Background and Aim

An international disease registry was started in September 2009 to evaluate the long-term disease course of NP-C in clinical settings.

Methods

Descriptive data from enrolment are presented for all patients with available data who were included in the Registry as of 19th August 2011.

Results

121 patients have been enrolled. The median (range) age at enrolment was 16.9 (0.9–56.6) years, age at onset of neurological manifestations was 8.2 (1–48.0) years (n=100), and age at diagnosis was 11.8 (0.1–53.9) years (n=110). A history of neonatal jaundice was recorded in 4/4 evaluable patients with early-infantile (EI) onset of neurological manifestations (at age < 2 years; n=9), 6/21 (29%) with late-infantile (LI) onset (at 2 to < 6 years; n=21), 6/21 (29%) with juvenile (JUV) onset (at 6 to < 15 years; n=21), and 3/20 (15%) with adolescent/adult (AA) onset (at ≥ 15 years; n=20). Migrulat therapy at enrolment was recorded in 88/121 (73%) patients; mean (SD) exposure 1.7 (1.5) years (n=86). Neurological manifestations were observed in 71/84 (85%) patients: ataxia (71%), vertical gaze palsy (68%) and dysarthria (62%) were most frequent. Median (range) disability scores (0=normal; 1=worst) were: 0.0 (0.0–0.94) in EI (n=7), 0.29 (0.0–1.0) in LI (n=28), 0.41 (0.15–0.88) in JUV (n=28), and 0.29 (0.06–0.81) in AA-onset patients (n=26). A low proportion of patients had normal language, manipulation, ambulation, and/or swallowing.

Conclusions

Over two-thirds of this NP-C cohort had infantile or juvenile onset of neurological manifestations; neonatal jaundice was observed more frequently in these patients versus adolescent/adult-onset patients.

Determination of Prealbumin, Selenium, Zinc and Iron Concentration in Serum for Monitoring the Nutrition Status of Phenylketonuric and Hyperphenylalaninemic Patients

Background and Aim

Phenylketonuria is an inherited disorder of metabolism of the amino acid phenylalanine caused by a deficiency of the enzyme phenylalanine hydroxylase. It is treated with a low-protein diet containing a low content of phenylalanine to prevent mental affection of the patient. The objective of the present study was to assess the compliance of our phenylketonuric (PKU) and hyperphenylalaninemic (HPA) patients; to determine the concentration of serum pre-albumin and trace elements to discover the potential correlation between the amount of proteins in food and their metabolic control.

Methods

The prospective study contained altogether 174 patients, of which 113 were children, 60 with PKU and 53 with HPA and 61 were adults, 51 with PKU and 10 with HPA.

Results

We did not prove a statistically significant difference in the levels of serum pre-albumin, zinc and iron among the respective groups. We proved statistically significant difference in the level of serum selenium among PKU and HPA patients in adulthood (p=0.006, Mann-Whitney U test).

Conclusion

The therapeutic restrictive diet for PKU and HPA makes the patient liable to the risk of nutritional deficit.