EURORDIS
European Organisation for Rare Diseases

EurordisCare® results

Survey results summary
21 June 2005

Contact: Dr François Faurisson
Telephone: + 33 1 56 53 52 60
francois.faurisson@eurordis.org

THE RARE DISEASE DILEMMA: EUROPEAN HEALTH CARE SYSTEMS UNABLE TO DIAGNOSE WHAT PATIENTS ARE SUFFERING FROM

EurordisCare® is a research programme initiated by Eurordis (www.eurordis.org) in 2002, involving rare disease (RD) patient groups, to survey and compare access to care between European countries and between different rare diseases.
EurordisCare (2) compared access to diagnosis between **8** RD[1], in **17 countries,** in **12 languages by 6 000 patients and families.** Members of **69 patient organisations** completed and returned a questionnaire (February to September 2004).

Main results, approximated figures

- **25%** of patients waited **from 5 to 30 years** between early symptoms and confirmatory diagnosis of their disease.
- Before receiving a confirmatory diagnosis, **40%** of patients received first an **erroneous diagnosis**, others received none.
    - This was the case for
        - 25% of patients suffering from Marfan syndrome
        - 50% of patients suffering from Ehlers Danlos disease
    - and for
        - **33%** of patients in Finland, Spain, United Kingdom and Ireland
        - **50%** of patients in Austria, Denmark, Germany, Romania, Sweden and Poland
- Such erroneous diagnosis led to medical interventions that were not based on a correct diagnosis, such as:
    - surgery for 16% of patients
    - medicinal treatment for 33% of patients
    - psychological care for 10% of patients
- Patient mobility: **25%** of patients needed to travel to a **different region** to obtain the confirmatory diagnosis, and **2%** needed to travel to a **different country.**
- The diagnosis was announced in **unsatisfactory terms** or conditions in **33%** of cases, and in **unacceptable** ones in **12.5%** of cases.

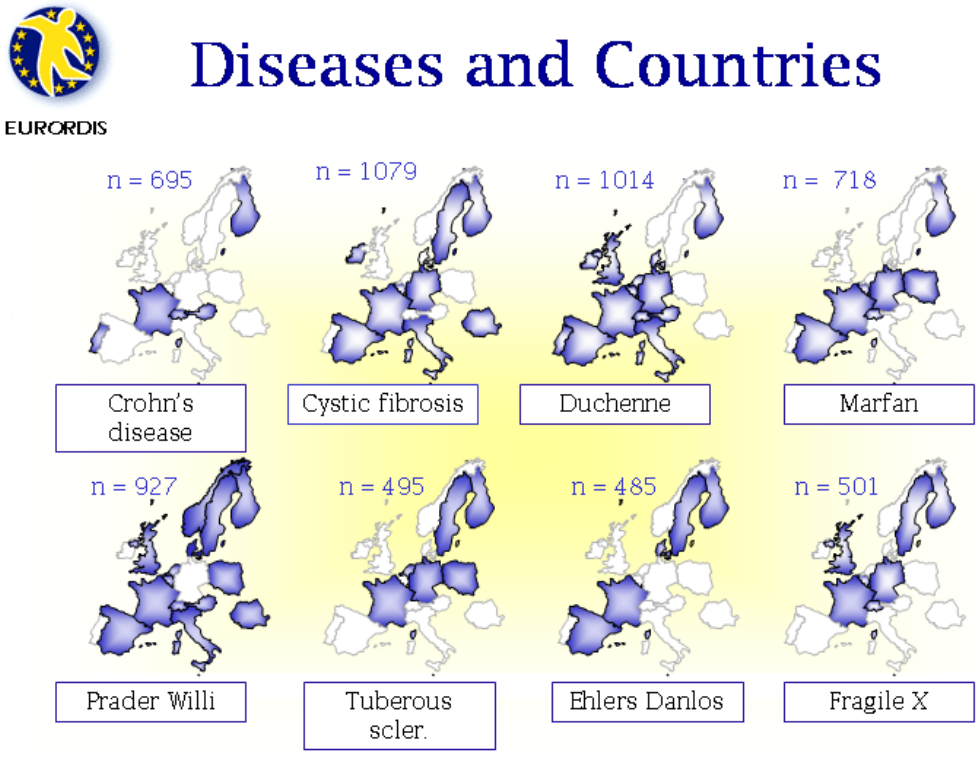
[1] Crohn's syndrome, Cystic fibrosis, Duchenne muscular dystrophy, Ehlers Danlos, Fragile X, Marfan syndrome, Prader Willi syndrome, and tuberous sclerosis

- The **genetic nature** of the disease was **not communicated** to the patient or family in **25%** of cases. This is paradoxical, given the genetic origin of 80% of rare diseases.
- Genetic Counselling existed only in **50%** of cases.
    - In only 40% of cases, an effort was made to discuss the diagnosis and the genetic risk
    - on the other hand, 80% of patients or their parents spontaneously engaged a debate within the family to help diagnosing or preventing other cases, and when this happened, it helped diagnosing others in 30% of cases (10% affected, 20% healthy carriers).

Conclusions

- The results of the survey highlight the dilemma of rare diseases: lack of information, lack of appropriate medical training, difficulties in accessing care, and as a result, loss of confidence of patients in the health care system and the medical profession.
- Detailed results, extended national data and an analysis of explanatory factors will be presented during the European Conference on Rare Diseases, ECRD 2005, in Luxembourg, 21 June 2005.
- Solutions exist and will be debated during the conference: reference centres, data bases for exchange of information, DNA and tissues banks, networks of professionals etc.
- A coordinated European policy for rare diseases is urgently needed.

Annex



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