Challenges in the diagnosis and management of children with spinal muscular atrophy in Ghana: a five-year retrospective review

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INTRODUCTION

Spinal muscular atrophy (SMA) is a rare autosomal recessive disease leading to degeneration of the anterior horn cells with progressive muscle weakness.[1] In limited-resource settings, it presents a diagnostic challenge, and the management remains supportive.[2] In Ghana, only a few cases have been reported in the literature.[2] It is assumed that cases are either missed or mis-diagnosed due to lack of expertise and resources.

OBJECTIVES

This study describes the challenges in the diagnosis and management of SMA in five paediatric centres in Ghana.

MATERIALS & METHODS

This is a retrospective folder review of children diagnosed with SMA at paediatric centres in Ghana between January 2018 and September 2023. Data retrieved included referral details, demography, SMA phenotypes, management, and outcomes. Supervising specialists were interviewed on what they perceived to be the major and minor challenges in the diagnosis and management of SMA in Ghana. Ethical approval was obtained from the Ghana Health Service Ethics Committee.

RESULTS

Seventeen children from 14 families were diagnosed with SMA, confirmed by SMN1 deletion at 5 paediatric centres. They included 9 females and 8 males, aged 3 - 108 months. Phenotypes were SMA1 (10/17), SMA2 (3/17) and SMA3 (4/17). Five came through emergency centres with respiratory distress, while 12 were referred to specialist clinics with various diagnosis (table 1). SMA diagnosis and management were supervised by 3 paediatric neurologists (11/17), 1 developmental paediatrician (3/17) and 1 general paediatrician (3/17). All genetic tests were done overseas at the patients' cost (5/17) or through special programmes (12/17). Treatment was mainly supportive. Two patients received genetic modification therapy. Nine cases (8 SMA1 and 1 SMA2) died from respiratory complications. The supervising specialists perceived lack of expertise (5/5), non-availability of genetic testing locally (5/5) and high cost of testing overseas (4/5) as major diagnostic challenges. Non-availability of gene therapies (5/5) was perceived as a major challenge in management (table 2).

Table 1: Referring clinicians vs. referring diagnosis

Referring clinician	Referring diagnosis				Total
	Respiratory distress	Suspected SMA	Floppy infant	Cerebral palsy	
Paediatrician (or trainees)	3	2	1	0	6
General practitioners	2	0	1	4	7
Nurse	0	0	1	0	1
Self-referral	0	2	1	0	3
Total	5	4	4	4	17

Table 2: Perceived challenges in diagnosis and management of SMA

	No. of respondents		
Diagnostic challenges	**Major**	**Minor**	**Not a challenge**
Lack of expertise	5	0	0
No newborn screening programme	1	2	2
Genetic testing not available locally	5	0	0
High cost of testing overseas	4	1	0
Challenges in management			
Lack of expertise	0	3	2
Gene therapy not available locally	5	0	0
Lack of supportive care	0	3	2

CONCLUSIONS

Lack of expertise, non-availability of genetic testing locally and high cost of testing overseas are perceived as major barriers to the diagnosis of SMA in Ghana. Misdiagnosis is common. We recommend careful histories and detailed physical examinations as the best approach to diagnosis. Current management remains supportive as genetic modification therapies are unavailable.

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