



Socioeconomic Data

Total population: 59 619 290 (2008)

Area of country: 301 338 sq km

Population density: 197.6/sq km

GDP: 1 888 trillion \$ (2007)

GDP/capita: 32 319 \$ (2007)

% of GDP spent on health: 8.9 (2005)

of physicians/100 000 inhabitants: 420 (2006)

Italy

Sections of this chapter were written with the collaboration of the Italian Alliance for Rare Disorders (UNIAMO).

National Initiative in the Field of Rare Diseases

The Italian healthcare system is a universal, regionally based public system. The National Ministry outlines funding needs based on historical funding patterns while the regional governments set their own budgets and organise healthcare delivery. The regions establish one or more Local Health Authorities (LHA), which are responsible for the provision of care. Private health insurance is available but not common. Financing comes from both payroll taxes and general revenues. Inpatient care and primary care are free of charge but co-payments are required for some services. Italians must register with a general practitioner in their LHA. A wide range of primary care, specialised care and additional health services are provided under the national system although some inequality exists between regions and overcrowding in hospitals is widespread.

Health authorities and professionals use the European Orphan Drug Regulation definition of a rare disease as having a prevalence of less than one per 2000 people. Discussion regarding a national plan for rare diseases is ongoing, with a scheduled approval in October 2008. Availability of non-medical services for rare disease patients varies considerably from region to region, with high quality services in some regions and no such services in others. Web sites, helplines and patient forums have been created by national alliances and single rare disease organisations as well as some public health institutions. No single coordinating body currently exists specifically for rare diseases and orphan drugs. Many state and regional agencies collaborate on these initiatives together. For example, Istituto Superiori di Santi (ISS) is responsible for the National Rare Disease Registry and Agenzia Italiana del Farmaco (AIFA) engages in discussions regarding orphan drugs and experimental therapeutic treatments and manages a special orphan drug research fund based on a pharmaceutical industry advertisement tax. Orphan drugs put on the market are distributed directly by or near hospitals, free of charge, in all regions of the country. Regions adopt their own policies for off-label use.

The National Ministry, the regions and AIFA provide important funding for rare disease research initiatives. Telethon, a non-profit organisation, is responsible for raising and distributing funds for the advancement of research toward diagnosis, cure and prevention of human genetic diseases

in universities and non-profit research institutes. It has also played a key role in raising awareness of rare diseases in Italy. Reimbursements for orphan drugs are based regionally and compassionate use is granted in very particular cases of patients suffering from very rare diseases. Neonatal screening exists for a few metabolic diseases. Genetic testing and counselling are provided for the most prevalent genetic diseases in Italy and diagnostic centres provide genetic counselling.

Diagnosis in Italy

PARTICIPANTS IN THE SURVEY

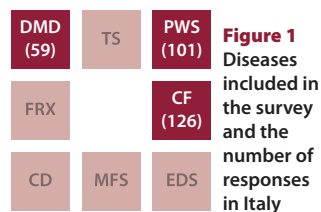


Figure 1
Diseases included in the survey and the number of responses in Italy

Responses from 286 families of patients with three diseases were analysed (*Figure 1*). An equal number of female and male patients were represented in the survey (47% and 53%, respectively).

DIAGNOSIS OVER THE FIRST THREE MONTHS OF LIFE

Neonatal diagnoses were obtained in 28% of patients, almost twice that observed for overall (15%), in particular for detection during pregnancy or at birth (14% compared with 7%) and neonatal testing (10% compared with 4%).

AWAITING THE DIAGNOSIS

Before obtaining the correct diagnosis, another diagnosis was given to 45% of patients, resulting in inappropriate treatments in 75% of cases, including medical (35%), surgical (7.5%) and psychological or psychiatric (7%) (*Figure 2*). During the quest for diagnosis, more than five physicians were consulted by 29% of families and more than ten by 9% of families. Physicians prescribed tests for 94% of patients, including biological tests (84%), genetic tests (42%), functional tests (51%) and X-rays (44%).

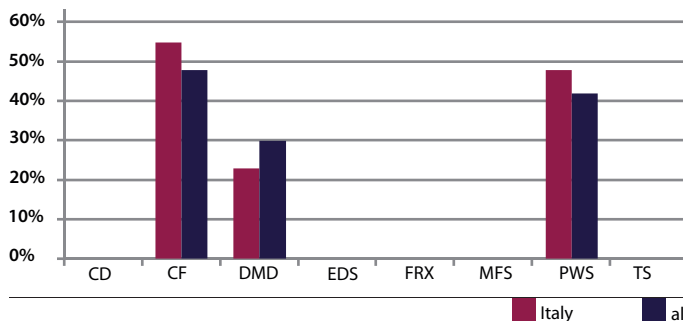


Figure 2
Percentage of patients initially receiving a misdiagnosis in Italy

DIAGNOSIS

The structures providing the diagnoses were hospital consultations (73%), specialised centres (23%) and rarely private practices (1% compared to

10% overall). These were located in another region in 21% of cases and in another country in 5.4%, representing almost three times the overall value of 1.9%. Personal cost was globally higher than that observed overall (Figure 3 & 4). Free diagnoses or low expenses were reported by 58% of respondents, whereas high or very high expenditure was reported by 21%. A second opinion was sought to confirm the diagnosis in one in three families, compared to one in five overall.



Figure 3
Personal expenditure for obtaining a diagnosis in Italy



Figure 4
Personal expenditure for obtaining a diagnosis overall

very high high moderate low no

ANNOUNCEMENT OF DIAGNOSIS

For 26% of patients, diagnoses were given without complete information on the disease; however 98% of respondents considered this information to be necessary. A total of 93% considered that psychological support was necessary at the time of the announcement of the diagnosis, whereas 78% of patients did not receive such support.

The genetic nature of the disease was explained to families in 86% of cases, with details given about the possibility of other cases in the family in 61% of cases. Genetic advice resulted in the diagnosis or identification of a carrier in family in 27% of cases.

Respondents considered the conditions of the announcement to be poor or unacceptable in 26% of cases (Figure 5).

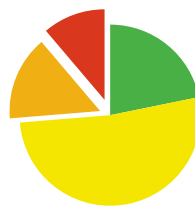


Figure 5
Conditions of the announcement of diagnoses in Italy

unacceptable poor acceptable well-adapted

For 77% of the families, delays in diagnosis were considered responsible for deleterious consequences, such as psychological consequences (6% compared to 13% overall) and birth of another affected child (2% compared to 7% overall).

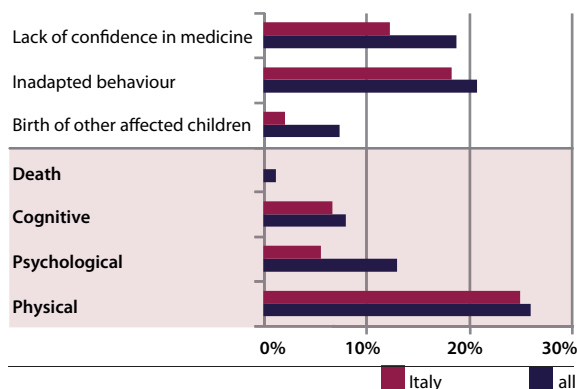
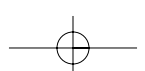


Figure 6 Medical and non-medical consequences of delayed diagnoses in Italy



Access to **Medical** and **Social Services** in Italy

PARTICIPANTS IN THE SURVEY

AH (16)	Ch11 (4)	WS (30)	PWS (111)
FRX (31)	EB (42)	TS (12)	CF (100)
ANR (31)	OI (84)	MFS (38)	EDS (21)
HD (33)	MG (79)	ATX	PAH (59)

Figure 7
Diseases included in the survey and the number of responses in Italy

Responses from 691 Italian families of patients with 15 diseases were analysed in the survey (*Figure 7*).

The proportion of female and male patients represented were 55% and 45%, respectively.

The mean age of patients was 29 years (mean age at diagnosis: 16 years).

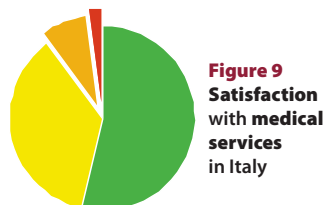
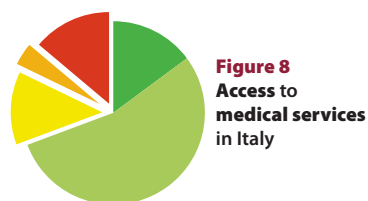
NEED FOR MEDICAL SERVICES

Overall, Italian patients needed 9.7 different kinds of medical services related to their disease, which was close to overall value. Hospitalisation occurred in 53% of patients for an average total duration of 21 days.

MEDICAL SERVICES

The access to eight essential services for each disease was easy in 70% of cases, difficult in 17% of cases and impossible in 14% of cases (*Figure 8*). Difficulty was mainly due to lack of referral (59%), unavailability (20%), as well as waiting time (20%), personal cost (9%) and location of the structures, including a location too far away (15%), no one to go with (5%) and difficulty in travelling (10%).

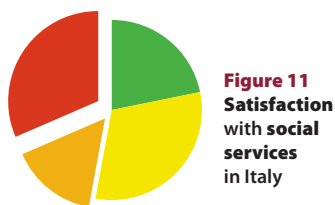
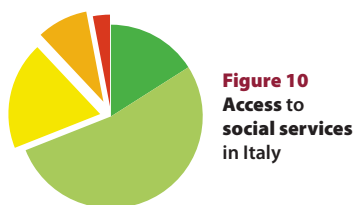
When obtained, the medical services responded well to patients' expectations in 90% of cases and poorly in 10% of cases (*Figure 9*).



impossible very difficult difficult easy very easy not at all poorly partially fully

SOCIAL ASSISTANCE

Amongst the 26% of Italian families that required social assistance, 3% failed to meet with a social worker, whereas 69% met with one easily and 28% met with one with difficulty (*Figure 10*).



impossible very difficult difficult easy very easy not at all poorly partially fully

Community care structures play a more important role in Italy than overall in Europe (71% compared with 53%). A total of 53% of Italian families were satisfied with this assistance while 32% were not at all satisfied (Figure 11).

REJECTION

Italian patients experienced rejection by health professionals slightly less frequently (14% compared to 18%) than respondents overall for the 16 surveyed rare diseases.

The reluctance of health professionals to treat patients due to the complexity of their disease was the main cause of rejection (85%), followed by disease-related behaviour (15%), communication difficulties (14%) and physical aspects (5%) (Figure 12).

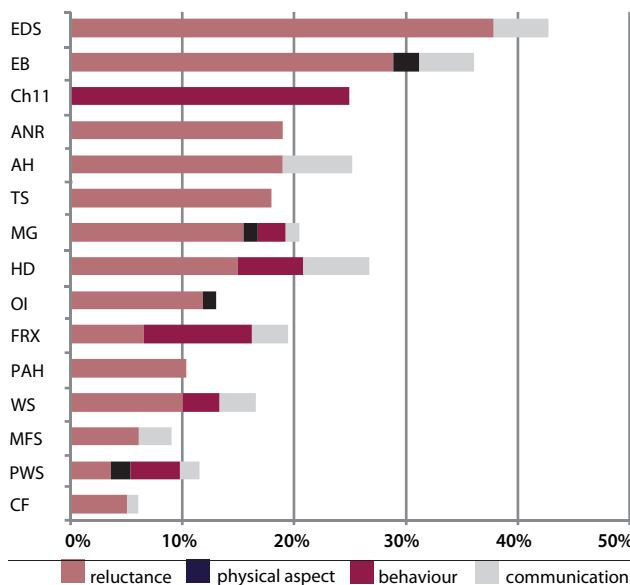


Figure 12 Cumulated frequencies of cause of rejection, by disease, in Italy. As patients may have been rejected more than once for more than one reason, the total number of rejections exceeds the number of rejected respondents.

CONSEQUENCES OF THE DISEASE

As a consequence of the disease, 12% of Italian patients had to move house. Amongst these, families most frequently moved to a more adapted house (54%), to be nearer to disease specialists (25%) or to be closer to a relative (25%).

As a consequence of their disease, 19% of patients had to reduce or stop their professional activity. In 36% of cases, a member of the family had to stop working in order to take care of a relative.