

18th E.S.PKU Meeting

29th–31st October
2004



Bled - SLOVENIA



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Program of the Meeting

Day 1 Friday, October 29th 2004			
09:00 onwards in the morning	Arrival of the delegates and guests -checking in according to personal arrangements		
10:00 - 11:00	Press Conference		Small Conference Room "Triglavska dvorana" Hotel Kompas
11:20 - 12:10	Official Opening of the Industrial Exhibition	Board of E.S.PKU	Coffee Room Hotel Kompas
12:10 - 18:00	Industrial Exhibition		Coffee Room Hotel Kompas
12:00 - 13:30	Lunch		Restaurant Hotel Kompas
15:00 - 17:15	General Meeting of E.S.PKU	<i>Moderator:</i> Jean-Marie Criem President E.S.PKU	Conference Room "Jezerska dvorana" Hotel Kompas
17:15 - 18:00	Financial Policy for PKU-Foods in Europe Presentation of the results of the Questionnaire	<i>Presentation:</i> Natalija Stošički <i>Moderator:</i> Dr. Tanšek, Slovenia	Conference Room "Jezerska dvorana" Hotel Kompas
18:45 - 19:00	Welcome-Address of President of E.S.PKU, Mr. Jean-Marie Criem Welcome of Slovenian Representative Mr. Ciril Kržišnik, MD, PhD, Medical Center - University Children's Hospital, Medical Director		Conference Room "Jezerska dvorana" Hotel Kompas
19:00 - 21:00	Wellcome-Dinner		Restaurant Hotel Kompas
21:15 onwards	Informal meeting of presidents of PKU Societies: introduction, presentation of work, initiatives, common issues, future cooperation		Small Conference Room "Triglavska dvorana" Hotel Kompas

Day 2 Saturday, October 30th 2004			
08:00 - 09:30	Breakfast		Restaurant Hotel Kompas Hotel Golf
09:30 - 18:00	Industrial Exhibition		Coffee Room Hotel Kompas
10:00 - 10:45 10:45 - 11:20	Scientific Advisory Committee Meeting I: Micronutrients and their bioavailability in the PKU Diet Discussion on Micronutrients and their bioavailability in the PKU Diet	<i>Moderator:</i> Prof. Dr. Baerlocher, Swiss. Dr. Petra Rust, Austria	Conference Room "Jezerka dvorana" Hotel Kompas
12:00 - 13:00	Lunch		Restaurant Hotel Kompas
13:30 - 15:00	Scientific Advisory Committee Meeting II: Laboratory Tests for the follow-up of PKU and other allied diseases Evolving methods for the measurement of Phenylalanine	<i>Moderator:</i> Prof. Dr. Baerlocher Dr. Herbert Korall, Germany Dr. Zoltan Lukas, Germany	Conference Room "Jezerka dvorana" Hotel Kompas
15:00 - 15:30	Coffee-break		Coffee Room Hotel Kompas
15:45 - 17:30	Scientific Advisory Committee Meeting III The Quality of Life with PKU Compliance in PKU treatment Coping Styles and Health Outcomes	<i>Moderator:</i> Prof. Dr. Baerlocher Dr. Tanšek, Ljubljana Dr. Francjan Van Spronsen, Netherland Dr. Fiona Kennedy, Ireland	Conference Room "Jezerka dvorana" Hotel Kompas
17:30 - 17:45	Closing Remarks	Jean-Marie Criem, President of E.S.PKU	Conference Room "Jezerka dvorana" Hotel Kompas
19:00 20:30 onwards	Dinner with Social Programme - Slovenian folk dances and songs (Folk dancing group from Trbovlje, Slovenia)		Restaurant Hotel Golf
scheduled on demands of the	Informal Meeting of Scientific Advisors Preparation of the Scientific Programme of 2005 Annual Meeting	<i>Moderator / Program</i> Prof. Dr. Baerlocher	Small Conference Room "Triglavka dvorana" Hotel Kompas

participans	New Researches		
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Day 3 Sunday, October 31st 2004		
08:00 - 09:30	Breakfast	Restaurant Hotel Kompas Hotel Golf
10:00 - 12:30 10:00 - 16:00 in the morning	Guided Excursions to the Bled Island Guided Excursion to Ljubljana, Slovenian capital (depends on weather) according to personal choice / meeting point is the lobby of Hotel Kompas - registration and price of excursion could be submitted in the lobby of Hotel Kompas - please register on 30th October 2004 the latest!	
	Departure of the delegates and guests according to personal arrangements	

Lecture Abstracts

Financial Policy for PKU-Foods in Europe

Ms. Natalija Stošicki

Slovenian PKU Society, president

Presentation of the results of the Questionnaire

	PKU formula	PKU food
Slovenia	✓ Case by case not from compulsory health insurance	payable
Croatia	✓ compulsory health insurance	flour ✓ other is payable
Austria	✓ compulsory health insurance	Family support for PKU children
Germany	✓ compulsory + supplementary health insurance	payable
Denmark	(✓) Refunded in 4 days	✓ Refunded in 20 days
Sweden	Payable by patients: up to 16yrs =10 EUR/month after 16yrs: 20-150 EUR/month depends on geographical area	Payable by patients: up to 16yrs = 6 EUR/month after 16yrs: 20-150 EUR/month depends on geographical area
Poland	✓ compulsory health insurance (formula=drug)	N/A
Hungary	✓ (formula = drug)	payable
Ireland	✓ compulsory health insurance	✓ On medical prescription
Spain	✓ (only SHS and Mad Johnson)	In some regions there is help of the government

	Tax reliefs and other benefits	Receipts
Slovenia	no	€ 83/month child care allowance
Croatia	no	0
Austria	N/A	N/A
Germany	€ 3.700/year deductible from taxable income € 924/year deductible for nursing a child	€ 205/month for care of handicaped
Denmark	no	€ 150 depending on expences
Sweden	no	Up to € 150/month child care allowance
Poland	N/A	N/A
Hungary	€ 48/year personal income tax relief for adult PKU Special aid for mothers with PKU child – 10 yrs at home	€ 60/month child care allowance
Ireland	no	no
Spain	N/A	N/A

N/A – information not available

✓ – free of charge

Micronutrients and their bioavailability in the PKU-Diet

Dr. Petra Rust

Institute of Nutritional Sciences, University of Vienna, Austria

Abstract

Micronutrients are a group of dietary constituents of low quantity, but nevertheless of great importance for disease prevention. Vitamins and trace elements are the major classes of micronutrients in the diet. In various situations micronutrient deficiencies occur despite adequate dietary intake. These observations indicate that the total amounts of micronutrients comprised in foods are not available for utilization by the body. The term bioavailability refers to the extent to which a bioactive compound elicits a response in a target tissue. Bioavailability influencing factors are: efficiency of digestion, previous intake of the nutrient, body "status" of the nutrient, gut transmit time, presence of gastrointestinal disorders or disease, other products with which the food stuff is consumed and prior-treatment (namely cooking or processing) of the product.

Thus, micronutrient interactions can affect absorption and bioavailability by a number of mechanisms. Minerals with similar chemical characteristics can compete for transport proteins or other uptake mechanisms, and they can form chelates with organic substances, facilitating or hindering absorption. The consequence of these interactions strongly depends on the relative concentrations of the nutrient. For example, negative effects of iron supplementation on indices of zinc and copper status and of zinc supplementation on iron and copper status have been reported. Ascorbic acid has a strong iron absorption promoting potential and in iron deficient subjects ascorbic acid supplementation improves iron status. Therefore, ascorbic acid supplementation or increased intake of ascorbic acid rich food could have health benefits, especially in subjects subsisting on a mainly plant food based diet like in PKU patients.

Potential risks of interactions have to be considered when food fortifications or supplementations are established, especially in population groups with higher micronutrient requirements. In populations where the major risk for iron deficiency is a poor availability of dietary iron, there is also a risk of marginal zinc status especially in growing individuals. Thus iron supplementation could cause impaired zinc status.

Perceptions of these interactions, evaluation of nutrient intake of subjects with regard to absorption inhibiting and promoting constituents could lead to more effective strategies to prevent micronutrient deficiency.

Laboratory tests for the follow-up of PKU and allied diseases

Dr. Herbert Korall

**Zentrum für Stoffwechselfdiagnostik Reutlingen GmbH (Metabolic Centre)
Reutlingen, Germany, herbert.korall-zfs@gmx.de**

Abstract

Tandem mass spectrometry (TMS) is now a sophisticated, high-tech method for the rapid analysis of pathognomic metabolites.

What is tandem mass spectrometry and why to do tandem mass spectrometry?

- It is a new, precise, accurate and sensitive technology
- By using stable isotopes as internal standards it is the golden standard of analytical methods
- Only smallest sample amounts are needed
- This method enables high and effective sample throughput
- TMS replaces HPLC and enzymatic assays for therapy monitoring of PKU and other inborn errors of metabolism (IEOM)

Advanced selective screening and therapy monitoring in PKU, isovaleric academia (IVA), methylmalonic (MMA) and propionic academia (PA), tyrosinemia (TYR) and glutaric aciduria (GA) may be done by only one single 3.2 mm punch of a blood spot on a Guthrie card. The blood spot may be easily taken at home by collecting a small blood sample from the finger tip by a SoftClik. By TMS therapy monitoring in PKU may be done by parallel determination of phenylalanine and tyrosine which is an essential amino acid in PKU patients.

What are the advantages of therapy monitoring by TMS?

- Only one single blood spot is sufficient
- Simple mailing of Guthrie card in an envelope
- Rapid analysis of relevant metabolites (i.e. phenylalanine and tyrosine)
- Results are available on the same day when the sample arrives at the lab
- Comparing quantification of phenylalanine and tyrosine in plasma by HPLC respective dried blood spots by TMS proves the high accuracy of TMS

We are now able to confirm or to reject a diagnosis of PKU from blood samples within the first 24 h of life. This is interesting especially in index families where a PKU case is already known.

Other inborn errors of metabolic diseases (see above) may be integrated as well, because these diseases:

- Need a similar protein restriction
- Show similar problems of compliance
- And need similar therapy monitoring

Application of therapy monitoring of other diseases from dried blood spots are shown. Also homocystinuria (a disease which is on dietary treatment by our new executive board member Fiona Kennedy) may be monitored by TMS in only 100 µl plasma by a runtime of less than one minute.

Conclusions: Tandem mass spectrometry is a highly efficient method in therapy monitoring of PKU and other allied diseases.

Evolving Methods for the Measurement of Phenylalanine –

A Vital Diagnostic Marker and Indicator for Follow-up in Cases of Hyperphenylalaninemia and Tetrahydrobiopterin-deficiency

Dr. Zoltan Lukacs

**University Hospital Hamburg-Eppendorf, Department of Pediatrics, Metabolic Laboratory,
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Abstract

Since the discovery of the biochemical backgrounds to hyperphenylalaninemia (HPA), the rapid determination of phenylalanine concentrations has been crucial for screening of potentially affected individuals and also for an effective treatment of the disease. Early on the original bacterial Guthrie test provided limited sensitivity but proved to be a useful tool for preventive screening of newborns. Numerous new methods which provide higher sensitivity and accuracy have been developed since. Among these, enzyme assays which use UV or fluorescence spectroscopy are widely used. However, those are slowly being replaced by tandem-mass spectrometry. For scientific purposes, breath analysis and MR spectroscopy have been applied to assess time-dependent phenylalanine metabolism and the kinetics of blood-brain transport *in vivo*. Furthermore, the tetrahydrobiopterin (BH4) loading test is particularly important for distinguishing between HPA and tetrahydrobiopterin-deficiency. More recently, BH4-sensitive HPA may be diagnosed with this assay when it is administered under highly reproducible conditions and thus, may open new possibilities for treatment of certain cases of BH4-sensitive HPA.

The Quality of Life with Phenylketonuria

Dr. Mojca Žerjav Tanšek

Medical Center - University Children's Hospital, Vrazov trg 1, Ljubljana, Slovenia

Introduction

Although there is some knowledge about psychological adjustment of children and adolescents with phenylketonuria (PKU), this is not true for health-related quality of life. The studies on behavior in early treated PKU children are consistent with the observations that as a group, are at greater risk of behavioral and psychological disturbance than their peers. They may have a higher risk of psychosocial maladjustment. Internalizing disorders like depressive mood appear to be more common, whereas externalizing symptoms are not. Some studies show that even early treated PKU adolescents are characterized by less autonomy, less achievement motivation, low frustration tolerance, more negative self description and a higher level of dependency from their families. Nevertheless several studies have shown that the discontinuation of diet led to an irreversible decline in IQ even in adolescence.

The data support the assumption that the strictness and flexibility of dietary practice have a profound effect on children's personality development. The characteristic behavioral and psychological disturbances may be regarded as a consequence of an insufficient coping with the diet and the chronic illness in general as well as a direct result of the metabolic disorder. At the other side a recent study indicates that PKU with a strict diet and the related medical controls do not significantly compromise health related quality of life and psychological adjustment (Markus, 2002). The present study was planned to evaluate the quality of life in PKU adolescents in Slovenia.

Materials and Methods

In the study quality of life (QOL) questionnaires of 15 adolescents with classical PKU (mean age 15.2 years (from 13 to 18 years of age)), 19 adolescents with insulin dependant diabetes mellitus (mean age 15.3 (from 12 to 18 years of age)) and 19 healthy controls (mean age 16.3 (from 11 to 18 years of age)) were investigated. All early treated PKU patients were on diet but the average phenylalanine value was not included in the questionnaire because the interview was anonymous. The adolescents are controlling their phenylalanine values 3 to 4 times per year.

All diabetic children (DM group) were treated with 3 to 4 daily injections of insulin and they follow the recommended diet in diabetes. The control group consists of the healthy children of the same age who were tested in different schools.

All three groups completed the QOL Questionnaire which was kindly provided by NEDEC as a collaborative project of different diabetes centers (an adaptation of QOL questionnaire Eli Lilly Company, USA).

The questionnaire consists of 126 questions which are arranged in different groups. The questions try to evaluate patient's perception about the general health and well-being, ability to perform various physical, school and social activities, flexibility in planning of meals, the common body complaints and emotional status.

The answers are descriptive and they represent the grades from 1 to 5 (from completely negative to completely positive answer). The results of all three groups (PKU, DM and controls) are compared.

Results

PKU and DM patients find their general health less good than the healthy children who feel excellent/very good in 63%. Only 46% adolescents with PKU (37% with DM) find their health excellent or very good.

PKU patients do not feel limited in moderate and daily activities but they feel less able to perform vigorous activities in 46% (DM group only in 26% and control group in 5%). It is interesting that the emotional causes are responsible for the difficulties in daily work in control group (in 48%) but only 10% DM patients and none from PKU group stated the emotional reasons. The physical causes for the difficulties in daily work were mentioned in 20% in PKU and DM group and in 15% in the control group, respectively. The others have listed the combination of emotional and physical causes.

All PKU patients stated that they had a lot of energy in the last 4 weeks before the interview but the statement was true just for 73% of adolescents in DM and 57% in control group. All groups have described a similar courage in resolving their health problems.

There were no important differences in feelings of happiness and sadness during the last weeks between the groups. It is very interesting that PKU and DM adolescents are very satisfied with their sleep (in 80% and 63%) comparing with the control group (only in 33%)...

The difference exists also for the time they spend exercising: PKU and DM patients are satisfied in 94% and 78% respectively but controls only in 47%. All three groups are comparatively equally satisfied with their friends and the leisure time.

47% of the PKU and 36% of the DM patients are embarrassed to deal with their disease in public but none from the control group has confirmed embarrassment in a case of health problems. PKU patients are keener to keep telling others about having disease (in 86%) but DM adolescent do it only in 52%.

PKU adolescents feel less restricted in the meal plan as DM children. From DM group 21% of patients feel often restricted but none from PKU group. On contrary PKU group is less satisfied with the kinds and amounts of food. They find the choice of food satisfied only in 20% comparing to 47% in DM and 73% in control group.

80% PKU adolescents deny that disease interrupts their spare time activities but DM patients admit in 69% that the spare time is disrupted because of the diabetes.

It is surprising that 52% of DM and 35% of PKU patients would rather break the diet than tell someone about the disease.

DM adolescents are far more worried about the medical insurance, employment, vacations and the complications from the disease in the future (in 68 to 90%) as PKU (in 26 to 40%) and control group (in 10 to 36%). PKU children do not find teachers treat them differently because of the disease but DM children have answered it positively in 57%.

On the scale from 1 to 10 (not harmful to very harmful) PKU group finds phenylketonuria harmful to their health with score 2.6. On the same scale DM adolescents have a score 4.5 and controls score 2. On a similar scale (not sure to very sure) PKU group expects a good control of the disease/health with score 8.2, DM group with score 7.2 and control group with score 7.

Conclusions

The results of QOL questionnaires have revealed that PKU adolescents feel limited in daily life because of the disease. PKU group shows positive life attitude. Their QOL is not compromised comparing to the control group. The exceptions are the food restrictions with the social dimension of eating and the worries about the personal life in the future.

The group of diabetic adolescents presents a comparison to PKU adolescents as a group of chronically ill children. The daily preoccupation with the diabetes and the expected apparent complications from the disease make this group more vulnerable. According to the results of our study their QOL is more compromised as in PKU group.

Compliance in PKU treatment: a complex issue.

Dr. FJ van Spronsen

Department of Metabolic Diseases, Beatrix Children's Hospital, Netherland

Abstract

When compliance in PKU patients is concerned a number of issues should be taken into account.

1. Nowadays, thanks to the early diagnosis and possibility of treatment PKU patients develop normally, whereas in the old days PKU individuals, left untreated, developed severe mental retardation.
2. Especially during the first years, parents of individuals with PKU usually do realise that treatment should be very strict. With time, fortunately, parents become more familiar and relaxed with the dietary treatment.
3. When dietary treatment is not as strict as advised in the literature, the individual patient will develop a degree of less optimal development that only may be detected on a population basis.
4. Notwithstanding the possibilities in the future with other treatment strategies (BH4, Large Neutral Amino Acid supplementation, phenylalanine lyase, gene therapy), at present dietary treatment is the treatment of choice and dietary treatment is burdensome because of various reasons.
5. When a child grows the diet becomes often more complicate to control rather than easier
6. Plasma phenylalanine concentrations increase with age that can not only be based on a decrease in tolerance, implying that compliance decreases with age.

But the central question is: how to define good and poor compliance? To define good and poor compliance we first have to decide our aims. Is good compliance just a "low" plasma phenylalanine concentration or a combination with good outcome measurements?

We consider good quality of treatment with "low" plasma phenylalanine concentrations important but at the same time we do not want to make patients well-developed-unhappy-strict diet-users. When we aim at a normal quality of life, we may have to change our aims!

At present we do not know what the effect is of very strict and less strict treatment on psychosoical developement, well-being of the patients, quality of life etc. So, we do not know anything about the balance between good neuropsychological development and good psychosocial development.

Therefore, one of the important questions is: Do people with PKU who are treated in a very strict way have a higher or a lower quality of life than people with PKU who are treated less strict? When we know this we might be able to define our aims and treatment strageties better.

We then may try to increase compliance by different strategies, including f.e.:

1. increase the level of knowledge of physicians (both pediatricians and adult physicians) and dieticians (both for children and adults) treating patients with PKU
2. increase the level of knowledge of patients and parents of patients with PKU
3. decrease the burdensomeness of the dietary treatment by searching for the do's and the don'ts in dietary treatment (f.e. should the individual phenylalanine daily intake be given as 3-4 equal amounts a day or can we give 50% in one meal? Should we divide the daily intake of

protein substitutes: may this be taken in one or two times a day or should this be divided into 3-4 daily equal parts?)

4. transfer the responsibility of the diet to the parents/patients to a large degree.
5. develop methods that can easen the stress of treatment in daily life, including f.e. a protein substitute with minerals, vitamins etc that smells and tastes well, developing another structure of the outpatient control with age related topics.
6. Investigate the usefullness of estimating instead of weighing the precise phenylalanine intake
7. Etc., etc.

During the presentation I will try to go into these issues a little further, showing that there is still a lot to do and that some studies can only be performed on a multi-centre-basis, while for other issues all centres/countries should be wellcomed to show their questions, problems, and searches for (practical) solutions.

A study on development and quality of life can be performed on a European level. On the one hand the research design is quite simple and asks “only” for data on outcome and quality of life instruments. These instruments are available on a European level with references for most countries. So, this asks for a multi-centre/coutry research basis that needs the involvement of some dedicated people from various countries/centres in which the ESPKU may play an important role.

Coping Styles and Health Outcomes

An Evaluation of the Coping–Health Relation in Cohort of Irish Adolescents, on Dietary Treatment for Classical Phenylketonuria

Fiona Kennedy, B.A. Hons. MPhil. Hons.

Acknowledgements: Dr. E. Naughten, Consultant Paediatrician with special interests in inherited metabolic disorders, Ms. H. Rushe, Senior Clinical Psychologist, University Hospital Temple Street, Dublin, Ireland, Dr. E. Quayle & Ms. A. Moloney, Lecturers, Dept. of Applied Psychology, University College Cork, Ireland

Abstract

With the advent of advanced diagnostic techniques, comprehensive immunisation programmes and effective therapeutic interventions, contemporary health care has seen a radical demise in the prevalence of acute, infectious disease. Consequently, predominant emphasis now rests with the sustained management of chronic, physical disorders. Amenable to symptomatic management rather than cure, such disorders are by virtue of definition of a protracted nature. Indeed, the parameters involved typically dictate that the condition last for a duration of six months or more. With treatment being largely directed at hindering the course of these conditions, and indeed preventing the premature demise of those afflicted, the PKU condition represents in many ways, an exemplary case of that which constitutes a chronic physical disorder. A complex inherited metabolic disorder, diagnosed by means of the Guthrie test, phenylketonuria (PKU), is perhaps the most prevalent of all inherited amino-acid disorders. Caused by a hereditary deficiency of the liver enzyme phenylalanine hydroxylase, associated disturbances in the metabolic chain, if untreated, can lead to profound consequences of a physiological, neurological, cognitive and behavioural nature. Thankfully however, early pre-symptomatic detection coupled with effective therapeutic interventions, mean that for the patient with classical phenylketonuria, the future now brings with it the hope of a relatively normal life span. Yet this is not to say that the PKU condition no longer impacts on those afflicted. To the contrary, the PKU condition like any chronic disorder has the potential to evolve and impact across all facets of the life cycle including; school, work, travel and dating. Frequently dictating that social demands be refined so as to accommodate treatment demands, for many patient's the quest to cope can be a formidable and at times, somewhat daunting task. Thankfully however, despite these difficulties the growing number of PKU patients' leading healthy fulfilled lives, would appear to provide testimony to the assertion that many patients' can and indeed do cope. Yet how do these patients' cope and in what way, if indeed it does, does the individual's choice of coping style impact on his/her subsequent health outcome? Eager to address this latter issue, this current presentation focuses on the coping-health relation in a cohort of Irish adolescents (N=50) on dietary treatment for classical phenylketonuria. Believing it time to move beyond traditional deficit-centred models of care this study sought therefore to embrace for the most part, an alternative, competency based model of practice. Consequently, employing the A-Cope Scale, attempts were made to evaluate not whether patients cope with life's demands but rather instead, how it is that they actually do cope. Employing second order factor analysis on the A-Cope Scale, results ascertained served to identify the use of two distinct coping styles, the first encompassing those coping strategies of an interpersonal nature [e.g. seeking support, investing in friendships, solving family problems] the second incorporating strategies largely of a personal nature [e.g. engaging in demanding activities, avoiding problems, developing a sense of self-reliance]. Following a series of multiple regression analyses; use of interpersonal type coping

was identified as the most significant predictor of metabolic control, only however in those patients already in good biochemical control. Such findings were ascertained having forced entered the identified coping strategies into a multiple regression analysis wherein a host of demographic [age, gender, number of siblings on dietary treatment] dispositional [self-esteem, self efficacy] illness related [knowledge of the PKU condition, acceptance of and attitude to, degree of responsibility assumed for treatment management] and multi-dimensional measures of health control orientation, [internal, chance, powerful others] were stepped through. Amongst patients deemed to be in poor metabolic control, use of a great array of more diverse coping strategies were noted. Here the use of diversion and escapism was deemed to be best predictive of improved biochemical control, over and above those other extraneous variables employed.

Official Sponsors

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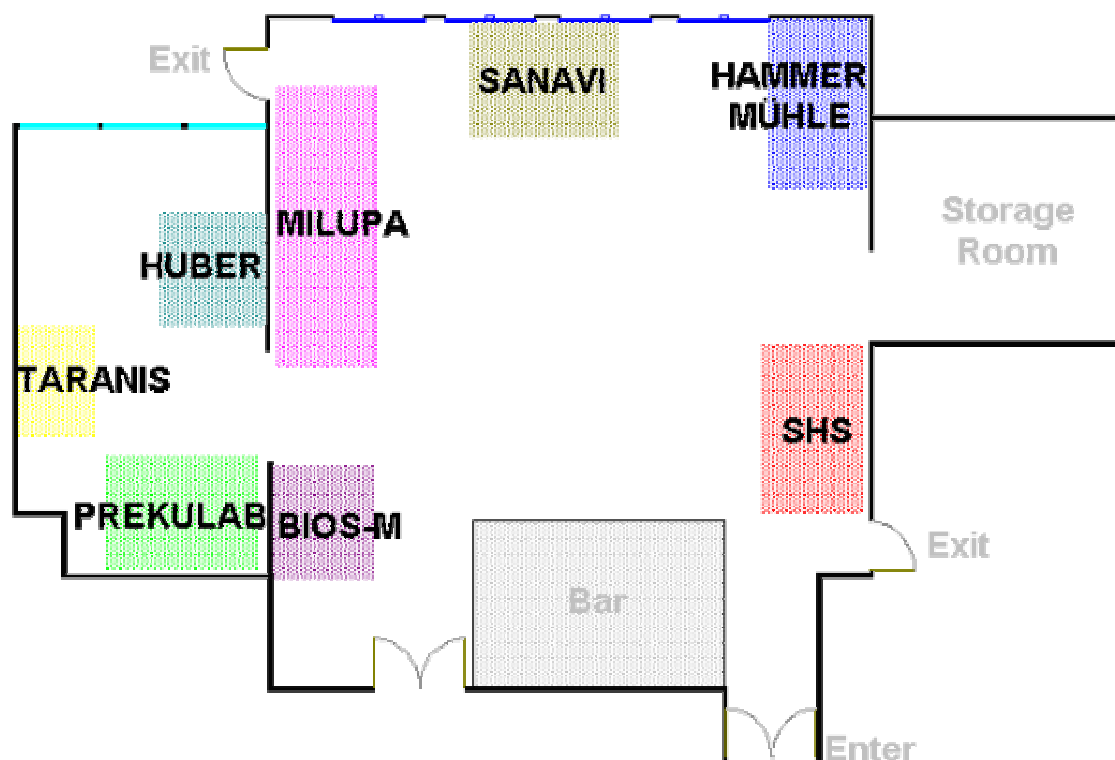
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Exhibition Ground Plan



Introduction of Slovenian PKU Society

In Slovenia, first efforts to start neonatal screening for phenylketonuria (PKU) were made in 1967. The screening has run successfully since 1979. In July 1992, the quantitative fluorometric method was introduced. Results of the 13-year period of Guthrie method application are compared to results of the present period. Additionally, appropriate treatment, therapy duration and maternal PKU program are discussed. Methods. In all newborns with elevated phenylalanine (PHE) levels detected by neonatal screening performed with a dried blood drop collected from the newborn's heel on a filter paper on days 3 to 5 (cut off level 0.12 mmol/l), serum phenylalanine determination was done. Patients were categorized according to the severity of the PHE elevation. Incidences were calculated and compared by chi-square test. The incidence of classical PKU is 1/8.000. The incidence of mild hyperphenylalaninemia (PHE 0.12-0.59 mmol/l) in the last period when the quantitative fluorometric screening method was used, is 1/476, which is significantly ($p < 0.0001$) higher than before. One patient with 6-pyruvoyltetrahydrobiopterin synthetase (6-PTS) deficiency was diagnosed. All patients with phenylketonuria are registered at Ljubljana Medical Centre, Department of Paediatrics, Department of Endocrinology, Diabetes and Metabolic Diseases.

PKU association of Slovenia was established in 1983. It counts 120 members in the whole of Slovenia.

Each year members come together at the PKU family meeting where experiences are exchanged and new members are introduced. Owing to the smallness of association and the country itself, all families know each other.

Each summer 14 days long free of charge holidays on the Slovenian coast are organised for children in cooperation with PKU association of Slovenia and Ljubljana Medical Centre, Department of Paediatrics.

During Christmas holidays children are visited by Santa Claus.

One of the main principles of PKU association is to assure good quality of life to their members. For this reason the publications stated below were issued:

1997

- "Phenylketonuria and nutrition"

2000

- "To live with Phenylketonuria"

- Translation of Czech PKU cookery book,

- Brochure: "Guidebook through PKU" for educators in kindergartens and school teachers,

- Brochure: "Have you heard for Phenylketonuria?",

2004

- »P planning and attending PKU women's pregnancy«

Planning and proper attention during pregnancy is significant for the state of health of PKU mothers-to be and their newborns.

Under the guidance of dr. Mojca Žerjav Tanšek (Ljubljana Medical Centre, Department of Paediatrics), who controls all PKU patients in Slovenia, PKU association of Slovenia has started to perform an educational programme for PKU women about planning and attending during pregnancy under medical supervision. The programme started in 2003 and is organised in the mode of one day meetings at the Slovenian countryside.

Important regular tasks of Slovenian PKU association are also:

- testing the presence of phenylalanine in low albumin products of domestic food manufacturers at the national Chemical Institute,
- active monitoring of tax, health, etc. legislation and participation in its modifications,
- free of charge distribution of PKU food to the families with low budget.

In the future PKU association of Slovenia would also like to include all people with un-treated PKU and encourage PKU teenagers to be independent in taking care of their own health.

PKU association of Slovenia is registered as self-supporting humanitarian association and is a member of E.S. PKU. Their activities are financed with the means of “Foundation of disabled and humanitarian organisations” and with the membership fees. They do not have regular sponsors.

Ms. Natalija Stošicki

The president

Slovenia in Brief

(<http://www.uvi.si/eng/slovenia/in-brief>)



The Republic of Slovenia lies at the heart of Europe where the Alps and the Mediterranean meet the Pannonian plains and the mysterious Karst. Slovenia has a population of 2 million and its capital city is Ljubljana. The official language is Slovene, one of the South Slavonic languages.



In Slovenia, the sun shines approximately 2000 hours per year. And yes, there is plenty of snow in winter.

As a small, beautiful and picturesque country, Slovenia makes a great tourist destination. Mountains, lakes, waterfalls, forests, caves, hills, plains, rivers and the sea - you name it, you can find it all within a modest 20.273 km² (half the size of Switzerland), as well as many natural and landscape parks. You can ski in the morning and surrender yourself to the luxury of the Adriatic Sea in the afternoon.

Slovenia has 46.6 km of sea coast - an inch per inhabitant.

The highest mountain is called Triglav - the name meaning "three-heads" - and it is 2864m high. The mountain is a true national symbol, featured on the national coat of arms and the flag.

Slovenia is also known for its great wines and delicious traditional food.

The currency is called the Slovenian Tolar (SIT).

It is very easy to get here as the country is connected with the rest of the world by a modern highway network, railway system, international airport and ports.

