Two Cases of Spinal Muscular Atrophy in Hyderabad, Sindh: A Case Report:

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Abstract: Spinal muscular atrophy (SMA) is a genetic disease that causes muscle weakness and progressive loss of movement. It is a hereditary condition marked by progressive weakening and wasting of the muscles. This is the case report and aimed to investigate **t**wo cases of Spinal Muscular Atrophy in Hyderabad, Sindh. The patients diagnosed with spinal muscular atrophy. Laboratory findings performed such as ECG, EMG, and serum AST, LDH, CK. The primary reasons are ignorance and poverty.
[Amber Mahmood, Saira Baloch. **Two Cases of Spinal Muscular Atrophy in Hyderabad, Sindh: A Case Report..** *Rep Opinion* 2014;6(9):6-7]. (ISSN: 1553-9873). http://www.sciencepub.net/report. 2

Keywords: Spinal muscular atrophy, Clinical investigations, case report

1. Introduction

Spinal muscular atrophy (SMA) is a progressive neuromuscular disease that results in muscle wasting due to deterioration and loss of the anterior horn cells. It is inherited in an autosomal recessive manner due to mutations in the SMN1 gene. In the most common and severe form, SMA type-I **(1-2).** This is the case report and aimed to investigate **t**wo cases of Spinal Muscular Atrophy in Hyderabad, Sindh. The patients diagnosed with spinal muscular atrophy. Laboratory findings performed such as ECG, EMG, and serum AST, LDH, CK. The primary reasons are ignorance and poverty.

Case: 1

Thirteen years old girl was admitted for the treatment of Spinal Muscular atrophy. She was a confirmed case of Spinal Muscular atrophy along with cerebral palsy, confirmed by a team of neuro-physicians.

Parents gave history of child. She was unable to walk not able to get up from lying down or sitting position since birth. She will require a support of a stool or chair to get up. She has weak muscles of lower limbs and hands. She was unable to speak and understanding. Her birth was normal. Her other milestones were normal. Her appetite was good; she was a pleasant, talkative and expressive child.

Her family history suggested that there was not family history of spinal muscular atrophy.

She was investigated thoroughly at Agha Khan Hospital. Her EMG was showing some deficits. The neuro-physicians suggested no major treatment and said that the prognosis was very poor and she would not improve; but gave medicine to keep up the hope

Her Liver enzymes, serum CK, lactate dehydrogenase, and Sodium, Potassium, Calcium aldolase were elevated According to examination of blood RBCs were normocytic and normochromic, platelets were adequate. Electrocardiography and cardiac echocardiography showed no abnormal findings. Accordingly, she was diagnosed as a Spinal Muscular Atrophy carrier.

Case: 2

A 16 year-old male was admitted with complaints of progressive limb weakness that started 9 years prior, with the provisional diagnosis of presumed Spinal Muscular atrophy. Physical examination shows dehydration, semi conscious, tachycardic, chest clear, NVB, abdomen soft non-tender. Aldolases CPK, LDH, Sodium, Potassium, Calcium, were elevated Haemoglobin-15.9, RBC-5.63, PCV-48, MCHC-33.1, and Platelets-75,000. Skin temperature was recorded 34.5 degree centigrade. Bilateral posterior tibial, right and ulnar motor nerves showed normal distal latencies. Patient's history was fails with gait difficulty and difficulty in rising from sitting position since 2 years. Current electro diagnostic study is abnormal; it is suggestive of diffuse neurogenic process involving both upper and lower limbs most probably anterior horn cell disorder like spinal muscular atrophy.

2. Results

Table show the lab investigations in patinets with Spinal Muscular Atrophy

Investigation	Male patient	Femalepatient
Haemoglobin	15.9	11.1
AST	97 U/I	82 U/I
ALT	241U/I	233U/I
CPK	289 U/I	232 U/I
CK-MB	291 U/I	281 U/I
Aldolase	11.4 U/I)	535 U/I
LDH	484 U/I	12.1 U/I)
Sodium	146	128
Potassium	5.1	4.5
Calcium	9.6	7.9

3. Final Outcome:

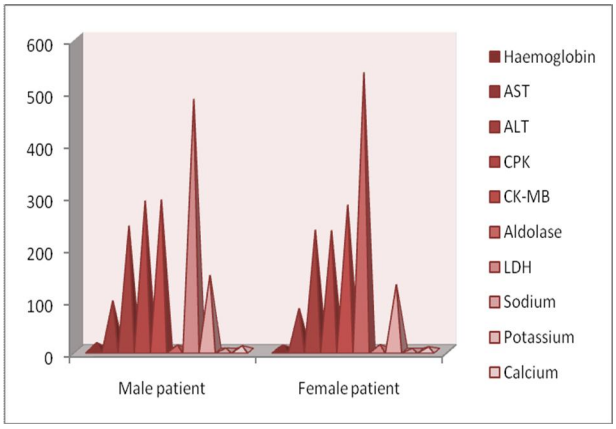

Figure show the lab investigations in patinets with Spinal Muscular Atrophy.

These patients are now (July 2014) about 14 and 16 years old. They live very worst life. They are fully un-movable. They need to support for day to day activities and clinical data confirm that subject's needs improvements. The primary reasons are ignorance and poverty.

We investigate that could not find such case reported earlier. These cases reveal a typical presentation of Spinal Muscular Atrophy, most likely type-I due to the age of presentation and severity of weakness. The management of these patients will require good preventative care and follow-up.

8/28/2014

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