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List of the Top Ten Ethical Tensions and Philosophical Issues *Working Group on History, Philosophy and Ethics*

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1. Prenatal and neonatal screening

The technologies are available or are in progress to diagnose accurately more and more RD. It is likely that they will be proposed as tools for both neonatal and prenatal screening. It is necessary too establish the criteria for their correct use attending to the principles of justice and beneficence.

Neonatal screening for conditions with no treatment may generate harms and social burden. Prenatal screening will pick up borderline conditions and will generate information difficult to interpret in the context of a possible decision of pregnancy termination.

Nevertheless, it is important to create the opportunity for making the choice even in the case of rare diseases. Producing quality information about good practices is an ethical need.

2. The role of the environment in the genetic make-up of the individual

The model of causation of genetic diseases is changing. A major role is attributed to the epigenetic factors and multi-step causality. Environment can play a fundamental role and it is a moral obligation to provide the best environment for development.

3. Health for all as a fundamental human right

Health is not simply the absence of disease, its value and significance being more general. It means the possibility for everyone to enjoy the best possible quality of life, independently of his or her biological, social and cultural characteristics.

Lessons should be learned, especially from parents' associations, for:

- Avoiding stigmatization and discrimination
- Increasing resilience
- Changing the conditions of life, social acceptance, and integration

4. Allocation of resources. Increase transparency of decisions

Given the limited resources available, public health policies must establish priorities, without excluding the most vulnerable, that may be the persons affected by the most rare diseases.

- How to establish priorities`
- What to do first?
- Who should decide?

The main social problem in this context is that the welfare state was created to offer social assistance to the higher number of citizens. As a consequence health services are in general built to treat frequent and acute diseases ('standard acute patient'), and very often they are inadequate in coping with chronic and rare illnesses, which are in large part individual and chronic. This inadequacy is causing a heavy social burden.

It would be unethical to create a new market with limited efficacies. It would be essential to establish a list of the most effective treatments available for each disease, which should be implemented in priority and in respect of equity.

5. Limits of the therapeutic interventions

Even if this is a difficult subject, it is sometimes ethically correct to decide not to treat in very severe conditions, especially if this is accompanied by effective psychological work with the families. Nobody should look for miracles.

This ethical debate is quite advanced in relation to very premature babies and to dying persons. Some lessons can be learnt from these debates and applied to the field of rare diseases.

6. «Feeling guilty»: an obstacle to the appropriate action

Frequently, the presence in a family of a patient with a rare disease is accompanied by a sentiment of guilt or shame. If in the case of infectious diseases «the enemy» is the germ, an entity outside our responsibility, in the case of genetic disease the cause is in our body, within us, and the responsibility for the birth of a baby carrying a rare disease is attributed to the parents and their behavior. This is a major cultural and social problem which deserves closer attention and the development of educational programs.

7. Increase the visibility of the most vulnerable populations. Increase social reconnaissance of rare diseases

Notwithstanding the great successes realized in the last decades, much social and cultural work is needed in this field.

8. Ethical issues of data collections: Databases, Biobanks, and cohorts for epidemiological and clinical studies

Many ethical and philosophical questions are connected to this topic, whose importance has enormously increased in the last couple of decades:

- Privacy
- Patenting
- The use of collected knowledge (i.e. registries) for scientific research
- Misuse of collected information (risks of discrimination in the workplace, schools, professions, insurances)
- Intellectual property
- Criteria for coding and classifying the specimen and the relative diseases
- Comparability of the data collected in different countries and often different institutions. The analyses of the registries for other diseases, such as cancer, can be of great help.

9. «Naming the disease»: philosophical aspects of the Classification of Diseases

As the number of known rare diseases increases, the need for an appropriate classification and codification becomes evident, in order to increase their rationality and visibility in national health systems and in epidemiological surveys.

The process of classification depends on several criteria, namely the accuracy of medical diagnosis, disease terminology, and the selection of classification systems. Diseases are usually classified according to their cause, the organ involved, the symptoms, and the syndromic complex. However, these criteria cannot be used easily for classifying genetic diseases, which are usually systemic and of unknown causation.

10. Decision-making and ethical issues

In the allocation of resources four main actors are present: Academia, Governments, Patients' Associations, Industry. Independent associations usually have very limited funding and that makes them quite vulnerable to external economic influences. The partnership public-private-associations is essential, but the decisional autonomy of the academia and patient's associations and organizations like ICORD must be preserved. This independence is an ethical obligation at the decisional level and ethic committees could be established, following the model established in many universities in relation to private funding.

The autonomy and the independence in decision making requires the availability of correct information at the scientific, economical, and political levels. Even if scientific knowledge is always incomplete, it is an ethical imperative to make the best information available, to diffuse it to the concerned individuals, associations, and institutions and to the whole society, in order to avoid the neglect of rare diseases and the neglect of the rights of people affected by them.

Actions to be undertaken

1. *Creation of a permanent working group within the ICORD on the historical, philosophical, ethical and social issues of Rare Diseases*

First report to be prepared for the ICORD 2011

2. *Implementation of the specific WEB Site opened by the Istituto Superiore di Sanità (Rome).
Implementation of a specific and permanent discussion forum*

A preliminary version is already available for evaluation on the web site of the ISS at the following address :

http://www.iss.it/bozza_nrar/

3. *Creation of an International Network on the historical, philosophical, ethical and social issues of Rare Diseases.*

Main aims: share the information; diffuse the relevant publications; creation of a specific bibliographical database; joint organization of conferences and workshops

4. *Organization of specific events*

6-11 septembre, 2010, Hermance (Geneva): Advanced Seminar on Philosophy of the Life Sciences: «Causation in post-genomic era».

More information on the website: <http://www.brocher.ch/pages/default.asp>