rare diseases are often complex syndromes with various definitions and synonyms. An enquirer may be calling in the absence of an exact diagnosis, or to obtain information on an already confirmed diagnosis. As rare diseases and syndromes have various names and synonyms, it is crucial to verify the enquirer is contacting the appropriate service.

- **COMPLEMENTARY ROLE:**
  Ensure that the information service only consists of explanation, translation in an understandable language and complementary information. The service does not intend to make a diagnosis or give medical advice.

- **VALIDATION:**
  Revisit information regularly and check its validity systematically.

- **IMPACT OF INFORMATION:**
  Be aware that information may be interpreted differently by people according to their emotional state, education and experience. Adjust your approach accordingly, being truthful without causing alarm and letting the enquirer set the pace at which information is given. Prognosis or progression of a rare disease is often severe.

- **CONTINUOUS EDUCATION:**
  Keep track with medical knowledge and medical progress, train volunteers and staff in a continuous manner.

- **ISOLATED PEOPLE:**
  Facilitate contacts between isolated people; establish structured channels of information for very rare diseases.

- **COMMUNICATION SKILLS:**
  Ensure that the enquirer is the centre of attention by demonstrating high communication skills, setting aside personal issues and allowing the enquiry to take as much time as is needed.

- **CLARITY:**
  Ensure that all methods in which information is delivered (information tools) are constantly monitored for accuracy, clarity and accessibility in terms of content, format and appearance.

Eurordis, the European Organisation for Rare Diseases, is a patient-driven network of rare disease organisations and individuals. Founded in 1997, Eurordis comprises 210 associations in 16 European countries.
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More information is available on:
www.eurordis.org
www.europa.eu.int/comm/health

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