Working Group Food, Environment and Rare Diseases

Summary

The Working Group has involved a limited group of participants to ICORD 2010, possibly also because the topic of the role of dietary and environmental factors in rare diseases is rather new and still not clearly defined. On the other hand the discussion was lively and put the basis for further development of priority issues within ICORD activities.
The following conclusions were derived and presented to the plenary ICORD meeting:

- There is no doubt that quality and safety of food and environment are relevant to rare diseases (RD), hence, these have to be considered within ICORD mission and activities So, the relevant question is not *whether*, but *how* and *in what instance.*
- In some instances, specific food- and environment-related factors are involved in the *pathogenesis* of RD. Telling, as well as widely recognized, examples are i) the insufficient intake of folates/folic acid concerning the pathogenesis of neural tube defects, representing the instance of a major risk factor; ii) aniline-denaturated rapeseed oil in toxic oil syndrome and *C.Botulinum* in infantile botulism, representing instances of primary causative agents. Likely, the progress of research on gene-environment interactions will increase the knowledge on specific risk factors involved in certain RD groups, such as birth defects and rare tumours.
- In many other instances, diet or living environment may have an *important modulating effect* on the course, the severity, the prognosis of rare diseases. Telling examples, quoted during the discussion, were the high copper (and/or low zinc) in the diet for Wilson disease and inadequate intake of dietary lipids in Marfan disease. In these, and several other examples (e.g, inheritable metabolic defects) the origin of disease is genetic, but the "environment" (in its broader sense of living scenario) may have important bearings on the clinical signs and in general on the quality of life.
- *Think positive*: the knowledge on causative agents, risk factors and/or modulating factors should *translate into prevention* of RD.

Prevention may be meant as primary, secondary or tertiary prevention according to specific cases. Actions may include measures, recommendations and information of health operators and the public and be aimed at the levels of general population, target groups or individual/families. In all cases, a *proactive* approach should be taken.

- *Research* to increase knowledge is critical, but making the *best possible use of available information* is critical as well.

The Working Group strongly endorses the diffusion to health operators, target groups and patients /families of *recommendations o*n the management of diet and living environment. Such recommendations should be devised in order to
i) provide an understandable message;
ii) support the awareness and empowerment of citizens, a point where patient associations may provide major inputs;
last but certainly not least iii) be *based on adequate scientific evidence.*

- Finally, the Working Group considers that general public health recommendations and measures on safety, diet and lifestyles must be included in the health management of citizens with RD. The Working Group notes that in some instances such measures/recommendations might be relevant to primary prevention of specific RD, as shown by the possible association of obesity and/or high periconceptional blood sugar with increased risk of neural tube defects and other malformations.