1. Biotinidase Deficiency : A Pilot Project

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Assay for Biotinidase enzyme activity is considered to be an important diagnostic tool in many neurological disorders and appropriate in neonatal screening programs. We conducted a pilot clinical study to assess the usefulness specificity, and sensitivity of a Biotinidase kit being developed by Bio – Rad Laboratories and Quantase, which uses filter paper blood spots for screening. A total of 265 Paediatric patients and normal newborns were screened. To increase the range of samples tested, we used selection criteria, whereby we had a mixture of all types of cases from paediatric section. We tested 18 severely deficient children of whom 10 were asymptomatic and did not require any treatment, and 8 required prompt treatment. Most of the patients responded favourably to Biotin treatment. Samples which showed intermediate, deficient, or severely deficient results were further checked by colorimetric method described by Barry Wolf. Patients who were symptomatic and had severely deficient results were crosschecked by Gas Chromatography and Mass Spectrometry and / or Tandem Mass Spectrometry, performed by Dr. M. Duran at Netherlands. One sample was sent for DNA analysis to Dr. Adolf Muehl in Vienna, which confirmed our diagnosis of Biotinidase deficiency (D44H/L71P). This sample had been negative by GC / MS. We found that this kit is more than 95 % sensitive and more than 95% specific. There was only one false negative and only two false positive patients. The screening test appeared to be very simple and economical.