α1 ALFA EUROPE Fed.
EUROPEAN FEDERATION FOR ALPHA1 ANTITRYPSIN DEFICIENCY

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Response to:
Public Consultation
Rare Diseases: Europe’s Challenges

Alpha 1 Antirtypsin Deficiency (AATD) is one of the most common hereditary disorders but is yet a rare disease as described in the document.. It is caused by the production of abnormal AAT protein in the liver. Persons with AATD are at high risk of developing life threatening lung and liver disease.

I propose to answer the questions as put and may comment on some as they apply to AATD/

Q1. We are satisfied with the current definition of RD

Q2. There is such a need for coding and classification.

Q3. A European inventory of RD would clarify a lot and would also raise the profile of RD.

Q4. AATD is often/generally misdiagnosed and can take years before proper diagnosis is made. Reference Networks would be helpful with patient group input.

Q5. Yes use should be made of e-tools.

Q6. Testing for AATD is quite simple. The problem lies not with labs but with primary healthcare where there is a lack of knowledge.

Q7. Appropriate screening and detection programmes are essential. Re AATD these programmes are fairly sketchy and differ widely in different countries. National government support is necessary.

Q8. Accessibility to orphan and other products is a problem and it is one that seems may get worse if healthcare companies reduce the production of these products as seems to be planned. Remuneration problems exist for many patients. A common best practice remuneration system might be worked out for all EU countries. This would need to be done in consultation with patient representative groups. Product availability should be common to all EU citizens.

Q9. This does not apply to AATD

Q10. With regard to AATD Information and proper counselling are essential not only for the person diagnosed but also for their family members who may have to be tested for the condition. Patient support groups and peer counselling need to be assisted. Psycho social services are essential to persons diagnosed with life threatening diseases as is financial support when they have to cease employment.
Access to transplant and other necessary medical therapies need to be easily available.

Q11. Registries are an essential tool in the treatment of RD and they need to be internationally compatible. As they are costly to set up and run they need to be financed by national governments. European databases and registries ought to be supported financially by the EU. All should be registered with national data protection commissioners.

Q12. The EPPOSI model as adopted by IPPOSI in Ireland is appropriate and works well. It would be well to set up such as IPPOSI in all EU countries. Patient groups, science and industry all have vested interests and common cause.

Q13. National plans and national centres of excellence (licensed) are essential to the progress of treatment and research for RD.
Further encouragement for both basic and clinical research should be recommended. The Irish model of Medical Research Charities Group and Health Research Board research funding scheme might be developed further in Ireland and in other EU countries.

Q14. We agree totally with the idea of a European Agency which will be representative of all stakeholders.

The main problem with AATD is at diagnosis level. There is an awareness deficit at general practice level throughout Europe. We need a lot of help to eliminate the awareness deficit. A lot of money and effort has been spent by patient groups in awareness campaigns and meetings without a lot if interest being shown by general practice healthcare professionals. Patient groups need more support at national and EU level. They are the best advocates for the RD populace.

Many thanks for your kind consideration,

Yours sincerely,

Larry Warren.