

PRIMARY ADRENAL INSUFFICIENCY IN SUDANESE CHILDREN

ETIOLOGY, CLINICAL PRESENTATION AND DIAGNOSTIC CHALLENGES

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Background

- ❖ Primary adrenal insufficiency, though uncommon, is a condition with high morbidity and mortality. Presentation may vary and could mimic many common childhood illnesses. Many etiologies have been reported, of which CAH followed by autoimmune causes were the commonest in children.
- ❖ Studies on PAI in children apart from CAH were scanty. We reported the first clinical data of PAI in Sudanese children aimed to determine the clinical features at presentation, etiology and diagnostic challenges of PAI in a limited resource country like Sudan.

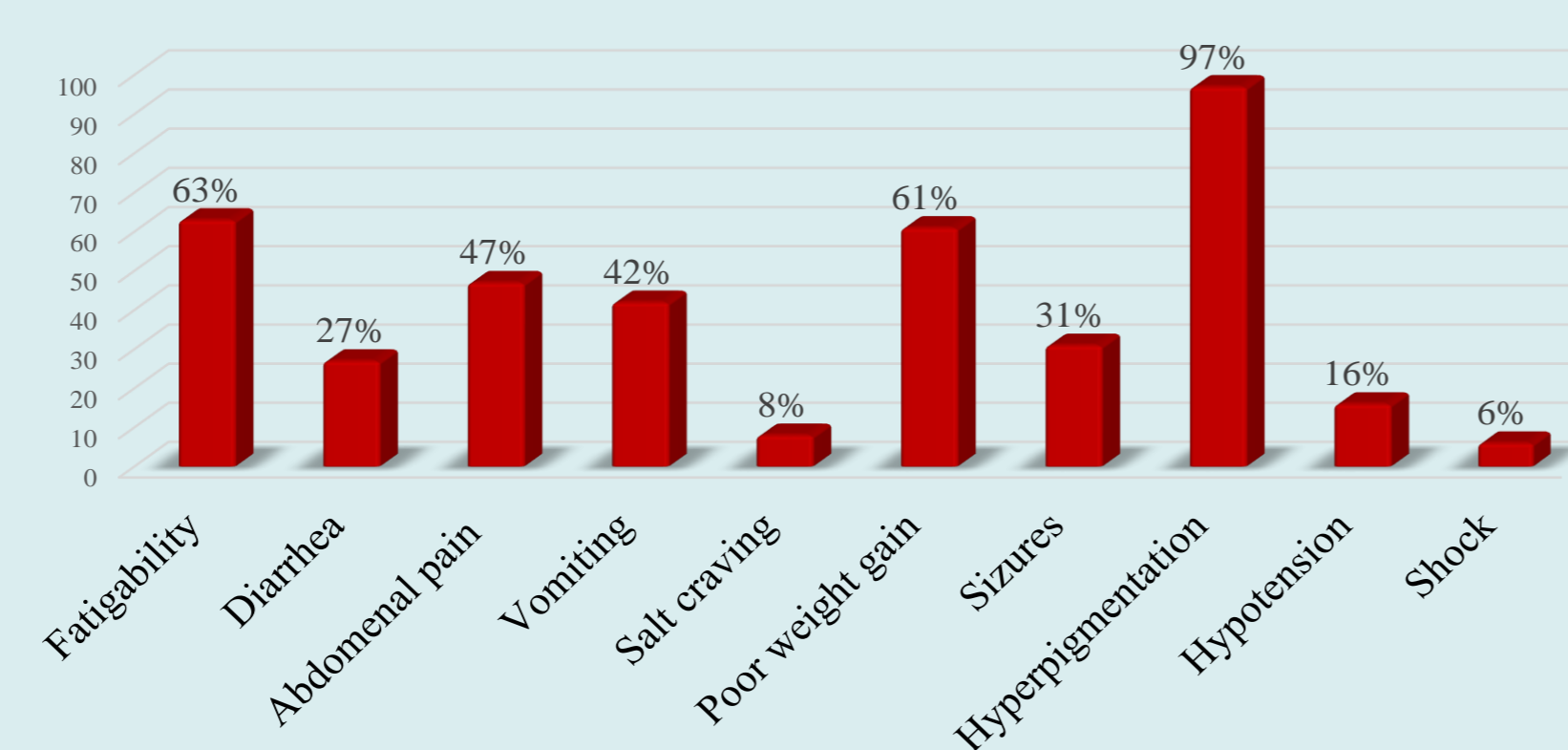
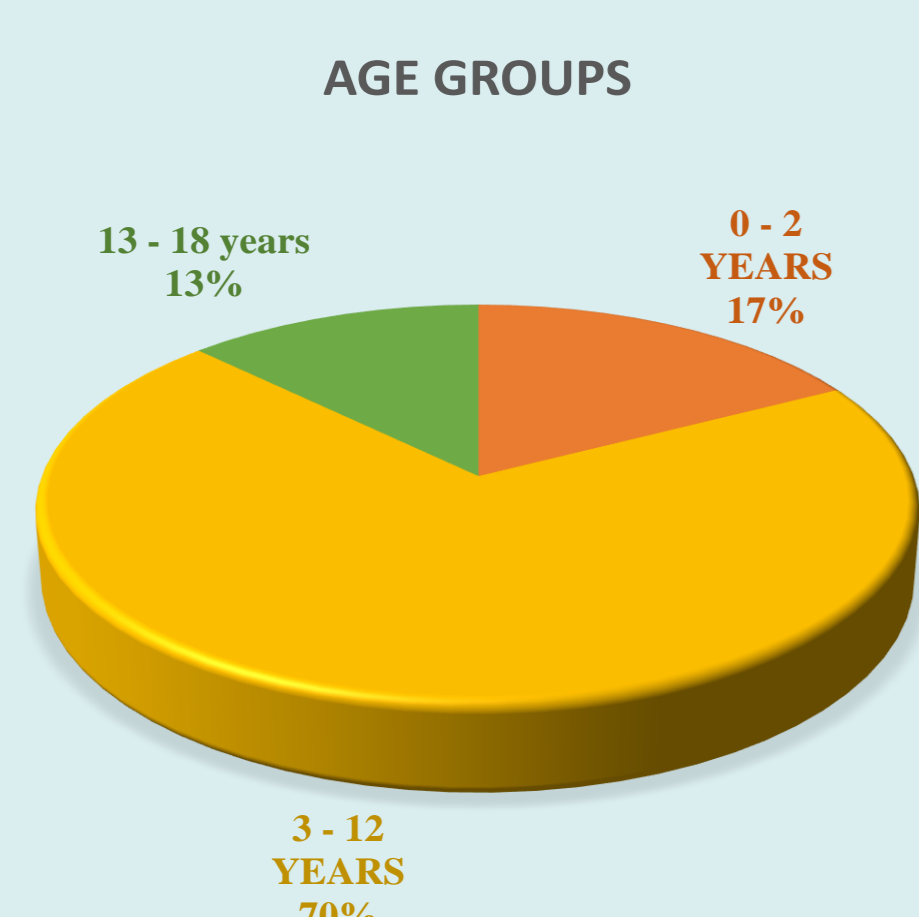
Subjects and Methods

- Descriptive, retrospective, hospital based study carried out in two major pediatric endocrinology centers in Khartoum state, Gaffar Ibn Auf specialized pediatric hospital and Soba university hospital from January 2006 up to December 2018.
- All patient's records were reviewed through a well-structured data collection sheet.
- Diagnosis of PAI was based on:
 1. Characteristic clinical signs and symptoms of PAI ± Symptoms and signs for specific etiologies.
 2. Serum cortisol below the reference ranges ($< 5\mu\text{cg/dl}$) with high ACTH level ($>100\text{pg/ml}$) and/ Or cortisol below 500nmol/l on ACTH stimulation test.
 3. Specific tests that directed towards certain etiology, like VLCFA, MRI for ALD, shimmer/barium swallow/genetics for Allgrove syndrome.
- Difficulties encountered in the diagnosis, availability of certain investigations and their costs as well as barriers of management and availability of certain medications were addressed.
- Patients with known diagnosis of CAH were excluded.

Results

- In a total of 97 patient's records who were referred as suspected PAI, 64 patients (40 males and 24 females) met the inclusion criteria for diagnosis.
- Median age at presentation was 6.27 ± 4.41 years (range: 0.02 to 17 years).
- Symptoms at presentation included hyperpigmentation, fatigability, abdominal pain, diarrhea, vomiting, seizures and shock. Hyper pigmentation was a dominant feature that presented in 62 (96.9%) patients.
- Duration of symptoms before first presentation varied and only (12.5%) came to medical attention within one month from first symptom. 50% had wrong diagnosis at first presentation.
- We were able to determine a diagnosis in 39 (61%) patients of whom 26(40.6%) were Allgrove syndrome, 7(11%) were ALD. 19 (29.7%) patients had possible autoimmune etiology from whom four had APS1 and one had APS2. One (1.6%) had PAI due to bilateral adrenal hemorrhage.
- We were not able to detect the etiology in eleven patients (17.2%).

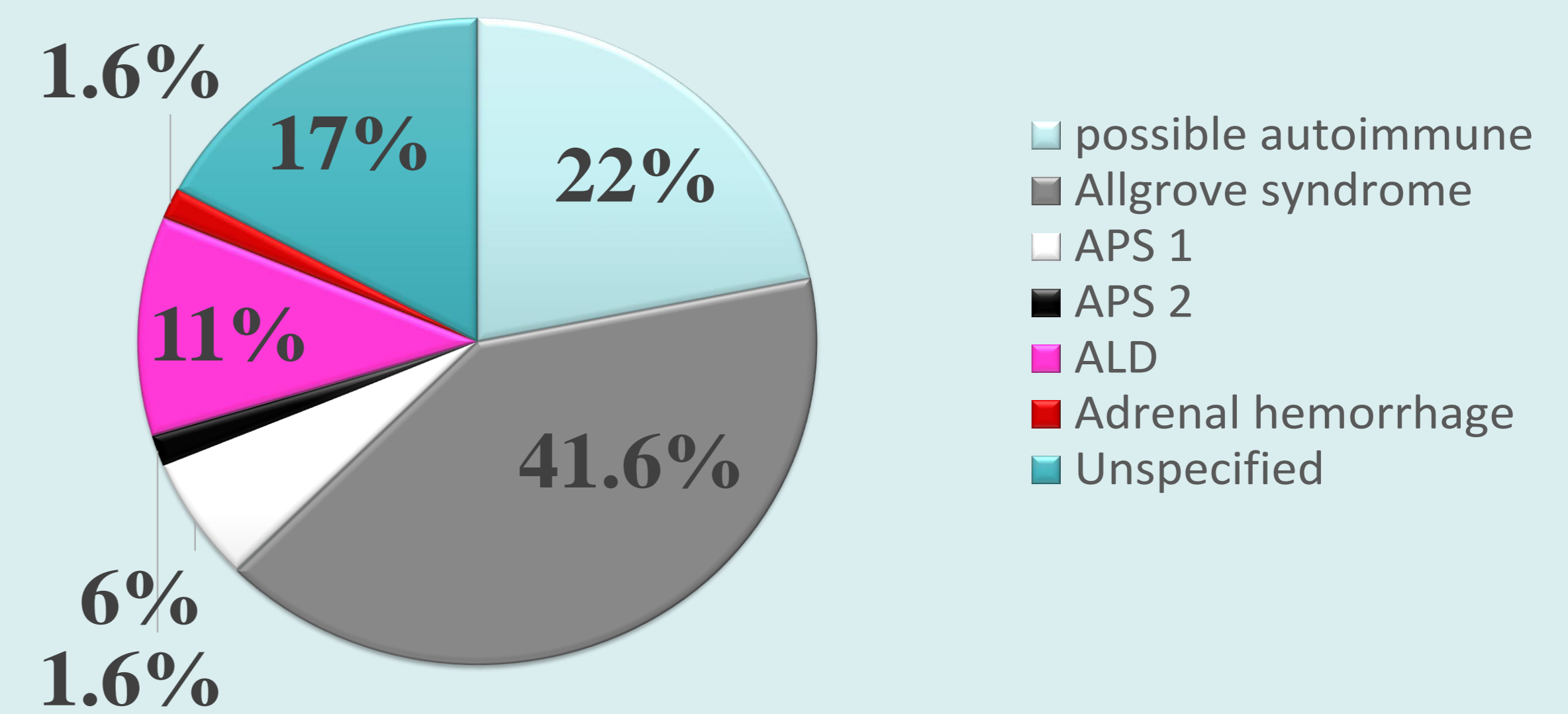
Charts/Graphs/Pictures



Age distribution of PAI patients (n=64)

Common symptoms & signs of PAI patients at presentation (n=64)

Etiology of PAI



Symptoms & Signs in relation to etiology

	Possible autoimmune N (%)	Allgrove N (%)	APS 1 N (%)	APS 2 N (%)	ALD N (%)	Adrenal hemorrhage N (%)	Unspecified N (%)
N	14	26	4	1	7	1	11
Fatigability	13 (93)	17 (65)	3 (75)	1	5 (71)	0	1 (9)
Abdominal pain	10 (71)	12 (46)	3 (75)	1	1 (14)	0	3 (27)
Vomiting	8 (57)	8 (31)	3 (75)	1	1 (14)	0	6 (55)
Diarrhea	3 (21)	9 (35)	1 (25)	0	0	0	4 (36)
Salt craving	3 (21)	0	0	1	0	0	1 (9)
Poor weight gain	8 (57)	16 (62)	4 (100)	1	3 (43)	0	7 (64)
Seizures	2 (14)	12 (46)	0	0	3 (43)	0	3 (27)
Hyperpigmentation	14 (100)	25 (96)	4 (100)	1	7 (100)	0	11 (100)
Hypotension	3 (21)	5 (19)	1 (25)	0	1 (14)	0	0
Shock	2 (14)	1 (4)	1 (25)	0	0	0	0

Discussion

- In Africa, studies of PAI were scanty and limited to Addison disease in general population. Up to my knowledge, no similar studies were published in children.
- The number of patients included in this study (64) was relatively large compared with other similar studies taking into consideration the duration of study which is 12 years and exclusion of the commonest etiology of PAI in children (CAH).
- The fact that Allgrove syndrome make the majority of our patients in this series reflect the rate of consanguinity in our population. Other possibility is that many symptoms in Allgrove syndrome could bring the affected children to medical attention much earlier such as alacrima and difficult swallowing.
- Difficulties included; late presentation or wrong diagnosis at first presentation, difficult access to medical services in some areas, high cost of of certain investigations and unavailability of others like adrenal antibodies and genetic tests.

Conclusion

- Features of increased pigmentation, lethargy, hypotension or electrolyte abnormalities, although unspecific and similar to many other childhood illnesses, yet should lead to a consideration of this diagnosis as early intervention further alters the outcome.
- Most patients were given a definitive diagnosis, while others remained with unidentifiable etiology and need accurate diagnosis after more specific tests and genetic testing.

References

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