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فَضْلُ اللَّهِ عَلَيْكَ عَظِيمًا ﴾



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

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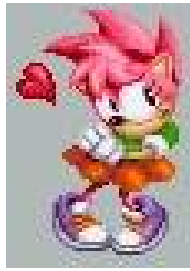
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**To My Parents  
and  
daughters  
Israa, Hager, Rehab**

## 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 umbilical 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 disorders. 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 disorders. 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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# CONTENTS

*Acknowledgement*

*Abstract*

*List of tables*

*List of figures*

*List of Abbreviation*

*Introduction*

*Aim of work*

	Page
<i>Review of literature</i> .....	1-51
<i>Theoretical review</i> .....	52-58
<i>Materials and methods</i> .....	59-86
<i>Results</i> .....	87-149
<i>Discussion</i> .....	150-155
<i>Summary and conclusion</i> .....	156-158
<i>References</i> .....	159-176
<i>Arabic summary</i>	

## LIST OF TABLES

	<b>Subject</b>	<b>Page</b>
<b>Table (1) :</b>	Nomenclature of glycosaminoglycan and proteoglycans	2
<b>Table (2) :</b>	Shows main genetic and manifestations among studied patients with different type of MPS	108
<b>Table (3) :</b>	Glycosaminoglycans (GAGs) level as cetylpyridinium chloride (CPC), hexuronic acid (H.A) and sulfate ( $\text{SO}_4^{-2}$ ) related to creatinine in urine of control and affected female children.	111
<b>Table (4) :</b>	Glycosaminocans level (GAGs) as cetylphridinium ehloride (CPC), hexuronic acid (H.A) and Sulfate ( $\text{SO}_4^{-2}$ ) related to creatinine in urine of control and affected male children.	111
<b>Table (5):</b>	Glycosaminoglycans level (GAGs) as cetylpyridinium chloride (CPC) hexuronic acid (H.A) and sulfate ( $\text{SO}_4^{-2}$ ) in leukocytes of control and affected children.	112
<b>Table (6):</b>	Glycosaminoglycans level (GAGs) as cetylpyridinium chloride (CPC) hexuronic acid (H.A) and Sulfate ( $\text{SO}_4^{-2}$ ) in leukocytes of control and affected male children.	112
<b>Table (7):</b>	Glycosaminoglycans (GAGs) of enzyme activities of hyaluronidase, sulfite reductase and sulfite oxidase in leukocytes of normal and affected female children.	113
<b>Table (8):</b>	Glycasaminogycans (GAGs) of enzyme activities of hydraluronidase, sulfite reductase and sulfite oxidase in leukocytes of normal and affected male children.	113



	<b>Subject</b>	<b>Page</b>
<b>Table (9):</b>	Concentrations of manganese ( $Mn^{+2}$ ): and molybdenum ( $Mo^{+2}$ ) in hair of normal and affected female children.	114
<b>Table (10):</b>	Concentration of manganese ( $Mn^{+2}$ ) and molybdenum ( $Mo^{+2}$ ) in hair of normal and affected male children.	114
<b>Table (11):</b>	Glycosaminoglycans (GAGs) level as cetylpyridinium chloride (CPC), hexuronic acid (H.A) and Sulfate ( $SO_4^{-2}$ ) related to creatinine in urine of fathers of control and affected children.	115
<b>Table (12):</b>	Glycosaminoglycans (GAGs) level as cetylpyridinium chloride (CPC) , hexuronic acid (H.A) and sulfate ( $SO_4^{-2}$ ) related to creatinine in urine of mothers of control and affected children	115
<b>Table (13):</b>	Glycosaminoglycans level (GAGs) as cetylpyridinium chloride (CPC), hexuronic acid (H.A) and Sulfate ( $SO_4^{-2}$ ) in leukocytes of fathers of control and affected children.	116
<b>Table (14):</b>	Glycosaminoglycans level (GAGs) as cetylpyridinium chloride (CPC), hexuronic acid (H.A) and sulfate ( $SO_4^{-2}$ ) in leukocytes of mothers of control and affected children	116
<b>Table (15):</b>	Glycosaminoglycans (GAGs) of enzyme activities of hyaluronidase, sulfite reductase and sulfite oxidase in leukocytes of fathers of control and affected children.	117
<b>Table (16):</b>	Glycosaminoglycans (GAGs) of enzyme activities of hyaluronidase, sulfite reductase and sulfite oxidase in leukocytes of mothers of control and affected children	117

	<b>Subject</b>	<b>Page</b>
<b>Table (17):</b>	Significant statistical correlation of control male children with different parameters in the study.	118
<b>Table (18):</b>	Significant statistical correlation of control male children with different parameters in the study.	119
<b>Table (19):</b>	Significant statistical correlation of control female children with different parameters in the study.	120
<b>Table (20):</b>	Significant statistical correlation of affected female children with different parameters in the study.	121
<b>Table (21):</b>	Significant statistical correlation of fathers of normal children with different parameters in the study.	123
<b>Table (22):</b>	Significant statistical correlation of fathers of affected children with different parameters in the study.	124
<b>Table (23):</b>	Significant statistical correlation of mothers of control children with different parameter in the study.	125
<b>Table (24):</b>	Significant statistical correlation of mothers of affected children with different parameters in the study.	126

## LIST OF FIGURES

	<b>Subject</b>	<b>Page</b>
<b>Fig. (A):</b>	Sugar constituents of glycosaminoglycans.	6
<b>Fig. (A-1):</b>	Biosynthesis of glycosaminoglycans.	7
<b>Fig. (A-2):</b>	Synthesis of 3-phosphoadenosine 5-phosphosulphate PAPS.	8
<b>Fig. (A-3):</b>	Repeating units (disaccharide components) of common glycosaminoglycans.	8
<b>Fig. (A-4):</b>	Repeat unit of chondroitin-4-sulphate.	13
<b>Fig. (A-5):</b>	Repeat unit of dermatan sulfate, catabolism of dermatan sulfate.	13
<b>Fig. (A-6):</b>	Repeat unit of heparan, catabolism of heparan sulfate.	14
<b>Fig. (A-7):</b>	Repeat unit of keratan sulfate.	14
<b>Fig. (A-8):</b>	Repeat unit of hyaluronic acid.	15
<b>Fig. (2):</b>	Schematic representation steps in the degradation of heparin sulfate.	17
<b>Fig. (3):</b>	Schematic representation of steps in the degradation of dermatan sulfate.	18
<b>Fig. (4):</b>	Formation of 3-phosphoadenosine 5-phosphosulfate, an active intermediate involved in sulfate reduction.	43
<b>Fig. (5):</b>	A simple correlations of group vibration to regions of infrared absorption.	51

	<b>Subject</b>	<b>Page</b>
<b>Fig. (6):</b>	Various modes of vibrations.	53
<b>Fig. (7):</b>	The group wave numbers of secondary amides.	58
<b>Fig. (8):</b>	Standard curve for protein Determination (Albumin was used as a standard).	82
<b>Fig. (9):</b>	Standard curve of inorganic sulfate used sodium sulfate as a standard.	83
<b>Fig. (10):</b>	Standard curve for creatinine determination (creatinine deissolve in N-HCl used as standard)	84
<b>Fig. (11):</b>	Standard curve of CPC, (chondroitin sulfate (CS) used as standard ).	85
<b>Fig. (12):</b>	Standard curve used (Hexauronic acid dissolve in benzoil solution as standard).	86
<b>Fig. (13-a):</b>	Glycosaminoglycans (GAGs) in normal urine as cetylpyridinium chloride in may (CPC) <sub>1</sub> mg creatinine.	128
<b>Fig. (13-b):</b>	Glycosaminoglycan (GAGs) in affected urine as cetylpyridinium chloride (CPC) <sub>1</sub> / mg cretinine.	128
<b>Fig. (14-a):</b>	Glycosaminoglycans (GAGs) in affected urine as hexuronic acid in mg (HA) <sub>1</sub> / gm creatinine.	129
<b>Fig. (14-b):</b>	Glycosaminoglycans (GAGs) in affected urine as hexuronic acid in mg (HA) <sub>1</sub> / mg creatinine.	129
<b>Fig. (15-a):</b>	Glycosaminoglycans (GAGs) in normal urine as sulfate in mg ( $\text{SO}_4^{-2}$ ) / mg creatinine.	130
<b>Fig. (15-b):</b>	Glycosaminoglycan (GAGs) in affected urine as sulfate in mg ( $\text{SO}_4^{-2}$ ) / mg creatinine.	130

	<b>Subject</b>	<b>Page</b>
<b>Fig. (16):</b>	Glycosaminoglycans level (GAGs) as cetylpyridinium chloride (CPC) <sub>1</sub> related to creatinine in urine of normal and affected children.	131
<b>Fig. (17):</b>	Glycosaminoglycans level (GAGs) as cetylpyridinium chloride (CPC) <sub>2</sub> related to protein in leukocytes of normal and affected children.	131
<b>Fig. (18):</b>	Glycosaminoglycans levels (GAGs) as hexuronic acid (H-A) <sub>1</sub> related to creatinine in urine of normal and affected children.	132
<b>Fig. (19):</b>	Glycosaminoglycans level (GAGs) as hexuronic acid (H-A) <sub>2</sub> related to protein in leukocytes of normal and affected children.	132
<b>Fig. (20):</b>	Glycosaminoglycans level (GAGs) as inorganic sulfate ( $\text{SO}_4^{-2}$ ) related to creatinine in urine of normal and affected children.	133
<b>Fig. (21):</b>	Glycosaminoglycans level (GAGs) as inorganic sulfate ( $\text{SO}_4^{-2}$ ) related to protein in leukocytes of normal and affected children.	133
<b>Fig. (22):</b>	Glycosaminoglycans level (GAGs) of enzyme activity of sulfite reductase (S-R) in leukocytes of normal and affected children.	134
<b>Fig. (23):</b>	Glycosaminoglycans level (GAGs) of enzyme activity of sulfite oxidase (S.O) in leukocytes of normal and affected children.	134

	<b>Subject</b>	<b>Page</b>
<b>Fig. (24):</b>	Glycosaminoglycans level (GAGs) of enzyme activity of hyaluronidase (H.A) in leukocytes of normal and affected children .	135
<b>Fig. (25):</b>	Manganese concentration in hair of normal and affected children	136
<b>Fig (26):</b>	Molybdenum concentration in hair of normal and affected children.	136
<b>Fig. (27):</b>	Glycosaminoglycans level (GAGs) as cetylpyridinium chloride (CPC) <sub>1</sub> related to creatinine in urine of normal parents and parents of affected children.	137
<b>Fig. (28):</b>	Glycsosaminoglcans levels (GAGs) as cetylpyridinium chloride (CPC) <sub>2</sub> related to protein in leukocytes of normal parents and parents of affected children.	137
<b>Fig. (29):</b>	Glycsoaminoglycans level (GAGs) as hexuronic acid (H.A) <sub>1</sub> related to creatinine in urine of normal parents and parents of affected children	138
<b>Fig. (30):</b>	Glycosaminoglycans levels (GAGs) as hexuronic acid (H-A) <sub>2</sub> related to protein in leukocytes of normal parents and parents of affected children.	138
<b>Fig. (31):</b>	Glycsoaminolglycans levels (GAGs) as inorganic sulfate ( $\text{SO}_4^{-2}$ ) related to creatinine in urine of normal parents and parents of affected children.	139
<b>Fig. (32):</b>	Glycsoasminoglycans levels (GAGs) as inorganic sulfate ( $\text{SO}_4^{-2}$ ) related to protein in leukocytes of normal parents and parents of affected children.	139

	<b>Subject</b>	<b>Page</b>
<b>Fig. (33):</b>	Glycsominoglycans levels (GAGs) of enzyme activity of sulfite reductase (S-R) in leukocytes of normal parents and parents of affected children.	140
<b>Fig. (34):</b>	Glycosaminoglycans level (GAGs) of enzyme activity of sulfite oxidase (S.O) in leukocytes of normal parents and parents of affected children.	140
<b>Fig. (35):</b>	Glycosaminoglycans levels (GAGs) of enzyme activity of hyaluronidase (Hyal) in leukocytes of normal parents and parents of affected children.	141
<b>Fig. (36-a,b):</b>	The FIIR spectra of the GAGs standard from 400-4000 wave number ( $\text{cm}^{-1}$ )	142-143
<b>Fig. (37-a,b):</b>	The FTIR spectra of the GAGs standers from 4000-2000 wave number ( $\text{cm}^{-1}$ ).	144-145
<b>Fig. (38-a):</b>	The FTIR spectra of the patients urine samples representing different types (I, IV (a,b) of MPS disease from 400-4000 wave number ( $\text{cm}^{-1}$ ).	146
<b>Fig. (38-b):</b>	FT-IR spectra of the patients urine samples representing type (V1 (a,b) of MPS disease from 400-4000 .wave number ( $\text{cm}^{-1}$ ).	147
<b>Fig. (39-a):</b>	FT-IR spectra of the patients urine samples representing different type (I, IV (a,b) of MPS disease from 400-2000 wave number ( $\text{cm}^{-1}$ ).	148
<b>Fig. (39-b):</b>	FT-IR spectra of the patients urine samples representing type (VI (a,b) of MPS disease from 400-2000 wave number ( $\text{cm}^{-1}$ ).	149