

A Report on the Incidence of Phenylketonuria (PKU) in Teheran/IRAN

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SUMMARY

To find the incidence of Phenylketonuria (PKU) in Teheran a study was conducted in different hospitals of Teheran for a period of Six Years (1974-1980) by screening 8633 neonates with Guthrie-test (4). Among these neonates, seven had minor hyperphenylalaninemia (5 with 6 mg% and 2 with 8 mg%). There was only one case with definite hyperphenylalaninemia (more than 20 mg%).

Material and Methods:

To study the incidence of PKU in Teheran/IRAN, 8633 Neonates were screened by Guthrie-test (4). All the neonates were 4 to 8 days old and came from different hospitals of Teheran.

Results and Discussion:

As seen in Tab. 1 from 8633 infants who were tested for serum Phenylalanine levels: 7392 had serum Phenylalanine levels less than 2 mg%, 978 2 mg%, 201 between

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2-4 mg% and finally 52 infants 4mg%. So if we consider the normal value of Phenylalanine to be 2-4 mg%, 99,89% of the cases had normal values, and the remaining 10 cases (0,11%) had the following results: 2 cases 4-6 mg% and in five cases the concentration of phenylalanine in blood was 6 mg%, and in two case 8 mg%, only in one case more than 20 mg%. With repeated determinations the first seven cases with minor hyperphenylalaninemia (6-8mg%)

showed normal values the reason of this transient hyperphenylalaninemia could be due to temporary phenylalanine hydroxylase deficiency. Only one of these infants with serum phenylalanine concentration more than 20 mg% after repeated examination was diagnosed as a definite case of PKU. It is worthwhile to mention that 3 babies of that 7 with minor hyperphenylalaninemia (6-8 mg%) were born with birth weights less than 2300 gms.

A number of studies conducted on the northern European populations show that the frequency ranges from 3-10 per 100.000 (Table 2). Such frequency has also been found in Japan and the middle East, but the disease is very uncommon in the eastern European jews and negross. PKU screening in Scotland shows an incidence of one in 8000 (11) Screening of 525240 newborns from eight European countries (Belgium, Denmark, Germany, France, England, Ireland, Holland and Switzerland) showed 668 positive cases, a frequency of 1: 8000 (1,2,3,5). The same study showed differences in PKU frequency in different countries: e.g. 1:5000 in Ireland, 1: 6000 in Germany and 1: 17000 in Switzerland (8). among the Japanese the disease is as common as the Americans and Europeans (6). For the cause of the high incidence of PKU in Ireland and West Scotland, there is a significant reduction of abortion

rate. In PKU families in comparison to the controls(14, 15).

A Screening programme for PKU undertaken in Denmark, shows a greater majority of cases in West of the country explained by . differences in ethnic background of eastern and western Denmark populations(13).

Analysis of the geographical distribution of the birth places of PKU patients, in Germany gives remarkably different frequencies, 1: 6275 in Cottbus, Leipzig, Halle and Frankfurt, but in Karlmarx stadt and Neubrandenburg the frequency is 1:14769(6).

Comparison of PKU incidence in eastern region of Austria with certain slavic and Hungarian immigrants, shows significantly more frequency than the western region with originally pure German people (12).

Study of 1000 individuals from Thailand for their fasting serum phenylalanine and tyrosin, gives a gene frequency of PKU about 0,0025 (7).

In screening of Australian newborns a PKU incidence of one out of 11608 was found (9).

The highest frequency of PKU seems to be in Belgium (1:4000) and lowest incidence in Sweden (1:21000). Among the Iranian population in our study only one Case of PKU was found in 8633 infants born in Teheran hospitals.

Tab.1 Distribution of Phenylalanine levels in serum of newborn

Serum phenylalanine levels (mg%)	Number of cases	Percentage %
< 2	7392	85.63
2	978	11.33
2-4	201	2.33
4	52	0.60
4-6	2	0.02
6	5	0.06
8	2	0.02
8-12	-	-
12-20	-	-
> 20	1	0.01
Total	8633	100.000
Temporary hyperphenylalaninemia	7	0.08
Phenylketonuria	1	0.01

Tab.2 Frequency distribution of PKU in the World
(Novitzky 1977)

Origin	Number Screened	Detected Cases	Frequency (1/?)
United States	2238634	205	11000
Poland	130912	21	6000
Germany	67309	10	7000
Canada	65898	6	11000
Israel	65000	5	13000
Ireland	25000	4	6000
Yugoslavia	23690	3	8000
Sweden	21505	1	21000
Belgium	20000	5	4000
Scotland	16500	2	8000
Denmark	12500	-	-
New Zealand	1840	-	-
Wales	1536	-	-
Australia	479	-	-
Teheran/IRAN (present Survey)	8633	1	8600

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