Case Report: Hypoalbuminaemia in THE

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ABSTRACT

Albumin is a plasma protein that constitutes 50% of the protein in the plasma and is a key factor in preventing extravasation of fluid in the extracellular space by the maintenance of oncotic pressure. A reduction in albumin will cause accumulation of fluid over the body due to the decrease of oncotic pressure, pooling fluid into the extravascular space resulting in symptoms of generalised oedema and facial puffiness. Let's consider the case of a 3-year-old boy with tricohepatoenteric syndrome, a rare genetic disorder with a median age of 3.7 that is characterised by chronic diarrhoea, patchy hair, triangular shape of the face, failure to thrive and liver disease and from that, bring forth a discussion forth regarding the implications of hypoalbuminaemia in the frail paediatric population.

INTRODUCTION

Albumin, a plasma protein that constitutes 50% of protein in the plasma, is an important factor in the maintenance of oncotic pressure within vessels and is a key factor in preventing extravasation of fluid in the extracellular space, the presence of which will lead to the presentation of generalised oedema. Causes of hypoalbuminaemia are vast, from congenital, reduced synthesis as a result of diseases such as liver disease, increased catabolism and increased losses, such as through the gastrointestinal tract. Consequently, the formation of ascitic fluid secondary to liver disease can also affect the distribution of albumin in the plasma.[1, 2]

CASE

Termed as *"syndromic diarrhoea",* trichohepatoenteric syndrome is considered to be a rare autosomal recessive genetic disorder characterised by woolly, patchy hair (*tricho*-), manifestations of liver disease in the form of hepatomegaly and cirrhosis (*hepato*-) and intractable diarrhoea often resulting in failure to thrive (*enteric*). Poor prognosis to this disease is often attributed to recurrent infection and eventual cirrhosis. The median age of life is 3.7 years. [3,4]

Thus, we present a case of such a child, a 3 years old male with tricohepatoenteric syndrome with failure to thrive, global and developmental delay and asthma on inhalers, presented to the emergency unit. The father's main complaint that since the last 3 days, the child had progressive generalised body swelling and facial puffiness, mostly noticed to be in the lower and upper limbs as well as the abdomen. The facial puffiness is noticed by the father to improve as the day progresses. These were the only reported symptoms of the visit. Otherwise, there was no underlying fever, urinary symptoms or vomiting. The patient was known to have chronic diarrhoea due to his syndrome, without deviation from his usual pattern of stooling. This patient had several admissions before due to bronchiolitis once and several for gastroenteritis causing multiple electrolyte imbalances, the last of which was dated only two months before his presentation to the emergency room.

He was vitally stable on arrival to the emergency with a blood pressure of 98/58, heart rate of 117, respiratory rate of 20 and saturation of 97% on room air. He was afebrile at a temperature of 36.8 oral.

On examination, he was well, alert but pale. He had dysmorphic features in way of his triangular face and fair patchy hair with short, small stature for age, weighing only 7.8kgs at a height of 79cm, all below the third centile for his age. His chest examination was clear. Heart sounds were heard without a murmur. His abdomen was overtly distended but soft without any illicit tenderness on examination. Shifting dullness was positive. Bowing of the lower limbs were noticed with bilateral lower limb swelling, pitting in nature. Both of his hands were also noticed to be oedematous.

He was fully investigated. He was noted to be not acidotic as a VBG showed a pH of 7.395, HCO3 26 and pCO2 43.4 was obtained during the consult. A blood count, renal profile and electrolyte panel showed a white cell count of 20,000 with associated reactionary thrombocytosis of 690, a haemoglobin of 8.2, a sodium of 135, potassium of 4.8, chloride of 105, urea of 2.6, creatinine of 26, calcium of 2.2, magnesium of 0.8, total protein of 45, albumin of 21, globulin of 24, direct as 2.2, and liver function tests all within acceptable ranges (alkaline phosphatase of 163, alanine aminotransferase of 11, an aspartate aminotransferase of 30 and G-glutamyltransferse of 9.9). The low haemoglobin was suspected to be dilutional due to his generalised oedema.

Additional studies included a chest x ray and a urine. The chest x rays were done, showing no congestion or patches. A urine in and out was performed as the child was not toilet trained as of yet, which showed no proteinuria.

The patient was seen by paediatric team and advised for admission.

During his admission, he was started on albumin, Lasix and was kept on half of his maintenance fluid. Due to his high cell counts, he was started on ceftriaxone, which was discontinued after 30 hours of a negative blood culture. His initial repeat albumin after the first albumin infusion showed an albumin of 19.9, but otherwise all other laboratory investigations were the same as his initial visit to the emergency room. His albumin after his second infusion showed improvement of 34.1 and the child was subsequently discharged after correcting of his imbalance and he did not revisit the emergency or other healthcare facility in the next subsequent weeks due to the same issue.

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Table 1. The trend of his laboratory investigations

Creatinine	25.9		20.6	48.9
Calcium	2.27		1.9	2.28
Inorganic Phosphate	0.76		0.74	0.39
Magnesium	0.72		0.65	0.77
Total Protein	56.3	44	45.1	68.5
Albumin	34.1	19.9	21.1	35.7
Globulin	22.2	24.1	24	32.8
Bilirubin Total	3.34	2.99	2.68	1.94
Bilirubin Direct	1.34	1.49	2.26	1.17
Bilirubin Indirect	2	1.5	0.42	0.77
Alkaline Phosphatase (Alp)	162	159	163	114
Alanine Aminotransferase (Alt)	12.4	12.4	11.7	10.1
G-Glutamyltransferase (Ggt)	11.4	14.1	9.89	9.36
Aspartate Aminotransferase (Ast)	29.8	29.8	29	29.1

DISCUSSION

In the emergency room, diagnosis of emergent conditions and prompt treatment of a patient's disease is a cornerstone of emergency medicine, and that includes patients with rare genetic diseases. More patients with rare genetic diseases are presenting to the emergency department, and this is due to advancements in genetic testing, including next-generation sequencing. However, still it is difficult for many emergency physicians to approach patients with rare diseases. A study by Zhou et al showed that there is "poor education and a need for information" amongst emergency physicians in China regarding knowledge about rare diseases.[5,6]

When it comes to diseases such as tricohepatoenteric syndrome, the likelihood of there being more than one or two patients in the same region afflicted with the condition are low, as there is 1 case in every 1 million births. For an emergency room doctor, dealing with such patients would be difficult, even with good knowledge as such patients might require specialised expertise. Access of which may be difficult.[7]

In the case above, the 3-year-old presented with generalised oedema and facial puffiness without any other symptoms. Causes of hypoalbuminaemia, as mentioned above, are vast. It can be attributed to either a disruption in the tightly controlled capillary haemodynamics, a drainage failure through the lymphatic system or retention of fluid through decreased excretion and filtration by the kidneys. Fluid retention due to increased hydrostatic pressure often occurs due to heart failure, glumeronephritis, renal failure and accidental ingestion of antihypertensive medications as often occurs in the paediatric populations. Obstructive causes such as that of venous obstruction are numerous as well, from malignancies to systemic diseases such as nephrotic syndrome can also result in the patient's presentation.[8]

Considering the patient's disease, presentation and his laboratory results, the most likely causes are protein-losing enteropathy or cirrhotic liver disease. The patient's syndrome, as noted above, is characterised by syndromic diarrhoea, which can result in protein-losing enteropathy including albumin. However, this low albumin could be an early indicator of cirrhotic liver disease.[9] Thus, tight monitoring of the patient's liver functions tests may be needed and trend of albumin and correction are required, hence why the referral to paediatrics was made and admission was advised. The trend of correction of albumin without deviation from his liver function tests go more with protein-losing enteropathy. It is important to note that this distinction would be difficult to make emergently.

This is just one specific case, but in the emergency room, we deal with much more. In a study by Kumar et al, it was noticed that in the retrospective study of 15,000 paediatric emergency visits, 18.6% were by patients with known or suspected genetic disorders.[10] It is important, therefore, to be firstly, consider rare diseases when evaluating a patient in emergency and also be prepared to handle them when they arrive to the emergency door. Application of generalised tools and protocols may be helpful in implementing a better standard of care for these patients, along with adequate education and physician training.

Another suggestion is by the way of a 2020 narrative review of genetic testing in the emergency medicine by Aghamir et al suggested the idea of personalized medicine even in emergency settings, with suggestions of cell-free DNA for diagnosis and pharmacogenetics for decreasing adverse effects of drugs and personalized medication being generated for patients with specific genetic diseases.[11]

CONCLUSION

Taking on cases of patients with rare genetic diseases presenting to the emergency is a cumbersome task. Patients may present with peculiar symptoms and it is our job to decide of such symptoms are attributed to the disease or not. Nevertheless, it is still the job of the emergency physician to optimise patient care and with the growing advancement of genetic testing, these methods of optimisation would look very different in the future.

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