

# Rare Diseases in ARGENTINA:

## 144 Survey of physicians' knowledge, practices and opinion

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### INSTITUTIONAL FRAME

As a part of a scientific research project entitled "Rare Diseases in Argentina: a socio-sanitary, legal and scientific approach. Communication and policy proposals for reducing vulnerability", a survey of physicians was performed in 2008. The research is being executed by Fundación FOP and supported by the Agencia de Promoción Científica y Tecnológica (part of the Argentinian Ministry of Science, Technique and Productive Innovation) and two private foundations: FUNI and Gianantonio.

### INTRODUCTION

Rare diseases affect 6 - 8% of the European population. Due to the absence of accurate local data, these rates help to estimate that approximately 3 million people in Argentina suffer from a rare disease.

Knowledge of rare diseases is still very recent and not so widespread in the health network.

Lack of related information by health professionals brings negative consequences to both patients and their families such as difficulty in obtaining a fast and correct diagnosis, lack of appropriate treatments and even the absence of specific medication.

**Primary care physician's role is essential** and must include: detecting early warning signs, accelerating the visit to specialists for diagnosis and guiding the patient to corresponding centres of references.

Knowledge of how and where to search information allows the diagnostic process optimization.

### OBJECTIVES

- To determine physicians' state of knowledge on the new concept of rare diseases as a public health issue.
- To know physicians' steps when diagnosing or treating a patient who suspects being suffering from an uncommon disease.
- To get information on proposals to improve rare diseases local situation among physicians.

### METHODS

- The survey was exposed to physicians users of INTRAMED (www.intramed.net), a Latin American medical website.
- Collection of responses: exclusively on line.
- Exposure time : from October 20th. to December 31st. , 2008.
- Exposed target population: 11375 Argentine physicians.
- The form's 17 questions described:

- Respondent's characterization and workplace.
- Knowledge on the new concepts: Rare Diseases and Orphan Drugs.
- Physicians' experiences in suspecting, diagnosing and treating rare diseases.
- Rare diseases diagnosis and treatment daily practice.
- Used sources of information on rare diseases.
- Knowledge of patient advocacy groups.
- Proposals to improve the local situation.



### RESULTS

950 Argentine answers

- Average age : 44,8 years
- Clinical Specialities 85,1%
- Surgical Specialities 14,9%

#### 1. KNOWLEDGE \* Do you know what the term "Rare Diseases" refers to?

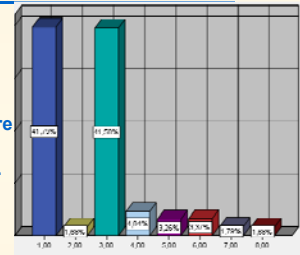
YES 749 (78,8%) NO 201 (21,2%)

##### \* What is the defining criterium of a Rare disease?

- Etiology .....31.....(4,1%)
  - Display of unusual symptoms .....48.....(6,4%)
  - Prevalence .....625.....(83,4%)
  - Lack of scientific knowledge .....45.....(6%)
- \* Orphan drugs definition:**
- Scarcely profitable medicinal products pharmaceutical industry is little interested in 77,4%
  - Medicines scarcely available in marketplace .....17.8%
  - Medicines no one indicates for their high price.....4,8%
- \* Rare Diseases common features:**
- Most of them are genetically originated .....32,7%
  - They are subject to public policies in Europe and US .....16.4%
  - Their diversity does not allow to create common health public policies .....67.4%

#### 2. ASPECTS USUALLY TAKEN INTO ACCOUNT WHEN REFERRING A PATIENT WITH A DIAGNOSED / SUSPECTED RARE DISEASE.

- Complexity of the Centre. (41,7%)
- Nearness of the Centre.
- Knowledge of rare diseases reference centres/services. (41,5%)
- Possibilities offered by patient's health-care coverage.
- Workplace procedure/administration rules.
- Patient and/or family's preferences.
- Kind of institution (public or private).
- Other.



#### 3. PATIENTS & FAMILY ORGANIZATIONS

##### \* Do you know the existence of organizations of patients and families of any RARE DISEASE?

YES 477 (50,2%) / NO 473 (49,8%)

- Are you currently linked to patient and families organizations of any RARE DISEASE?
- YES 392 (17,8%) / NO 85 (82,2%)

#### 4. SOURCES OF INFORMATION ON RARE DISEASES.

- Basic bibliography .....71.9%
- Expert Professionals on the subject.....69.6%
- The Internet.....68.9%
- Interhospital Communication Systems.....23.4%
- Other.....15.1%

#### 5. PROPOSAL TO IMPROVE LOCAL SITUATION.

97.1% of respondents believe it is necessary to improve knowledge of Rare Diseases among physicians.

Selected options to improve physicians' daily practice of diagnosing and treating rare diseases patients:

- Internet Sites with specific local information.....62,2%
- Exclusive rare diseases national/local consultation office .....20,9%
- Interhospital Communication Systems .....13,9%
- Other .....3%

### CONCLUSIONS

- Even though physicians consider and work on these pathologies, knowledge of the new approach of rare diseases as subject to policies is limited.
- Although rare diseases issue is slowly being installed in Argentina, physicians' daily practice indicates difficulties in making right diagnoses and accessing to appropriate information. In addition, their scarce of knowledge on the genetic origin that most of rare diseases have, may limit appropriate referring that may accelerate diagnosis process.
- A website including local data may improve information circuit among professionals, accelerating the diagnosis process for health teams, patients and families.