﴿ وَعَلَّمَكَ مَا لَمْ تَكُن تَعْلَمُۚ وَكَانَ

فَضْلُ ٱللَّهِ عَلَيْكَ عَظِيمًا ﴿١١٣﴾ ﴾

(سورة النساء:آية ١١٣)

IDENTIFICATION OF SOME DIFFERENT TYPES OF MUCOPOLYSAECHARIDOSES DISEASE IN AFFECTED CHILDREN IN EGYPTIAN POPULATION BY BIOPHYSICAL AND BIOCHEMICAL METHODS

Thesis

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Biophysics

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التعرف على بعض الانواع المختلفة لأمراض السكريات المتعددة المخاطبة فى الاطفال المصابين فى التجمعات السكانية المصرية بواسطة الطرق الفيزيائية الحيوية والكيميائية الحيوية

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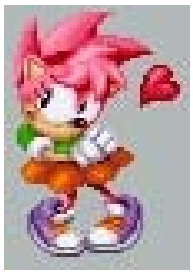

To My Parents
and
daughters
Israa, Hager, Rehab

Abstract

MPS are inheritable disorders caused by deficiency of lysosomal enzymes. The most commonly detected clinical manifestation were coarse facies and mental retardation in all cases followed by hepatosplenomegaly, skeletal deformity, cataracts umblical hernia, heart affection and hearing affection.

Our study detected increase in the activity of hyaluronidase in leukocytes in MPS patients. Abnormalities of hyaluronidase showed very significant elevation of hexuronic acid in leukocytes of affected children (males and females) and their parents. A decrease in sulfite oxidase and sulfite reductase activities in leukocytes. This decrease in enzyme activity leads to increase of sulfate radicals in leukocytes of affected patients.

Our results indicate a significant increase of manganese in hair of affected children (males and females) with MPS than normal controls. Molybdenum in hair was very highly significantly decreased in female children but with highly significant decrease in male affected children compared with significant decreasing of sulfite oxidase activity in leukocytes.

Five standard types of glycosaminoglycans were analyzed by furier-transform infrared spectroscopy (FT-IR) for affected children by MPS disrders. They were classified into three groups. The first group was diagnosed as Hurler syndrome(MPSI) with the presence of dermatan sulfate in urine. The second group diagnosed as Morquio syndrome(MPS-IV) in 16.6% of cases with MPS, 2 cases were (MPS-IVa) type and 2 cases were (MPS-IVb) type. MPS-IVa type had keratan sulfate and chondroitin-6-sulfate in the urine while MPS-IVb showed the presence of keratan sulfate and chondroitin-4-sulfate in urine. The third group was diagnosed as Moroteoux-Lamy syndrome(MPS-VI) in 33.3% of the cases with three cases diagnosed as (MPS-VIa) and five cases diagnosed as (MPs-VIb) respectively. In conclusion quantivative estimation of glycosaminoglycans by measurment of hexuronic acid content in urine and leukocytes are good markers to detect MPS disrders. Our results recommend FT-IR spectroscopy as a very new molecular diagnostic technique to MPS disease in addition to the fact that it is reliable,rapid and cheap method.

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