5th Meeting of the International Society for Neonatal Screening

Genova, Italy

26 – 29 June 2002

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*The presenting author:*

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X **Poster presentation**

❑ **Oral Presentation at workshops** (overtype with a **X** one of the following boxes)

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|---|---|---|
| ❑ Hypothyroidism and CAH | X Hyperphenylalaninemias | ❑ Cystic Fibrosis |
| ❑ Quality control programs | ❑ New technologies | ❑ Databases and newborn screening |
| ❑ New paths in neonatal screening | ❑ Tandem MS | ❑ Screening for Type I Diabetes |

The number of oral presentations is limited. Abstracts not accepted for oral presentation will be considered for poster presentation.

***Abstracts submission deadline: 15th January 2002***

Type your abstract inside the box according to the instructions

## EPIDEMIOLOGY OF HYPERPHENYLALANINEMIA (HPA) IN LEBANON.


Touma E H [1], Romstad A[2], Khneisser I [1], Hsiao K-J[3], Loiselet J[1], Dhondt J-L [4] .

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During a 6-year period (1996-2001) about 60,000 newborns were screened using the enzymatic assay Quantase®. The cumulative incidence of HPA-PKU was 1/7500 (7 cases). Three cases were considered as mild benign HPA. In particular, the cumulative incidence of BH4-deficiency (1 case) and of classical PKU (3 cases) was 1/15000. The relative incidence of atypical PKU due to BH4 deficiencies was 25% while globally it is only 1-2%. This may be attributed to higher levels of consanguineous marriages in Lebanon.

Including the 7 cases diagnosed by neonatal screening, a total of 22 children in 19 families were followed for HPA under metabolic investigation or dietary monitoring. Of those, two infants had PKU with associated disorders: fatal intestinal dysplasia in one and cardiomyopathy in another. Only one of five infants who had mild benign persistent HPA needed special low PA diet because of blood PA levels above 360 µmol/L.

Tetrahydrobiopterin (BH4) deficiencies were determined in three infants who were investigated for HPA. The first case had GTP cyclohydrolase deficiency for which she received adequate treatment starting 5 months of age with normal growth and development at 5 years of age. DNA analysis revealed a homozygous mutation in exon 6, M213T (ATG→ ACG) not described previously. The second case, with 6-pyruvoyl-tetrahydropterin synthase (6PTPS) deficiency, was diagnosed at 8 months of age but she was not adequately treated and died at age 1.5 years. DNA analysis demonstrated a homozygous mutation C to T at cDNA position 200 within the exon 4 of the PTPS gene. The third case was detected by neonatal screening and confirmed as Dihydropteridine reductase deficiency at 1 month of age. DNA analysis revealed two distinct mutations R221X and Y150C, the first being a severe mutation inherited from the mother while the second a milder one. The patient responded well to low-PA diet and special treatment by BH4, L-Dopa+carbidopa, 5-OH Tryptophane, Selegeline and folinic acid with normal development at 2 years of age.


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