

A UNIQUE PRESENTATION OF PYROGLUTARIC ACIDURIA

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ABSTRACT:

Pyroglutaric aciduria or 5-oxiprolinemia is an under recognized cause of high anion gap metabolic acidosis which is a derangement of gamma- glutamyl cycle. Most common presentation would be vomiting, acidosis, increased anion metabolic acidosis. It is common in paediatric population with inherited autosomal recessive enzyme deficiencies. This paper is emphasized on presentation of pyroglutaric aciduria in a 2 month old .

Keywords: pyroglutaric aciduria, high anion gap metabolic acidosis , 5-oxiprolinemia

INTRODUCTION:

It is a condition in which the body is unable to produce glutathione. Glutathione is generated in the small intestines, kidneys, and liver by gamma-glutamyl cycle. This gamma-glutamyl cycle is responsible for maximizing the absorption of amino acids and plays an important role in chemical detoxification. Elevated levels of gamma-glutamyl amino acids levels can lead to chronic acidosis, central nervous system damage, hemolytic anemia. And as the levels increase, patient becomes more acidotic.

CASE PRESENTATION:

A 2 month old female child born to a 25 year old primi with h/o pih in last trimester and was on tab. labet and h/o hypothyroidism in first trimester and was on tab. thyronorm and the baby was breast fed only for 2 weeks , later the mother was not getting breast milk , so the baby was started on formula feeds. The complaints started later after starting the formula feeds. This baby presented with complaints of loose stools since 4 days, 2 episodes of vomittings since 1 day, 1 episode of convulsion described as gtc which lasted for 5 minutes. .An mri was done, which was suggestive of mild viral encephalitis . csf analysis was done ,which showed protein:102, glucose: 76, TLC : 10, lymphocytic 100% Acyclovir and becef were started, fever, loose stools vomittings subsided but respiratory distress was present. Baby was shifted to picu in view of respiratory distress. On admission necessary investigations were sent. On asking the past history the birth weight of the baby was 2.4kg and after 2 months of age the weight is still 2.4 kg. There is no weight gain and severe failure to thrive was present

ON EXAMINATION:

On examining the baby, baby was requiring O2 support, acidotic breathing was present

Temp: afebrile

Hr: 162/min

Rr: 68/min

Spo2 : 100 % on bc pap

Bp: 98/66 mmhg

Pp: well felt

On systemic examination:

Cvs: s1 s2 heard ,systolic murmur +

Cns: conscious ,irritable, b/l pupils reactive to light, no signs of encephalopathy gc-13/15

Rs: a/ebe+ ,b/l crepts + ,deep breathing + ,scr+

p/a : soft, nt, no hsm+

On examining the stools were white, and multiple episodes of loose stools were present. Sepsis screen was positive. Crp was elevated, HB decreased and platelets reduced. So in view of severe sepsis patient was upgraded to meropenam. Intermittently patient was having fever spikes and high grade which reduced on medication. One pcv transfused at 15cc/kg in view of low hb and o2 requirement. Still the condition was not improving so an abg was sent which showed high anion gap metabolic acidosis.

On admission	After 2 days
Ph: 7.21	PH: 7.24
P02:154 on 5l O2 by bcpap	PO2:154
Pc02:18	PC02:16
Hc03- : 7.7	HC03-: 6.4
Na:145	Na: 138
Cl:118	Cl: 108
Anion gap : 29	23

This metabolic acidosis was persistent, and a suspicion of metabolic disorder was aroused. Then a metabolic workup was done, which showed elevated ammonia (224), elevated lactate. Further the feeds were held, and the ammonia levels repeated after 24 hrs, the levels were shown to be decreasing. A metabolic cocktail was started and on re-introducing the feeds the baby started having loose stools again. So a GCMS/TMS was sent which was suggestive of pyroglutaric aciduria. Further the lactose feeds were stopped and started on soyamilk formula, the baby started getting better, loose stools reduced and then got discharged on soyamilk formula.

CONCLUSION:

Pyroglutamic acidemia is an underrecognized cause of elevated anion gap metabolic acidosis. The objective of this case is to increase awareness that not just one factor alone but a combination of factors, such as sepsis, malnourishment can contribute to the cause. Contributing factors should be identified and corrected, including discontinuing any potentially contributing medications and treatment of underlying conditions.

This paper is to emphasize that pyroglutaric aciduria is one of the common causes of High Anion Gap Metabolic Acidosis, but goes unrecognized due to the rare presentation.