ISSN 0972-3757
INTERNATIONAL JOURNALOF HUMAN GENETICS

Int J Hum Genet, 1(3): 211-217 (2001)
PRINT: ISSN 0972-3757 ONLINE: ISSN 2456-6330
DOI: 10.31901/24566330.2001/01.03.09

Chemical Diagnosis of Congenital Metabolic Disorders by Gas Chromatography / Mass Spectrometry (GC/MS) in India

Usha P. Dave[1], Vrajesh Udani[1], Josephine Fernando[1], Chunhua Zhang[2] and Isamu Matsumoto[2]

1. CREMERE: Centre for Research in Mental Retardation, Mumbai, India
2. MILS: Matsumoto Institute of Life Science, Kanazawa, Japan

KEY WORDS GC/MS; developmental delay; IEM; genetic counseling

ABSTRACT The present study reports the chemical diagnosis of 254 high–risk children by GC/MS using MILS method, which detects biochemical marker compounds specific for congenital metabolic disorders. Air–dried "urine filter paper" allows simple bedside collection, transportation and a link with other hospitals. Simultaneous analysis of 101 metabolic disorders has the advantage of a wide range of detection to the attending doctor having limited health infrastructure in small towns and rural areas. The definite diagnosis was done in 44 samples (17.3%), reflecting the vast unexplored population of varied genetic etiology. This higher incidence in comparison with the earlier reports (4-5%) from India, based on conventional methods indicated the urgency of latest technology. In critically–ill neonates, the metabolic abnormality was 24.3%. This emphasizes the crucial role of GC/MS in preventing mortality and morbidity. The high–risk genetic factors were consanguinity (13%), family history of mental retardation (13%), and stillbirth and deaths (33%), indicating the racial and ethnic diversity, as well as cultural and traditional impact. The 3 interesting cases among many are discussed where successful management and therapy was done. Accuracy of GC/MS analysis made genetic counselling more effective in evaluating the risk of Inborn Errors of Metabolism (IEM) in future pregnancy.

Home Back