During the years, a self-criticism of our screening program was necessary to orientate it towards the detection of disorders that can cause severe clinical problems, such as urea cycle disorders, methylmalonic acidemia, prolidase deficiency, hyperglycinemia and more ambivalent disorders such as sarcosinemia. The total incidence of these disorders is 1:28,000, whereas the total incidence of disorders necessitating surveillance and follow-up is 1:3,800 for transport disorders (Hartnup syndrome, cystinuria, dicarboxylic aminoaciduria, etc...) and 1:7,000 for enzymatic disorders (histidinemia, cystathioninuria, etc...). We conclude that such screening programs are technically feasible, inexpensive and furnish a valuable clinical service.

0402 FIRST OBSERVATION OF HEMOGLOBIN D PUNJAB IN AN AUSTRIAN FAMILY

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In the course of a routine screening program for gestational diabetes, the glycosylated portion of hemoglobin A (Hb A_{1c}) is quantitated by High Performance Liquid Chromatography. Incidentely an aberrant hemoglobin fraction was found in a native Austrian woman by means of this screening program. Subsequent further analysis of this hemoglobin variant revealed an heterozygote HbD Punjab state.
In an extensive field study 49 members of this family, covering 4 generations were examined. 22 of them showed the same heterozygote HbD state as the propositus.
Comparing various hematologic parameters in heterozygote carriers and normal subjects of this family, evidence was obtained that heterozygote carriers of the HbD Punjab gene are not at all impaired by this genetic variant. Considering age distribution and reproduction rate among carriers and normal subjects in this family, the heterozygote carrier state does not seem to have any negative influence on life quality and life span also.
Some hypotheses on the ways of distribution of the HbD Punjab gene from India to this remoted rural area in Austria are presented.

0403 THE INVESTIGATION OF PORPHYRIN METABOLISM IN DERMATOLOGY PATIENTS.

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Patients seen by dermatologists are referred to clinical biochemistry if their skin symptoms are suggestive of porphyria. These include bullae, blistering, burning, skin fragility and other clinical signs suggestive of photosensitivity.

Total red cell porphyrins are measured and ethanol extracts of samples with elevated levels subjected to fluorimetry. This distinguishes zinc protoporphyrin (ZnPP) from free acid protoporphyrin (PP). Elevated levels due to iron deficiency, lead exposure, chronic infection, inflammation or malignancy (ZnPP) are thereby distinguished from those due to erythropoietic protoporphyria (PP).

Total plasma porphyrins are measured and diluted plasma scanned spectrofluorimetrically using an instrument equipped with a red-sensitive photomultiplier tube. An elevated plasma porphyrin result together with a distinctive fluorimetric scan allows provisional diagnosis of either porphyria cutanea tarda (PCT), variegate porphyria (VP) or erythropoietic protoporphyria (EPP). Diagnosis is confirmed by measuring total porphyrins in urine and faeces followed by separation of the component porphyrins into diagnostically distinctive patterns using high pressure liquid chromatography (HPLC).

Investigation of 258 patients showed the following: 182 (71%) normal porphyrin metabolism, 64 (25%) PCT, 10 (4%) EPP, 2 (0.8%) VP.

Thus the main types of porphyria presenting with skin symptoms can be provisionally diagnosed within hours of specimen collection. The use of HPLC allows a definitive diagnosis to be made.

0404 PHENYLKETONURIA IN MENTALLY RETARDED CHINESE CHILDREN IN TAIWAN.

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It was believed that phenylketonuria (PKU) is a rare inborn metabolic disease in Chinese population and very few cases have been reported in the past. The blood of 4744 children were collected on filter paper from total 4994 registered students in public school mentally retarded classes all over Taiwan in the Spring of 1983 in order to study the morbidity of PKU in mentally retarded Chinese children. The blood phenylalanine (Phe) were screened by the Guthrie's becteria inhibition assay. Three of them were positive (Phe 14-20 mg/dl) and were recalled. Two cases of PKU (male, 16 yr. & female, 11 yr.) and one case of hyperphenylalaninemia (male, 13 yr.) were confirmed by positive urinary $FeCl_3$ test, serum Phe (fluorometric method) of 26.7, 33.1, 13.6 mg/dl, and Phe/Tyrosine ratio (amino acid analyzer) of 27.0, 51.0, 13.4, respectively. Two elder sisters, one of each PKU patient, were also diagnosed as PKU with mental retardation in the follow-up family study. Using the same screening and confirmatory methods, a 12 years old PKU boy was detected from 551 institutionized children in northern Taiwan. A 20 month old boy and an 8 year old girl were identified as PKU from 147 selected pediatric neuropsychiatrical patients in our hospital within a year (Nov. 1982-Oct. 1983). From these data, the morbidity of PKU in mentally retarded Chinese children may be estimated at around 0.04-0.2% and may be as high as 1.4% in clinically selected pediatric patients. These results show that PKU does cause mental retardation in Chinese children on this island and may not be as rare as we thought. Therefore, a neonatal screening study is indicated both for estimating the prevalence of PKU in Chinese and for early diagnosis and treatment to prevent PKU babies from mental retardation in Taiwan.

0405 METABOLIC SCREENING IN IRELAND

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The Irish National Neonatal Screening Unit is housed at the Children's Hospital Temple Street. Blood drawn by heelprick is despatched to the National Neonatal Screening Centre where microbiological inhibition assays are carried out for the detection of PKU, MSUD, Galactosaemia and Homocystinuria. TSH in dried blood spots is assayed by radioimmunoassay. Incidences range from 1:4400 for CHT to 1:140,000 for MSUD.

Confirmatory and monitoring procedures were installed to complement the National Neonatal Screening Programme. Further spot tests and screening profiles were added in 1972. This group of tests are applied in the detection of inherited metabolic diseases in
(a) Severely ill children
(b) Older children in whom an inherited metabolic disease is suspected
(c) Offspring of women with recurrent miscarriages and neonatal deaths, and
(d) women whose offspring have congenital malformations

These methods are limited to a range of spot tests and screening profiles including sugar chromatography, amino acid chromatography, glycosaminoglycan quantitation and electrophoresis. Routinely included is serum amino acid chromatography, estimation of blood galactose, galactose -1- phosphate uridyl transferase screening with the Beutler method, blood ammonia, lactate and pyruvate.

Organic acid screening has not yet been included in our programme. Since 1972, 3,800 blood and 8,300 urines have been analysed.

In 69 patients an inherited metabolic disease had been identified.

0406 OROTIC ACID IN URINE. REFERENCE VALUES AND DIAGNOSTIC USEFULNESS.

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Orotic acid is an intermediary metabolite of pyrimidine synthesis. Increased concentration of urinary orotic acid excretion has been reported in primary orotic aciduria and in some congenital defects of the urea cycle enzymes. Since the urinary excretion doesn't increase in organic acidurias, its determination is a tool for differentiating the hyperammoniemic disorders.

We give the reference values of orotic acid found in the 24 h. urine of 500 children aged from 0 to 15 years. We don't found differences between sexes.

Orotic acid in urine was performed according to Adachi(*) et al. and values have been related to creatinine excretion: µmol/g. creat..

We have related the urinary orotic acid values with the blood ammonia in patients suffering from ornithine trascarbamylase deficiency (1 case), hyperammoniemia, homocitrullinuria and hyperornithinemia (3 cases), organic acidurias (glutaric aciduria 1 case, methylmalonic acidemia 1 case, lactic acidemias 5 cases) and those taking valproic acid as antiepileptic therapy.

(*) Adachi T. et al.: A colorimetric determination of orotic acid. J. Vitam. 9, 217, 1963.