

The Italian National Centre for Rare Diseases: the experience of the Telephone Helpline

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Background

A national Telephone Helpline for rare diseases is established at the Italian National Centre for Rare Diseases (CNMR) in Rome since February 2008 as a public service reached on 800 896949. The helpline is free and anonymous and works 5 days per week from 9 am to 1 pm. A team of psychologists, sociologists and medical doctors trained and experienced on telephone counseling, public health policies and management of rare diseases are involved. Main aim of the service is to inform health operators, citizens with rare diseases and their relatives on the exemptions from the costs of medical cares as well as on the national network of specialized centers according to the Italian Ministerial Decree (M.D.) n°279/2001.

A web-based system is used for data collection and access to data regarding centers, support groups, advocacy organizations and online scientific literature.

Objectives

To evaluate the number of calls, details of callers, time and nature of queries and the relative proportion of rare diseases enquired.

Methods

Observational study on telephone calls from March 10th 2008 to March 10th 2010.

Results

We recorded a total of 6781 queries equally distributed throughout the country. A limited number of e-mails, faxes and ordinary mails were not taken into account.

Calls were from northern (34%), central (26%) or southern [+ islands] (35%) Italy [FIGURE 1].

Main enquirers were patients (48%), patients' relatives (36%), general practitioners or specialists (7%) [FIGURE 2].

Callers were females (68%), males (31%), unknown (1%). Age of callers was 41-50 years (16%), 31-40 years (15%) and 51-60 years (13%) [FIGURE 3].

Patients were females (54%), males (33%), unknown (13%). Age of patients was <18 years (31%) [FIGURE 4].

As represented in [FIGURE 5] queries regarded:

- 1) Public Health (54%) [Benefit or tax credit issues, information re medical centers].
- 2) Clinical (18%) [General guidance in the field of rare diseases, research of specific topics in the scientific literature].
- 3) Social Area (10%) [General benefits for disability; lay groups, complaints].
- 4) Clinical trials and orphan drugs (5%).
- 5) Psychological support (1%).
- 6) Other (12%).

Interestingly, *i*) many enquiries did not refer to diseases included in the M.D. n° 279/2001 (data not shown) and *ii*) the commonest calls and complaints were about undiagnosed diseases [FIGURE 6].

Conclusions

- ✓ The service represented a benefit to patients, their families, caregivers and health professionals seeking advice and information about rare diseases.
- ✓ The multidisciplinary team did effectively run the helpline, which may be a valuable support strategy to facilitate the management of rare diseases.
- ✓ The web-based system allowed to keep a database of comprehensive information on rare diseases in Italy useful for a broad range of purposes from basic and clinical research to the development of a generic general practice strategy for individual with rare diseases to improve their overall care.



FIGURE 1 - Geographic distribution

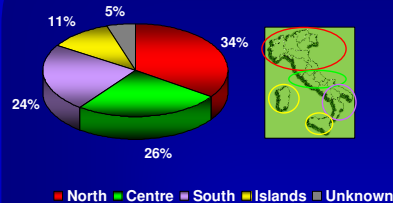


FIGURE 2 - Enquirers were ...

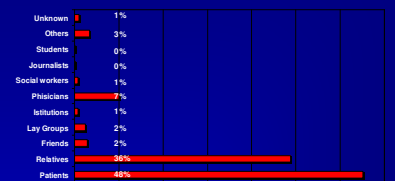


FIGURE 3 - Gender and Age of Callers

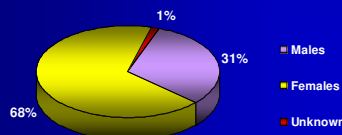


FIGURE 4 - Gender and Age of Patients

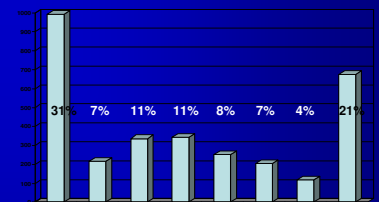
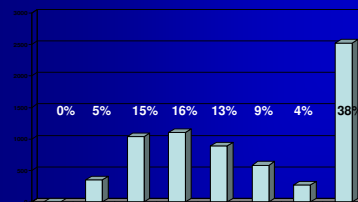
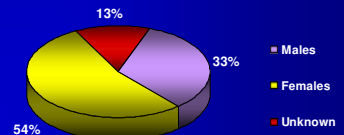


FIGURE 5 - Nature of enquiries

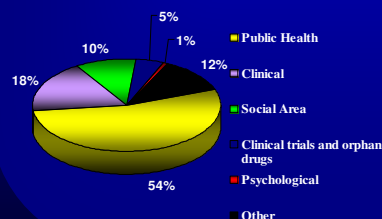
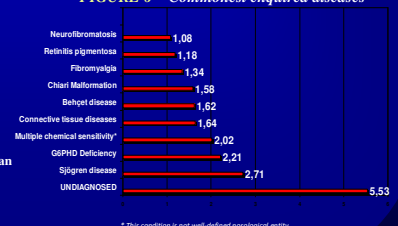


FIGURE 6 - Commonest enquired diseases



* This condition is not well defined nosological entity