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Bartter Syndrome - A Community Based Case Management

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ABSTRACT

Bartter syndrome is a rare genetic disorder affecting kidney function. It is found in both gender and in all ethnic background. As clinical manifestations differ according to different age group, a detailed history and a thorough physical examination will enable the nurses to detect problems at the earliest. Early investigations and prompt treatment will help the patients prevent complications and enhance longevity. Nurses have a pivotal role in providing a comprehensive care to the patients with Bartter syndrome. Fluid and electrolyte balance, nutrition and prevention of infection are some of the factors to be emphasized for patients with Bartter syndrome. This article describes the pathophysiology, clinical manifestations, medical and nursing management regarding Bartter syndrome.

Key words: Bartter syndrome, rare genetic disorder, kidney function, Gitelman syndrome.

INTRODUCTION

Bartter syndrome was first described in the medical literature in the 1960s by Dr. Frederic Bartter. Bartter and associates in 1962, described two patients who had hyperplasia and hypertrophy of juxtaglomerular apparatus, hyperaldosteronism, hypokalemic metabolic alkalosis, and normal blood pressure. Since that time, many cases have been reported with these findings, and the entity has come to be called Bartter syndrome. Through the years, different terminology has been used to describe these disorders (Kurtzman& Gutierrez, 1975).Bartter syndrome is a general term for a group of rare genetic disorders in which there are specific defects in kidney function. These defects impair the kidney's ability to reabsorb salt and cause imbalances in various electrolyte and fluid concentrations in the body. The electrolytes affected are primarily mineral salts such as potassium, calcium, magnesium, sodium, and chloride. Some researchers classify these disorders based on their clinical appearance, while others classify them based on the underlying mutated gene (National Organization for Rare Disorders).

Epidemiology

The Bartter syndromes affect males and females in equal numbers. Bartter syndromes can occur in individuals of any race or ethnic background (National Organization for Rare Disorders). Exact prevalence of this disorder is unknown, although it likely affects about 1 per million people worldwide. The condition appears to be more common in Costa Rica and Kuwait than in other populations (Genetics Home Reference, 2011).

Etiology

Bartter syndrome is caused by mutations in any one of at least 5 genes and is usually inherited in an autosomal recessive manner.

- Mutation in the SLC12A1 gene causes type 1Bartter syndrome.
- Mutation in the KCNJ1 gene cause type 2Bartter syndrome.

- Mutation in the CLCNKB gene is responsible for type 3Bartter syndrome.
- Mutation in the BSND gene or combination of mutations in the CLCNKA genes cause type 4 Bartter syndrome (Genetics Home Reference, 2011).

These genes are essential for normal kidney function-they are involved in the kidney's ability to reabsorb salt. Abnormal changes in these impair these abilities.

Pathophysiology

Bartter syndrome is a renal tubular salt wasting disorders in which the kidneys cannot reabsorb chloride in the Thick Ascending Loop of Henle (TALH) or the Distal Convoluted Tubule (DCT), depending on the mutation. Impairment in the sodium - potassium - chloride cotransporter or the potassium channel affects the transport of sodium, potassium and chloride in Thick Ascending Loop of Henle. This results in increased distal delivery of the ions, where only some sodium is reabsorbed and potassium is secreted.

Chloride is passively absorbed along most of the proximal tubule but is actively transported in the TALH and DCT. Failure to reabsorb chloride results in a failure to reabsorb sodium and leads to excessive sodium and chloride(salt) delivery to the distal tubules, leading to excessive salt and water loss from the body (Frassetto, 2019).

The resultant volume depletion causes activation of the Renin-Angiotensin-Aldosterone System (RASS) and subsequent secondary hyperaldosteronism. Long term stimulation causes hyperplasia of the juxtaglomerular apparatus and hence increased renin levels (Bokhari, 2020).

Classification

- Classification of Bartter syndrome according to the clinical appearance:
- Neonatal (antenatal) Bartter syndrome
- Classical Bartter syndrome
- Gitelmansyndrome

Classification of Bartter syndrome according to the mutation of genes:

- Mutation in the SLC12A1 gene cause type 1
- Mutation in the KCNJ1 gene cause type 2
- Mutation in the CLCNKB gene are responsible for type 3
- Mutation in the BSND gene or combination of mutations in the CLCNKA genes cause type 4

Once the genetic causes of Bartter syndrome were identified, researchers also split the disorder into different types based on the genes involved.

Type 1, 2 and 4 have the features of antenatal Bartter syndrome. Because type4 is also associated with hearing loss, it is sometimes called antenatal Bartter syndrome with sensorineural deafness. Type 3 usually has the features of classical Bartter syndrome (Genetics Home Reference, 2011).

Clinical manifestations

The antenatal forms of Bartter syndrome may be first be characterized by

- Polyhydramnios
- Premature delivery
- Life threatening salt loss.

Affected newborns may experience

- Excessive urination
- Polyuria
- Fever
- Dehydration
- Vomiting
- Diarrhea
- Some affected infants have distinctive facial features-triangular face, prominent forehead, large eyes, protruding ears, and drooping mouth (as shown in figure 2), failure to thrive, delayed growth, and or intellectual disability.

Classical Bartter syndrome typically becomes apparent in childhood and is characterized by muscle weakness

- Cramping
- Spasms
- Fatigue

- Polydipsia
- Excessive urination
- Dehydration.
- Constipation
- Vomiting
- Elevated body temperature
- Growth and developmental delay.

CASE REPORT

Mrs. R delivered a term boy baby via normal vaginal delivery at a private hospital with the birth weight of 3.2 kgs. The child was apparently well till 8 months old after which the child showed symptoms like vomiting and diarrhea along with intermittent fever on and off. He was referred to a tertiary level hospital by a community health nurse who regularly made home visits. The child was suspected to have AGE with severe dehydration. Post rehydration electrolyte showed hypokalemia. A possibility of barter syndrome was considered in view of severe dehydration, hypokalemia and normal blood pressure. Serum potassium was normalized with oral replacement and the child was discharged when his condition improved.

At one year of age, child showed the symptoms again and was readmitted with severe dehydration and hypokalemia. Child's Blood pressure measured 90/60 mmHg. Heart rate was 100/minute, respiratory rate was 40/minute, Serum potassium - 3.1 mEq/L, creatinine - 0.3 mg/dl with Bicarbonate - 30 mEq/L. hence, started on syrup. Potassium chloride 7.5 ml thrice a day, tab. Indomethacin 25 mg OD and the child was discharged. The dosage of medicine was then tapered. Within few months the symptoms subsided and the medications were stopped. The child is now 15 years old, leading a healthy life.

Diagnosis

Bartter syndrome is usually diagnosed after combinations of tests are performed on an individual with the signs and symptoms of the condition.

Laboratory tests include the following:

- Blood tests to measure serum electrolytes (specifically magnesium, renin, and aldosterone)
- Urine tests to determine the presence of prostaglandin E2 and urine electrolytes (sodium and potassium).

Antenatal subtypes can be diagnosed before birth of the child (prenatally) when polyhydramnios is present without associated congenital malformations and when there are elevated levels of chloride and aldosterone in the amniotic fluid (Genetic and rare diseases information center).

MANAGEMENT

Medical management

- Limited clinical evidence regarding treatment exists due to the rarity of the syndrome.
- Sodium and potassium supplements for the electrolyte imbalances
- Aldosterone antagonists and diuretics Spironolactone are mainstays of therapy.
- Indomethacin or Ibuprofen is used to decrease prostaglandin excretion.
- Growth hormone to treat short stature.
- Low dose ACE inhibitors can help limit the aldosterone mediated electrolyte derangements. Calcium and Magnesium supplements may also be needed (Frassetto, 2019) & (Larosa, 2020).

Nursing consideration

Nurses have major role in performing a complete assessment and providing need based care to the patient and the family. Following are the needs and problems anticipated in a patient with Bartter syndrome, and the specific nursing interventions are listed below.

- Monitor vital signs including temperature frequently.
- Monitor laboratory values for serum electrolytes
- Monitor the weight and dietary intake.
- Assess the skin turgor, mucous membrane and signs of dehydration.
- Maintain intake and output chart.

- Administer prescribed medications, IV fluids and supplements.
- Encourage the client to take plenty of oral fluids.
- Educate the family about the care, treatment and need for compliance.
- Provide emotional support to the family.
- Refer to psychological counseling as needed.

Prognosis

Little information is currently available on the progress of these patients, but most of them tend to present a satisfactory prognosis after a follow-up of at least 10 years although late manifestations such as proteinuria and impaired renal function have been also described (Cunha&Heilberg, 2018).

Complications

Some individuals with bartter syndrome have significant electrolyte imbalances which can lead to irregular heartbeats (cardiac arrhythmias). This can increase the risk for sudden cardiac arrest, growth delays, developmental problems and kidney failure (Genetic and rare diseases information center).

REFERENCES

1. Kurtzman N A, Gutierrez L F. The pathophysiology of Bartter syndrome. Nov 17, 1975) JAMA. 1975; 234(7):758-759. Doi:10.1001/jama.1975.03260200074030
2. National Organization for Rare Disorders. Retrieved from https://rarediseases.org>rare-diseases
3. Genetics Home Reference, reviewed Feb 2011, published June 9 2020. Retrieved from https://ghr.nlm.nih.gov/condition/barttersyndrome
4. Frassetto L A. Bartter syndrome treatment and management May 30, 2019. Retrieved from https://emedicine.medscape.com/article/238670-treatment
5. Bokhari SRA et al April 23, 2020. Retrieved from https://www.ncbi.nlm.nih.gov/books/NBK442019/#_NBK442019_pubdet_
6. Larosa C J. Bartter syndrome and Gitelman syndrome. MSD manual professional version. Jan 2020. Retrieved from https://www.msdmanuals.com/professional/pediatrics/congenital-renal-transport-abnormalities/bartter-syndrome-and-gitelman-syndrome
7. Cunha T S, Heilberg I P. International Journal of Nephrology and Reno vascular Disease. 2018; 11: 291-301 published online 2018 Nov 9. PMCID: PMC6233707 PMID: 30519073 DOI https://doi.org/10.2147/IJNRD.S155397. Retrieved from https://www.dovepress.com/bartter-sydrome-causes-diagnosis-and-treatment-peer-reviewed-fulltext-article-IJNRD
8. Genetic and rare diseases information center. Retrieved from https://rarediseases.info.nih.gov/diseases/5893/bartter-syndrome

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