CLINICAL AND HEARING FUNCTION ASSESSMENT AMONG CHILDREN WITH MUCOPOLYSACCHARIDOSIS ATTENDING ALEXANDRIA UNIVERSITY CHILDREN HOSPITAL

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Introduction

Mucopolysaccharidosis (MPS) is a genetic, progressive and clinically heterogeneous disorder that usually involves multiple body systems due to the deficiency of one of the enzymes that play a role in the metabolism of glycosaminoglycans (GAGs), which accumulates in the lysosomes of multiple organs. It involves wide spectrum of clinical manifestations. Hearing dysfunction is a well known clinical association found in most patients with Mucopolysaccharidosis that can't be underestimated as children acquire language basically by hearing what is being said by those around them, and good hearing is involved in the development of speech and language as well as socialization.

Aim of the work

This work aimed at clinical characterization of MPS cases attending genetics clinic at Alexandria University Children Hospital, enzymatic and genetic diagnosis of the cases and detailed evaluation of their hearing function.

Subjects and Methods

This descriptive cross sectional study was conducted on seventy-five patients with confirmed diagnosis of different types of Mucopolysaccharidosis diagnosed in the genetics clinic of Alexandria University Children Hospital from the year 2010 till the year 2020.

History and clinical evaluation, GAGs detection in urine, serum enzyme assay , molecular diagnosis and hearing function assessment either by auditory brain stem response or pure tone eudiometry was done for the cases .

Results

Table: Distribution of the studied MPS cases according to variant coordinate in each gene (n = 68)

Disease	Gene	Variant Coordinate	No.	%
MPSI (n = 8)	IDUA	c.1598C>G	5	62.5
		c.1045G>C and c.1160T>C	1	12.5
		c.395T>C	1	12.5
		c.766delC	1	12.5
MPSII (n = 12)	IDS	c.1122C>T	3	25
		c.36G>A	3	25
		c.1224dup	2	16.6
		c.1006+1G>A	1	8.3
		c.329G>A	1	8.3
		c.103+1G>A	1	8.3
		c.935G>T	1	8.3
MPSIIIA (n = 5)	SGSH	c.658G>A	2	40.0
		c.88+2T>C	2	40.0
		c.1127T>G	1	20.0
MPSIIIB (n = 10)	NAGLU	c.1444C>T	3	30.0
		c.1412dupT	1	10.0
		c.1558C>T	1	10.0
		c.1592A>G	1	10.0
		c.2T>C	1	10.0
		c.456dupA	1	10.0
		c.802T>C	1	10.0
		c.889C>T	1	10.0
MPSIIIC (n = 1)	HGSNAT	c.1542+1G>C	1	100.0
MPSIVA (n = 22)	GALNS	c.472G>T	13	59
		c.1174C>T	2	9
		c.3381G>A	2	9
		c.1156C>T	1	4.5
		c.1503C>G	1	4.5
		c.452C>T	1	4.5
		c.490G>T	1	4.5
		c.470C>T	1	4.5
MPSVI (n = 8)	ARSB	c.691-1G>A	3	37.5
		c.1036delG	1	12.5
		c.158A>G	1	12.5
		c.245T>G	1	12.5
		c.349T>C	1	12.5
		c.960C>G	1	12.5
(MPSVII) (n = 2)	GUSB	c.1065+5G>A	2	100.0

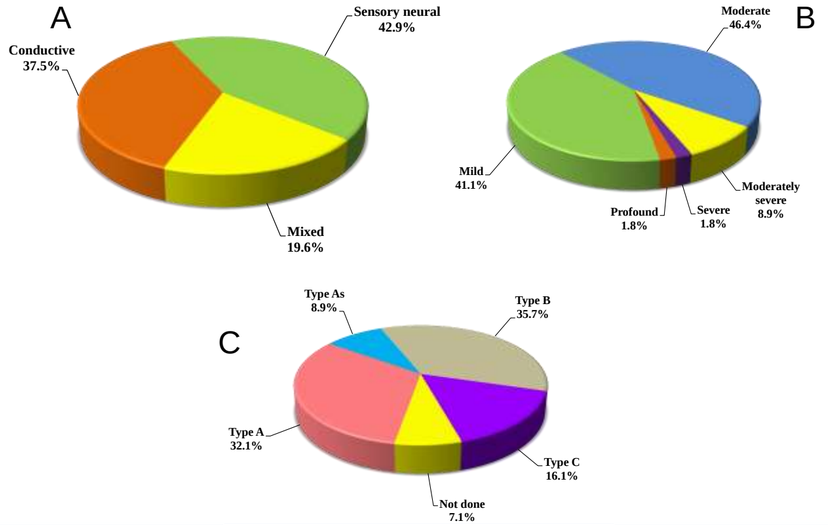

Figure: Distribution of the studied cases according to (A) type and (B) degree of hearing loss and (C) tympanometric curve

Conclusion

- The findings of the current study suggest that raising the awareness towards rare diseases among primary health care physicians to detect the patients in early stage and hence make the best use of treatment. It is important to emphasize that otological evaluations of MPS patients should be performed as early as possible to avoid middle ear conditions that interfere with the quality of life and language development of young children with the need for audiology, speech, and language therapy services to work together as soon as a child is diagnosed with severe, ongoing hearing difficulties.