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(CASE REPORT)

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Addison's disease with marfanoid features

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Abstract

Addison's disease is a rare endocrine disease resulting from adrenal insufficiency due to various causes. This is a case report of Addison's disease in a young boy 13 year old who presented with 2 months History of darkening of the skin, not gaining weight and decreased appetite was combined with subsequent biochemical testing a diagnosis of Addison's was made and on General Physical examination patient had marfanoid features.

Keywords: Endocrinopathies; Addison's disease; Adrenocortical insufficiency; Hyper pigmented; CT

1. Introduction

Addison's disease is a relatively rare endocrine condition resulting from adrenal insufficiency with a prevalence of 1:100,000 can affect any age group. Autoimmune destruction of the adrenal glands is the most common cause of Addison disease[1] Autoimmune destruction can be an isolated finding or autoimmune polyglandular endocrinopathies (type 1 and 2). Patients with the autoimmune adrenal disease are more likely to have polyglandular autoimmune syndromes[1][2][3]. Addison disease usually manifests as an insidious and gradual onset of non-specific symptoms, often resulting in a delayed diagnosis. The symptoms may worsen over a period of time, which makes early recognition difficult. A high clinical suspicion should be maintained to avoid misdiagnosis [4].

2. Case Presentation

A 13 yr old male child was admitted in June 2022 with a 2 months history of darkening of the skin, not gaining weight and loss of appetite. Basal cortisol plasma level in the morning was 0.3 (5-25) µg/mL and ACTH was done which was 799(0-37)pg/ml. TSH was within normal limits. CT abdomen revealed neither classification nor hemorrhage. Primary adrenal insufficiency was established. Tuberculosis is frequently reported in Addison. In our patient the Tuberculin skin test revealed negative. Treatment planned to be given was oral hydrocortisone 15 mg/m2 and oral fludrocortisone. Based on history, physical examination and laboratory findings, Addison's disease was established.

3. Discussion

The above case highlights the importance of the differential diagnosis especially when the presenting symptoms are similar. Patient had been initially thought of vitamin B12 and folic acid deficiency which came out to be normal. The wide variety in symptoms means that the diagnosis can be attributed to other conditions and the more cardinal features such as skin or mucous membrane pigmentation may be missed although these may not always be present.

Addison's disease is a relatively rare condition. It can be very difficult to diagnose and easily missed due to its presentation with non-specific symptoms. Investigations can provide many clues to the clinician to make them suspect

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Addison's disease [5]. Raised TSH may be a feature and sometimes Addison's disease can be worsened by starting thyroxine [6].

A clinician may then suspect the diagnosis and consider a random cortisol level. However, this can be inaccurate due to the circadian rhythm of cortisol production with peak levels in the morning and low levels at midnight and also an increase in production in times of stress [7]. An unusually low cortisol in the presence of clinical features of Addison's disease should prompt a diagnosis and this may be confirmed by a trial of hydrocortisone. Cortisol and adrenocorticotropic hormone (ACTH) levels should also be taken prior to steroid administration.

Figure 1 Hyperpigmented knuckles

2(A): Arachnodactyly

2(B): Wrist sign

2(C): Thumb sign

Figure 2 Marfanoid features

Before Treatment After Treatment

Figure 3 Before and after starting of the treatment

4. Conclusion

The presence of hyper-pigmentation with normal Serum electrolytes and normal BP initially thought of megaloblast anemia. On review of history the causes of not gaining weight and decreased appetite was combined with subsequent biochemical testing a diagnosis of Addison's was made. Addison's disease although rare does occur in the community. Diagnosis is usually late, leading to increased morbidity and mortality Thus, physicians should keep a high index of suspicion for adrenal insufficiency in unexplained illness.

Compliance with ethical standards

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Disclosure of conflict of interest

There is no conflict of interest between Institution and funding agency

Statement of ethical approval

Not applicable for ethical approval , It is an observational study.

Statement of informed consent

The consent was obtained from the patient care takers.

References

[1] Michels AW, Eisenbarth GS. Immunologic endocrine disorders.JAllergyClinImmunol. 2010Feb; 125(2Suppl2):S226-37.

[2] FischliS.[CME:AdrenalInsufficiency].Praxis(Bern1994).2018Jun;107(13):717-725

[3] SinghG,JialalI.StatPearls[Internet].StatPearlsPublishing;TreasureIsland(FL):Sep 28,2021.PolyglandularAutoimmuneSyndromeTypeII.

[4] ErichsenMM,LøvåsK,SkinningsrudB,WolffAB,UndlienDE,SvartbergJ,Fougner KJ,BergTJ,BollerslevJ,MellaB,CarlsonJA,ErlichH,HusebyeES.Clinical, immunological,andgeneticfeaturesofautoimmuneprimaryadrenalinsufficiency: observationsfromaNorwegianregistry.JClinEndocrinolMetab.2009Dec; 94(12):4882- 90.

[5] Red Larsen P, Kronenberg HM, MelemedS, etal. Williams text book of endocrinology. 10th edn. Saunders Publishing, 2002:525–32.

[6] ShaikhMG,LewisP,KirkJMW. ThyroxineunmasksAddison'sdisease. Acta Paediatr 2004;93:1663–5.

[7] LaycockJ, WiseP. Theadrenal cortexessentialendocrinology.3rd edn.OxfordUniversityPress1996:102-9.