

DEVELOPMENT OF BIOCHEMICAL PROCEDURES FOR THE DIAGNOSIS OF GENETIC DISORDERS

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ABSTRACT

Normal patterns of carbohydrates, amino acids, alpha keto-acids and mucopolysaccharides in the urine of newborns, infants and children (n=118) were established. Comparison with those found in the case group (n=429) that included cases of developmental delay, seizure disorders, mental retardation, autistic behavior, multiple congenital anomalies, multiple skeletal deformities, sepsis, meningitis, etc. highlighted significant findings.

The biochemical procedures done include the common screening procedures for phenylketones, α -keto acids, reducing sugars, tyrosine, cystine and mucopolysaccharides; one- and two- dimensional thin layer chromatography for identification of amino acids, ketoacids and sugars and cellulose acetate electrophoresis for identification of mucopolysaccharides.

In addition to glucose, several in the case group had other sugars like fructose, galactose, xylose and lactose in various combinations. There were twenty in the case group found to be positive for phenylketones, 24 for alphaketones and one positive for cystine. Various amino acid patterns: a generalized increase or an increase in specific amino acids (e.g. glycine, alanine, beta-alanine, methionine, proline, OH-proline, tyrosine, histidine, lysine, glutamate, leucine, isoleucine and valine) were also demonstrated.

The normal samples that were positive in the turbidity test for mucopolysaccharides were found to have chondroitin sulfates A and C by electrophoresis. However 48 in the case group were found to have urinary heparan sulfate and dermatan sulfate either singly or in combination in addition to chondroitin sulfates A and C.

The study established reference data on urinary metabolites in infants and children. It also documented the presence of inborn errors of metabolism among Filipino children.

INTRODUCTION

There is a discernible change in attitude towards genetic disorders amongst medical practitioners. The present attitude is founded on the hope that medicine can help the afflicted and that if the disorder is diagnosed early the undesirable consequences can be prevented. Much of this change in attitude can be traced to better