

WORKING GROUP D: OBTAINING THE DIAGNOSIS OF RARE DISEASES

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At the beginning, the coordinator of the working group proposed two points for the discussion:

- 1) To talk about the problems connected to diagnosis of rare diseases
- 2) To discuss newborn screening

High quality of the diagnosis is crucial

It was recognized that methods and techniques for obtaining diagnosis are well developed. Instead, the interpretation and the use of analytic results is susceptible to improvement. The best way to overcome the problem is to participate to one of the existing external quality control systems, such as CF network, EQAS, EMQN, EUROAGENTEST, etc.

However, there are more than one 1000 tests and less than 50 are covered by quality control systems. The organization of ring test could support the extension of quality control in the field of diagnostic tests.

Another suggestion would be a platform specific control by assuring the quality of a single technique (e.g. DNA sequencing).

ICORD should collect existing guidelines and support setting of international standards on diagnosis.

Is the quality control of diagnostic test regulated by law in various countries?

Regulations that demand quality control of diagnostic tests do not exist in all countries. For instance, in Germany regulations demand EQAS, but do not require ISO; in Brazil, developed diagnostic tests often do not translate from research projects to health practice. The group concluded there is no consistent approach between countries. Therefore, ICORD should support transnational information sharing and suggest the grounds for regulations of diagnostic tests at the international level.

Genetic counseling of a patient and its family

Parents of a child with a rare disease often expect that the test will bring change to their life. Unfortunately, diagnostics is not always followed by existing treatment or drug. Thus, the existence of a genetic counseling is very important; the clinical usefulness of each test should be established.

The existence of informed consent is by all means the prerequisite for any genetic analysis.

Reference centers and networking

The information about the availability of a test within the country is frequently hard-to-get. There should be good networks within countries in order to support information flow and to address samples to reference centers, thus reducing costs and patients distress (e.g., need to travel).

ICORD should discuss and disseminate information on:

- a) requirements of centers for molecular and clinical testing;
- b) fitting diagnostic tests in the public health framework of rare diseases (diagnostics, classification, reference centers, treatment, care ...).

ICORD can also contribute to disseminate information on existing actions and activities on rare diseases in countries in order to share experiences and support priority setting at national level for the coming years. This preparatory work will facilitate a policy framework. Policy makers should be informed that poor quality diagnostics will bring more costs to the social system.

Examples were made:

- Italy: each region has its own reference centers and a network operates to coordinate them. However after ten years of such organization, an effort still has to be made to maintain communication between centers.
- Japan: there are no diagnostic reference centers, every expert is working inside its own institution.
- Brazil: there is no general plan, there is no contact between academia and government, there is no good information flow, it is a problem to collect data and calculate the statistics.

Networking is all important when there is no test for diagnosis of a rare disease. In such cases networks will help address samples outside the country, where the test can be performed.

Information flow

There are several information networks (e.g. ORPHANET) and they should be diffused. Better links between existing information websites should also be made.

Training of medical operators

Students should be trained to know more about genetics and rare diseases. But, we should also think about the education of other medical operators – nurses, physiatrists, acupuncturists – they should also be able to recognize when they are incapable to help and when a serious patient needs to be sent to a diagnostic centre.

For instance in Argentina medical doctors began to receive additional training to recognize inborn metabolic diseases. It was recognized as a useful approach.

Conclusion points:

ICORD should discuss and support the following actions:

- 1) To make international quality control systems by using and expanding experiences of the existing systems
- 2) To set reference centers
- 3) To organize networks between reference centers
- 4) To collect guideline for diagnostics
- 5) To train students, medical doctors and other medical operators to know where to send the patient when they can not make diagnosis
- 6) To disseminate validated and up to date information

FUTURE WORK PLAN:

- 1) To collect information on actions and policies from countries in order to share experiences along the line set by EUROPLAN
- 2) To share experience from existing sources and make those information available at the ICORD website
- 3) To start reflection on how to build a public health policy in this field