Research Article **ISSN: 2581-8015**

International Journal of Nursing and Healthcare Research

Journal home page: www.ijnhr.com

https://doi.org/10.36673/IJNHR.2022.v06.i01.A19

BARTTER SYNDROME

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ABSTRACT

Bartter syndrome is one of the autosomal recessive disorder of renal system. The clinical manifestation and predicting results of Bartter syndrome depends upon mutation category and very bad mutation were repeatedly go along with bad prognosis. Bartter syndrome can be unpredictable categorized as a kidney tubulopathy (certain small tubes within the kidneys are affected), a salt-losing disorder (excrete excess amounts of salt), a salt-relesing tubulopathy, and a channelopathy (the ion medium in the kidneys are damaged). Bartter syndrome is a kind of renal disorders that leads to disproportion of electrolytes such as potassium, sodium, chloride, and correlated molecules in the body. Decreased potassium, decreased chloride, metabolic alkalosis, and growth failure are the most repeated clinical manifestations of Bartter syndrome. Even though Bartter syndrome can be fragmented into subcategories based on the gene occupying or symptomatology, significant overrun of symptoms and disease presentation exists among the sub classification and Bartter syndrome may be greatest consideration of as range of disease caused by some dissimilar gene mutations.

KEYWORDS

Bartter's syndrome, Mutations, Gene, Metabolic alkalosis and Tubulopathy.

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INTRODUCTION

Bartter syndrome is a set of indistinguishable renal disorders that cause variance of electrolytes such as sodium, potassium, chloride, and other molecules in the body. In some cases, the disorder manifests in utro with more amniotic fluid surrounding the affected fetus (polyhydramnios).

Cause: Hereditary

Type 1- to IV ---------- Autosomal recessive disorder

Type V -- Autosomal dominant disorder.

Types

Type 1 and type II are neonatal Bartter syndrome, type III is classic form, type IV is Bartter's syndrome with (Sensorineural deafness) and type V is Bartter's syndrome associated with autosomal dominant hypocalcemia.

Associated gene mutations are SLC12A1 (NKCC2) - type I, It develops from mutations in the Nacl or Kcl co-transporter gene. NKCC2 is accountable for bear sodium come out from the tubular lumen into the cell linked with potassium and chloride. Detect in Chromosome 15q21. ROMK/KCNJ1 - type II, It results from mutations in the ROMKgene (locus KCNJ1 on chromosome bands 11 q24-25).

Pathophysiology

CLCNKB - type III, BSND (provide instructions to form barttin protein, It present in the kidneys and inner ear – type IV) and CASR – type V.

Mutations of genes encoding with proteins that transfer ions on top of renal cells in the ascending loop of henle.

Mutations directly or indirectly involving of one sodium, one potassium and 2 chloride ions (Na-K-2Cl) covering the apical membrane of the tubule.

The proteins are manufacture from these genes, It involved in the kidneys' reabsorption of salt. Mutations in any of the five genes impair the kidneys' ability to reabsorb salt, leading to the loss of excess salt in the urine (salt wasting). Abnormalities of salt transport also affect the reabsorption of other charged atoms (ions), including potassium and calcium.

Potassium is able to diffuse back into the tubule- It leads to paracellular reabsorption of both calcium and magnesium ions.

Loss of reabsorption of sodium - Hyper tonicity of the renal medulla- Diuresis - Volume depletion. Finally variation of ions in the body leads to extensive features of Bartter syndrome.

The renin angiotensin aldosterone system is activated with volume depletion – vasoconstriction – to prevent systemic hypotension – increase proximal tubular sodium reabsorpion (compensatory mechanism)

Renal vasoconstriction along with potassium deficiency – vasodilating prostaglandin E (PGE) - high PGE –growth retardation

Decrease chloride reabsorption promotes inadequate exchange of bicarbonate – excessive bicarbonate retention leads to metabolic alkalosis.

Clinical manifestations:

Vomiting

Failure to thrive

Polyuria

Polydipsia

Constipation

Salt craving

Dehydration

Antenatal: polyhydramnios

Increased urine production result from losing too much salt (sodium chloride) in the urine, weakening of the bones can occur due to excess loss of calcium.

Low levels of potassium in the blood (hypokalemia) can cause muscle weakness, cramping, and fatigue

Diagnostic investigations

Electrocadiography

It shows low and flat T wave, accompanied with U wave.

Serum electrolytes

It shows decrease sodium, potassium and chloride (hyponatremia, hypokalemia, and hypochloremia).

Serum aldosterone, rennin activity and angiotensin II activity- it shows high

Blood gas analysis

It shows metabolic alkalosis

Urine test

To determine the presence of prostaglandin E2 and urine electrolytes

Antenatal:

Amniotic fluid analysis –increased levels of chloride and aldosterone

Management

Sodium, potassium,chloride supplementations

Potassium sparing diuretics such as spiranolactone and triamterine – to reduce potassium loss

Non-steroidal anti-inflammatory drugs (indomethacin) with antacid

Angiotensin Converting Enzyme (ACE) inhibitors – to reduce glomerular filtration rate.

Renal ultrasound – to monitor f or the development of nephrocalcinosis
Genetic counseling
Food high in salt, water and potassium

Food high in potassium

Bananas, oranges, apricots, grapefruit (some dried fruits, such as prunes, raisins, and dates, are also high in potassium), spinach, broccoli, Potatoe, Mushrooms, Peas, Cucumbers, Pumpkins, Leafy green, tuna,soyabeans

Fruits and vegetables that are high in water content include

Watermelon, Strawberries, Pineapple, Oranges, Broccoli.

Food high in salt

Salted nuts, pickles, salt biscuits,

Complications

Cardiac arrhythmias
Nephrocalclinosis
Hypokalemic alkalosis
Hypocalciuria

NURSING MANAGEMENT

Appropriate management is vital to prevent potentially life-threatening hypovolemic shock. Children are more likely to develop fluid imbalances. The goals of management are to treat the underlying disorder and return the extracellular fluid compartment to normal, to restore fluid volume, and to correct any electrolyte imbalances.

Fluid and electrolyte imbalance related to impairment of kidney function as evidenced by hypokalemia and signs and symptoms of dehydration.

Risk for complications (cardiac arrest) related to electrolyte imbalance as evidenced by irregular heart beats

Goals and Outcomes

Goals and outcomes for fluid volume deficit:

Child is normovolemic as evidenced by systolic BP greater than or equal to 90mmhg, HR 60 to 100 beats/min, urine output greater than 5mL/hr and normal skin turgor.

Nursing Assessment for Fluid and electrolyte imbalance

Assessment is necessary in order to identify potential problems that may have lead to *fluid volume deficit*

NURSING ASSESSMENT

Monitor and document vital signs especially BP and HR. Increase HR is a compensatory mechanism to maintain cardiac output. Hypotension is evident in hypovolemia

Assess fontenells, skin turgor and oral mucous membranes for signs of dehydration

Assess color and amount of urine. Report urine output less than 5ml/hr for 2 consecutive hours.

Monitor and document temperature. (Insensible water loss occur due to pyrexia)

Find out the refusal of feed, nausea, vomiting and pyrexia. These findings are important to replace fluid requirement.

Check serum electrolytes and urine osmolality, and record values

Check weight daily

Maintain accurate intake and output chart

During treatment, monitor closely for signs of circulatory overload (headache, flushed skin, tachycardia, venous distention, elevated central venous pressure [CVP], shortness of breath, increased BP, tachypnea, cough) during treatment. (Close monitoring for responses during therapy reduces complications associated with fluid replacement.

Monitor and document hemodynamic status including CVP, pulmonary artery pressure (PAP), and pulmonary capillary wedge pressure (PCWP).

Assess the developmental mile stones for the age appropriate (Early detection and intervention can improve the skills and decrease the severity of complications.)

NURSING INTERVENTIONS

Aid the child if he or she is unable to feed, and encourage the mother to assist with feedings, as necessary

Emphasize importance of oral hygiene.

Insert IV cannula and administer parenteral fluids as prescribed. Consider the need for an IV fluid

challenge with immediate infusion of fluids for patients with abnormal vital signs.
Administer blood products as prescribed.
Maintain IV flow rate. Stop or delay the infusion if signs of fluid overload
Provide measures to prevent excessive electrolyte loss (e.g., resting the GI tract, administering antipyretics as ordered by the physician).

Impaired growth and development related to changes in electrolytes as evidenced by muscle weakness

Encourage the parent to feed/ give the amounts of fluid as tolerated or based on individual needs. (Age related)
Educate parent about possible cause and effect of fluid losses or decreased fluid intake.
Teach family members how to monitor output in the home. Instruct them to monitor both intake and output.
Educate parent to monitor the growth, development and report any warning signs.

CONCLUSION

It is a kidney disorder, it will cause fluid and electrolyte imbalance in the body. Early genetic diagnosis and appropriate management may improve the patient health status. The level of disability depends on the severity of the receptor dysfunction, but the prognosis is good in many patients and able to run normal lives.

ACKNOWLEDGEMENT

The authors wish to express their sincere gratitude to Department of Child Health Nursing, Sacred Heart Nursing College, Madurai, Tamilnadu, India for providing necessary facilities to carry out this research work.

CONFLICT OF INTEREST

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Please cite this article in press as: Jothilakshmi R and Sarojini G. Bartter syndrome, *International Journal of Nursing and Healthcare Research,* 6(1), 2022, 100-103.