

From: Milan Macek [milan.macek.jr@lfmotol.cuni.cz]
Sent: vendredi 18 janvier 2008 23:47
To: SANCO RARE DISEASES CONSULTATION
Subject: full support from the Czech Society for Medical Genetics to the Public Consultation

Ref. Public consultation regarding European Action in the Field of Rare Diseases

To Whom It May Concern

the Czech Society for Medical Genetics (www.slg.cz) fully endorses the Community Action Programme on Rare Diseases, which still form the backbone of genetic testing with Europe with clear clinical utility. Rare diseases are still an useful useful model and create important paradigms for the study of multifactorial disorders. Their research, traditionally strong in Europe, should receive institutional support from the EC.

Itemised answers to Questions in the Document

1/ Question 1: is the current EU definition of a rare disease satisfactory - YES

2/ Question 2: Do you agree that there is a pressing need to improve coding and classification in this area - Yes

3/ Question 3: Can a European inventory of rare diseases help your national / regional system to better deal with Rare diseases - Yes

4/ Question 4: Should the European Reference Networks privilege the transfer of knowledge? Yes, in particular using electronic tools

5/ Question 5: Should online and electronic tools be implemented in this area - Yes

6/ Question 6: Do you see a major need in having an EU-level assessment of potential population screening for rare diseases - Yes, since this is due to there „rarity”, which precludes generation of sufficient cohorts at a national level

7/ Question 7: Do you see a major need in having EU level assessment of potential population screening for RD - Yes this is will be beneficial

8/ Question 8: Do you envisage the solution to the Orphan drugs accessibility problem on a national scale or on a EU scale - EU scale is much better in order to pool resources

9/ Question 9: Should the EU have an orphan regulation on medical devices and diagnostics - Yes and it should always work with professional societies and patient organisations

10/ Question 10: What kind of specialised social and educational services for RD patients and their families should be recommended at EU level and national level - the list provided is comprehensive and sufficient

11/Question 11: What model of governance and of funding scheme would be appropriate for registries, databases and biobanks - the answers provided in the text are sufficient

12/ Question 12 - How do you see the role of partners (industry and charities in an EU acition on rare diseases? - an appropriate answer was provided

13/ Question 13: Do you agree with the idea of action plans - yes including creation of a specialised agency

Should you have any further questions to the document, please do not hesitate to contact us

Prof. Milan Macek and Prof. Petr Goetz /President of the Society/

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