QUESTIONNAIRE ON THE PKU MANAGEMENT AND THE NEWBORN SCREENING IN THE REGION OF THE SOUTH-EASTERN EUROPE

I. ***NEWBORN SCREENING PROGRAM CHARACTERISTICS***
1. The first year of the PKU screening in the whole country:
2. The estimated number of the screened newborns in year 2012:
3. The number of all the newborns in your country in year 2012:
4. Age of newborn when screened (**in hours**):
5. The estimated incidence of classic PKU in your country: Please provide a published reference if exists:
6. The estimated incidence of all hyperphenylalaninemias (cases over cut-off value in screening) in your country:
7. The current laboratory method of the PKU screening:
8. The cut-off value for the phenylalanine in the screening:
9. The estimated proportion of the patients who ever require the diet, comparing to all newborns above the cut off value in the PKU screening program:
10. Number of screening centers in your country: Total population of your country:
11. Which diseases are mandatory screened in newborns in your country (please, list all of them with the year of the introduction of the screening for each)?
12. Which laboratory methods are used for newborn screening in diseases ***other*** than PKU (please, list all)?
13. Do you plan to expand the newborn screening program in the next 5 years (if yes, when expectantly)?
14. Which additional diseases would you like to add to the existing newborn screening program (please, list all)?
15. Is the existing newborn screening program country wide or regional or hospital based (please specify)?
16. Who is currently financing the existing newborn screening program (e.g. national insurance system, or the ministry for health, or regions, or private, …)?
17. Could you estimate the current costs per screened newborn (please calculate to Euro/newborn)?
18. Which are the main obstacles in expanding the newborn screening program (please list three or more)?
19. Are you a part of any international cooperation/program in newborn screening (please specify)?
20. How urgent do you find expanding the existing newborn screening program, please give an estimate (on a 5 degree scale; from 0 - not at all, to 5 - extremely)?
II. ***NATION-WIDE ORGANIZATION OF THE PKU CARE AND ITS CHARACTERISTICS***
21. Do you have national guidelines for PKU management (if yes, please add the publication details)?
22. Which international PKU guidelines do you primarily follow?
23. How many PKU centers are in your country (please specify according to the following categories)? a. Pediatric only PKU centers: b. Adult only PKU centers: c. Combined – pediatric and adult PKU centers:
24. Do you have a national PKU registry in your country?
25. How many patients are currently included in the national PKU registry?
26. How many patients with BH4 deficiencies are currently included in the national PKU registry?
27. How do you define PKU metabolic phenotypes (write lower limit in µM of phanylalanine) and the recommendation for the treatment (until which age do you treat patients with given phenotype)? a. Classic PKU (lower Phe limit in µM; until which age treated): b. Moderate PKU (lower Phe limit in µM; until which age treated): c. Mild PKU (lower Phe limit in µM; until which age treated): d. Mild hyperphenylalaninemia (lower Phe limit in µM; until which age treated):
28. The most common background of the PKU physicians (pediatricians, internists, others - specify):
29. Who cares for the adults with PKU (pediatricians, internists, others - specify)?
30. Are amino-mixtures (PKU preparations) free of charge for the patients?
31. Any age limit for free prescription of amino-mixtures for adults?

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32. Do the patients get free low protein food; if yes please specify the amount per patient/month?
33. Do they get any extra money/financial support for the low protein food (if yes, any age limit?)?
34. Is BH4 treatment free of charge?
35. Age limit for BH4 treatment (please specify the lower and upper age limit)?
36. Does your country have a patient's/parent's PKU society?
37. Could you estimate the proportion of patient members in the society comparing to the whole population of the PKU patients?
38. Is the national PKU society a member of E.S.PKU?
III. ***ORGANIZATION OF THE PKU CARE IN YOUR CENTER AND ITS CHARACTERISTICS***
39. Number of the patient (having a PKU diet) at your center in age groups: a. <1 y: b. 1-18 y: c. >18 y:
40. Could you estimate the proportions of the patients who are lost to follow up in the same age groups (only the patients who need the diet)? a. <1 y: b. 1-18: c. >18:
41. Could you specify the number of the patients with classical PKU (Phe > 1200 µM or Phe tolerance <250 mg in early childhood), and the number of other hyperphenylalaninemias on diet in the group of the patients in your PKU center:
42. Could you estimate how many (number and/or % of all) are on diet (Phe < 800 µM) in >18 years group?
43. Number of patients with BH4 deficiencies in your center:
44. Do you routinely perform genotype analyses in PKU patients on diet?
45. Do you routinely perform genotype analyses in HPA/PKU patients not requiring diet?
46. The lab method used for genotype analyses in your center:
47. The lab method(s) for Phe analyses in your center:
48. Do you use BH4 in your PKU patients?
49. Do you routinely perform BH4 loading tests in PKU patients?
50. At what age do you perform BH4 loading tests in PKU patients?
51. The members of the PKU team in your center (number of physicians, dietitians, psychologists, nurses, others - specify (please, list all)):
52. Do dietitians prepare dietary plans for the patients?
53. How frequently are patients psychologically assessed by a clinical psychologist (at what ages)?
54. Does your PKU center treat the PKU adults?
55. Does your center take care of pregnant PKU women?
56. How many PKU pregnancies did you have till now?