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# A unique mutation profile is a significant feature in several pediatric genetic disorders in India

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Mendelian genetic disorders constitute an important health problem worldwide and cause extensive mortality among children in India. With no effective treatment options, prenatal diagnosis and genetic counseling remain principal management options for most genetic disorders. We have performed mutation screening for several common and rare genetic disorders among Indian children and carrier parents following informed consent and clearance from ethics committee. Our analysis included relatively well characterized disorders such as β-thalassemia and Phenylketonuria (PKU) and other rare diseases including Mayple Syrup Urine Disease (MSUD), Farber disease and Hypohidrotic Ectodermal Dysplasia (HED). Our work has revealed differences in the frequency of relatively rare β-globin gene mutations among β-thalassemia patients from North and South India. More importantly, we detected a high proportion of transcript modifying mutations in the Phenylalanine hydroxylase (PAH) gene in PKU patients from India as against other world populations where patients are known to harbor a high proportion of missense (PAH) mutations. Presence of a founder ectodysplasin A receptor mutation has resulted in a high frequency of autosomal recessive form of HED in India unlike other populations wherein a high frequency of ectodysplasin mutations make the X-linked hypohidrotic Ectodermal dysplasia the most common HED subtype. In addition, C-terminal truncating BCKDHA and BCKDHB mutations are a frequent cause of MSUD in India, unlike other populations. Our work constitutes the first mutation analysis for PKU, MSUD, Farber, and HED from India and the first PKU mutation analysis from the South Indian state of Andhra Pradesh and reveals important insights into a unique mutation profile among affected Indian children.

## Biography

Murali Bashyam is Staff Scientist & Chief, Laboratory of Molecular Oncology and has completed his Ph. D in Biochemistry from University of Delhi. He had been working as Visiting International Investigator at Department of Pathology, Stanford University. He has till now got 17 awards including Young scientist medal at Indian National Science Academy (INSA) and Young scientist award at Association of Microbiologists of India. He is a member American Association for Cancer Research and also of Association of UICC. He has till now 38 publication to his credit, and has presented 61 papers at Conferences/Symposia and Invited talks.

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