Evaluation of Phenylketonuria program in the Gaza Strip

By
Nabegha K. Abushahla

Supervisor

Dr. Yehia Abed

Adviser
Dr.A. Abushahla

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Supervisor:	**Tittle**
Dr.Yehia Abed	Dean of School of Public Health At-Quds University
	Signature Date Jan 18 2000
Adviser:	
Dr. A.Abushala	Head Section El Azhar University
	Signature Date Jan. 18th/2000
External Examiner:	President of Islamic University
Dr. M. Shubair	Signature Date Jan 18/2000

ABSTRACT

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Phenylketonuria is a rare inherited metabolic disorder, it affects the way by which the body processes protein. It is a genetic defect in phenylalanine ($C_9 H_{11} NO_2$) metabolism resulting in mental retardation (West Es1964), the disease is characterized by elevated blood levels of phenylalanine, which is an essential amino acid, and excessive excretion of phenylpyruvic acid. This condition is presumably caused by a deficiency in the liver enzyme, phenylalanine hydroxylase, which converts phenylalanine to tyrosine, (Hsia DYY1964). With a very strict initiation of dietary correction and prevention of severe mental retardation, affected children can enjoy a normal development and life span.

Treatment of patients with PKU requires frequent determination of blood phenylalanine and tyrosine levels, to assess dietary compliance and to evaluate the changes in the dietary intake of phenylalanine (Guthrie 1963).

An evaluation study for the phenylketonuria program in the Gaza Strip was conducted to highlight dangerous consequences of the disease, and to stimulate the policy maker to have the right decisions, to reach a maximum screening coverage.

The coverage of the PKU testing in the Gaza Strip was found to be 35.3% of the total of newborn babies. It covers only the registered newborn

babies in the governmental clinics. The rest of the newborn babies in Gaza are not tested unless they are at a high risk and have a family history.

The study revealed significant differences in blood sampling between different health centers in the Gaza Strip, the sampling accuracy in Rafah clinic was 100%, in Der Elbalah clinic it was 40%. There were other differences with respect to the card dryness and the period requires for transferring and testing the sample, (P value = 0.005).

The study showed that PKU tests were made for 61% of the babies in the second week of the baby age, ranging between 11 – 17 days of life. While the American Academy of Pediatrics (1982) recommended that the optimum age for screening of full term healthy newborn babies is between 48 – 72 hours after birth and All Saint Healthcare system (1999) stated that the test should take place before the baby leave the hospital.

The study also defined the exact material and cost needed for the PKU tests, to help the decision-makers to evaluate the cost and benefit when taking decisions for a maximum coverage program.

Questionnaires were completed for 65 PKU cases. Describing the demographic distribution, prevalence of the disease in the different provinces and localities in the Gaza Strip, the level of education of the patients, the number of PKU patients in the family, and the frequency of the PKU patients since 1982 till 1998 according to birth year & complication appearance.

The prevalence was very high in the south (8.9/100,000) compared to the Mid-Zone (4.8/100,000). The highest prevalence was in the rural villages

Chapter I

Introduction

1.1 Overview

Phenylketonuria (PKU) is an inherited metabolic disorder that results in progressive mental retardation, avoidance of brain damage and mental retardation can be prevented if the diagnosis was made early.

Asbjrn F`lling 1934 fortunately discovered the PKU disease in Norway and in the year 1960 Robert Guthrie develop the Guthrie inhibition test. Since then the early diagnosis of the disease allowed young people to grow up normally and live full productive life.

PKU is characterized by the deficiency of the enzyme phenylalanine hydroxylase, which convert excess phenylalanine to tyrosine. Phenylalanine is an essential amino acid necessary for growth. However, any excess must be depreciated by conversion to tyrosine. Any infant with PKU lacks the ability to make this necessary conversion to tyrosine. Thus phenylalanine accumulates in the blood stream causing many complications.

Routine screening of newborn infants for PKU is now mandatory in most countries of the world. The Guthrie blood test is normally done before discharging the newborn from the hospital. It is important to note that this test is not valid until the newborn has taken an ample amount (for 2 or 3 days) of the amino acid phenylalanine, which is a constituent of both, human and cows milk.

If the tests are positive, dietary control must begin immediately to prevent brain damage. Substituting normal milk by Lofenalac milk and adding strained foods low in protein to the infant's diet, vegetable and fruits are allowed in the diet, as they are low in protein.

Treatment must be initiated during the first few days of life to avoid mental retardation and to prevent the accumulation of phenylalanine in the body; blood and urine testing are made regularly to make the dietary monitoring.

Newborn screening for PKU has been conducted in the Gaza Strip in the governmental clinics since 1995, while the UNRWA clinics do not conduct the test and this leads to a low over all percentage of coverage.

Follow up of the positive cases takes place in specialized centers in the Shikh Radwan clinic, where the patients receive the health services, diet instruction and Lofenelac milk.

1.2 Problem statement

Newborn babies who receive vaccination and health services in the governmental clinics, in Gaza Strip are the only ones who are being tested, as tests are made in the central laboratory of Ministry of Health. About 66% of the newborn babies receiving their vaccination and health service in UNRWA and are not being tested for PKU.

Any estimation about the prevalence and incidence of the disease in the Gaza Strip is still unclear due to lack of research on the subject.

Low income of the parents and having more than one PKU patient in the family increases the suffering. Also not believing in diet taking protein for the children, as most parents due to human thinking believe that children have to eat lots of protein to grow up healthy, in addition the localizing of the service in Gaza city centers while other areas do not have service centers.

1.3 Importance and significance of the diet for PKU patients

Mental retardation can be prevented if the baby is treated with a special diet low in phenylalanine beginning before the fourth week of life. Treatment includes avoiding breast milk, regular formula cow's milk, cheese, meat and fish, because these foodstuffs contain too much phenylalanine. Patients should be subjected to strict follow up at a medical care center or clinic specialized in treatment of this disorder.

However recent studies showed that the high phenylalanine level in the blood, resulted after the special (Lofenelac) diet is stopped can lead to differences in IQ learning disabilities and behavioral disturbances.

1.4 Neurological deterioration

PKU is an inherited disorder of body chemistry that if untreated causes some degree of mental retardation. The majority of untreated PKU patients tend to have lighter colored skin, hair and eyes than the other unaffected family members due to the lack in the synthesis of Melanin. A number of infants have a rash similar to eczema.

Many neuralgic symptoms and signs, especially affecting reflexes occur, seizures are common in older children, and the incidence of abnormal EEGs is 75-90% (Robert Berkow 1997).

Conclusion

The evaluation study of the phenylketonuria program was conducted in the Gaza Strip to highlight the disease and to comprehend the importance of the maximum screening test. The study design was cross sectional, using evaluation of the program in the different stages, laboratory, patients, and clinic.

The data from the governmental central laboratory for all registered newborn babies was used. Abstract sheet was also used to evaluate the procedure of testing in the different related health services. Finally a questionnaire was made to give us a full picture about the PKU patients and their families concerning the gender, residence, complications, parents consanguinity, diet management and health services.

The sample size was 65, all of the patients or the patients' families were interviewed and the investigator completed the questionnaire, and finally a blood test that was done for all the patients. The parents and families of the patients were instructed in their homes or in the nearest governmental clinic on diet management and the importance of follow up.

The coverage in the Gaza Strip is 35.3%, therefore all the findings of the study need implementation for a better program and maximum coverage in the Strip, also the procedures for blood collection and testing needs more strict organization. Cooperation with different health providers is necessary to include the other newborn babies who receive their vaccination in the UNRWA clinics (2/3 of the newborn babies).

The PKU test took place in the second week after birth (61.0%), although it is recommended that the tests should take place in the first week after birth if not before (48-72 hours of life). The earlier the PKU is diagnosed and discovered the more chances for a healthy baby and less complications.

The PKU health services in the Sheikh Radwan clinic needs more staff and facilities to satisfy the required health services including a dietician, a social worker and a psychologist. This is also important to meet the expansion of the screening program to include all the newborn babies in the different provinces in the Gaza Strip.

Health education must concentrate on this subject where good information and lecturing for the population and the parents will prevent or minimize mental retardation of some of our new born babies giving them a better chance to live a normal life