JOURNAL OF CLINICAL AND DIAGNOSTIC RESEARCH

How to cite this article:
KUMAR A. INVESTIGATING FIRST PAEDIATRIC CASE OF PHENYLKETONURIA FROM NEPAL. Journal of Clinical and Diagnostic Research [serial online] 2008 August [cited: 2008 August 14]; 2:1028-1029.
Available from
http://www.jcdr.net/back_issues.asp?issn=0973709x&year=2008&month=August&volume=2&issue=4&page=1028-1029;&id=253

## LETTER TO EDITOR

# Investigating First Paediatric Case Of Phenylketonuria From Nepal

**KUMAR A**

**Corresponding Author**
Dr. Arun Kumar
MSc Medical Biochemistry, PhD (Med)
Assistant Professor, Department of Biochemistry
Manipal College of Medical Sciences, Deep Heights
Pokhara, Nepal

## Introduction

Inborn errors of metabolism are the result of mutations in the DNA deriving altered gene, and ultimately, proteins of abnormal function [1].Phenylketonuria (PKU) results due to the absence of the phenylalanine hydroxylase enzyme.

## Case Report

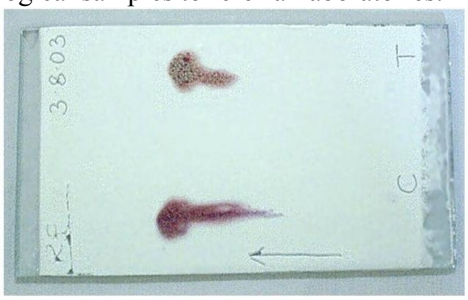
We came across one such suspected case of PKU in a 1.8 years male child, presented by his mother. She complained about delayed development, persistent vomiting after feeding and on micturation, and the bad odour of the urine of her child. After getting a clear consent from his mother, we collected samples for investigation. For collection of blood sample, the plantar surface of the foot was used for skin puncture, and blood was collected using a Natelson micro collection tube. The urine sample was collected in a sterile bottle, and was brought to the Clinical Biochemistry Department for further analysis. Ferric chloride test showed a dark-blue green colour, and was positive. Quantitative analysis of serum showed a value of 14 mg/dl. The finding was further supported by thin layer chromatography.

## Discussion

PKU is an inborn error of phenylalanine that occurs in 1 of every 12,000 births in North America [2]. Early screening and detection can effectively reduce the incidence of phenylketonuria, as every woman is at risk of giving birth to a child with mental retardation, microcephaly, congenital heart disease, and low birth weight. The incidence of this maternal PKU syndrome is 1 of every 30,000-40,000 pregnancies [3]. Guthrie test is the principal screening test for PKU, as the chances of false results in this case are very rare[4]. Gene probes have cracked the possibility of prenatal diagnosis by analyzing DNA isolated or chorionic villi[5]. Though Guthrie test was not done to confirm this case due to lack of infrastructure, this case was confirmed to be positive, as the male child presented with clinical features of PKU, which was further supported by elevated phenylalanine levels of up to 14 mg/dl. Secondly, the detection of phenylalanine in the urine sample by $FeCl_3$ test, was followed by confirmatory test using thin layer chromatography.

Though this is the first case reported from Nepal, we cannot expect to carry out a large scale screening program work in Nepal, where already so many health related problems exist. It is at least expected of the pediatricians not to over look any case, and they have been urged to send biological samples for effective detection and monitoring as soon as possible. In India, the incidence of phenylketonuria has been reported, but no such case is as yet reported

from Nepal, most probably this being the first one. General awareness of in born errors should be given to health workers of rural set up, so that at least they can be aware of such cases and could send biological samples to referral laboratories.




[Table/fig 1] Chromatogram showing phenylketonuriain "Test" compare to 'Control" with Phenylalnine and tyrosine standard

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