INTERNATIONAL PATIENT ORGANISATION FOR PRIMARY IMMUNODEFICIENCIES

A response to the EU Commission's public consultation on Rare Diseases – Europe's Challenge

IPOPI welcomes the publication of this Consultation document. Anything that serves to highlight the problems of people with rare disorders is to be welcomed and this particular publication shows how well the Commission staff has co-operated with and listened to those who represent people with rare disorders. As we learn more about the incidence of rare disorders it becomes increasingly clear that the combined incidence is much greater than had been anticipated and, equally, that inappropriate and late diagnosis and management is a costly factor. Our comments are made against that background of making the most effective use of existing health care resources.

Questions One and Two:
There is a clear need to have an agreed codification and classification for rare disorders so that an accurate record can be made on the life expectancy/mortality of that disease. An effective starting point is that of making sure that there is a commonality of terminology and that this is kept current in the light of evolving medical knowledge. Through treatment with new drugs and/or therapies a continued assessment of a particular rare disorder can be achieved.

Question Three
The experience gained from national knowledge can be useful but with rare disorders it is more effective to collect data more widely and this is an opportunity afforded by the EU. A major obstacle to the collection of meaningful data is that not all clinicians are happy to 'share' their anonymous, codified, data – even knowing that by doing so they might have access to more meaningful data collated from a wider input. Funding and indeed incentives are needed to achieve a widespread of participation.

Question Four
Patient organisations strive to enable people to be aware of their need of formal diagnosis. We have known people to take an effective step towards their diagnosis after reading a single line on a patient

organisation Christmas Card – how much more effective we have found easily accessible and well informed patient organisation websites. However, the 'reach' of those sites is limited and there can be little doubt that a solidly funded site for Rare Disorders such as that envisaged by the Commission in this Document would serve a wider community more effectively. The imprimatur of the Commission adds great value and credibility to the information provided.

Question Five
The experience in primary immunodeficiency has shown very clearly that lack of early diagnosis as a result of (a) inadequate medical awareness and knowledge and (b) a reluctance to make referrals to other specialists. This leads to delays in diagnosis and appropriate therapy.

Question Six and Seven
More dialogue between various medical experts might result in earlier detection of a RD as would inclusion of RD recognition in CME (continuing medical education). It is interesting that in some US States there is screening of all babies for severe combined immunodeficiency (SCID) – the screening in Wisconsin costs $70,000 a year, whereas repeated admissions hospital would cost more and not be effective. In order to obtain data from a critical mass it is essential that the collection and surveillance be on a Europe-wide basis. This makes it imperative that European guidelines on the diagnosis and treatment of patients with a rare disease should be developed asap.

Question Eight and Nine
Once an orphan drug has been approved by EMEA and a marketing authorization has been given then this drug should be made available in all member states – if this is not immediately possible, then it should be made available at centres of network or reference.

Prevention of certain rare disease is difficult and therefore there should be a greater emphasis on diagnosis and treatment.

"WHO empowerment – a pre-requisite for health and a pro-active partnership and patient self-care strategy to improve health outcomes and quality of life amongst the chronically ill" – How is a severely handicapped/chronically ill patient going to organise a "self-care strategy"? Patient organisations - especially for small groups of rare diseases are essential and where possible should join with their partner organisations in other European countries and further more form international groups, thereby becoming a stronger voice.

A European database/inventory of rare diseases is invaluable and will help to raise awareness and improvement of quicker diagnosis and consequently treatment. With the implementation of national reference centres this should further improve the European cohesion on dealing with RD.

Knowledge should be shared. There are so many eu based health organisations and medically based institutions., that there should be more sharing of information rather than aiming for individual funding for yet another project. Cross-border treatment especially for RD should be made easily available.

On-line and electronic tools should have a classification of quality of information.