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Case Report

Abnormal newborn screen in an ex-premature infant fed goat milk

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ABSTRACT

Newborn screening is an important tool in the early diagnosis or detection of rare genetic and metabolic conditions. There are external factors that may influence results. One such is infant nutrition. This article discusses the case of a three month old infant with an abnormal newborn screen in the setting of being fed whole goat milk as well as the importance of reviewing infant nutrition with parents.

Keywords: Infant nutrition, Metabolic acidosis, Milk composition, Goat

INTRODUCTION

Goat milk differs in composition to that of human milk. These differences may play a role in the changes seen in the plasma metabolites of infants fed this milk.[1]

With the introduction of new alternative infant formulas to the United States (US) baby food industry, it is crucial for pediatricians to initiate discussion and address questions or concerns related to infant nutrition especially when parents resort to non-traditional options.

Herein we describe the case of an infant with a false positive newborn screen and abnormal laboratory results caused by feeding with whole goat milk. Following cessation of the causative agent, laboratory findings improved.

CASE REPORT

A 3-month-old female presented from an outside medical facility with laboratory findings of metabolic acidosis in the setting of an abnormal newborn screen collected on day of life 90. The initial newborn screen done at birth resulted as insufficient which prompted the pediatrician to repeat the screen which showed an elevated methionine and propionyl carnitine (C3).

She was born at 33 weeks' gestation (GA) and was admitted to the neonatal intensive care unit (NICU) for one month which was complicated by respiratory failure and anemia of prematurity. After NICU discharge, the infant was seen regularly by the pediatrician and was evaluated for complaints of spit ups and vomiting after formula feeds. Thorough investigation revealed a diagnosis of infant gastroesophageal reflux (GER). The infant formula was then switched from Enfamil to Similac, and rice cereal was added to minimize symptoms and increase caloric intake. Mom reported dissatisfaction with symptom resolution and without physician input, switched feeds to undiluted whole goat milk three weeks prior to presentation. Infant was fed two ounces of goat's milk every two hours with an average of twenty four ounces in twenty four hours.

On physical examination, the infant was afebrile, anicteric, well appearing with good tone and all vital signs were within normal limits for age. A venous blood gas reflected a metabolic acidosis with pH 7.22, pCO_2 46 mm Hg, bicarbonate (HCO_3) 19 mEq/l (19 mmol/l) and base excess of -9. Electrolyte panel revealed an elevated serum

chloride of 109 mEq/l (109 mmol/l), phosphorus 7.2 mg/dl (2.3 mmol/l) and potassium 6.0 mEq/l (6.0 mmol/l) with a blood urea nitrogen (BUN) of 19 mg/dl (6.8 mmol/l).

The genetics team was consulted and genetic workup was initiated. Plasma amino acid profile showed an elevated methionine, phenylalanine, leucine and tyrosine with multiple amino acids in no specific pattern. Plasma acylcarnitine, homocysteine and urine organic acids (metabolites) were all within normal limits. After thorough discussion and evaluation by the geneticists, a specific genetic diagnosis could not be determined based on the reassuring results of a normal homocysteine, acylcarnitine, carnitine and urine organic acids level.

Goat milk was discontinued and infant was restarted on cow milk protein-based formula.

Repeat labs drawn after discontinuing goat milk were as follows: potassium of 5.5 mEq/l (5.5 mmol/l), chloride 105 mEq/l (105 mmol/l), HCO_3 28 mEq/l (28 mmol/l) and BUN 4 mg/dl (1.4 mmol/l). A diagnosis of goat's milk induced metabolic acidopathy was determined to be the most likely cause of presentation.

Parents were thoroughly counseled on the infant's diagnosis and the need to discontinue feeding with whole goat milk at home. Instructions were also given on infant gastroesophageal reflux (GER) and how to appropriately manage at home. Following inpatient discharge, the infant was seen two weeks later with the pediatric genetics specialty. Repeat plasma amino acid profile showed gross improvement with methionine, phenylalanine and leucine levels being within normal limits.

DISCUSSION

Goat milk in its natural unmodified form may contain three times as much protein when compared to human milk with a whey to casein ratio of 20:80 in contrast to the 60:40 accounted for in breast milk.[1] Another important compositional difference is its increased methionine and decreased cysteine amino acid concentrations. This can explain why plasma methionine and phenylalanine, both synthesized de novo by dietary sources, may be elevated in infants who are fed goat milk.[1]

Plasma amino acid profiles attained from this infant were similar to that of premature infants fed a casein predominant cow's formula in one report by Kashyap et al.[2] These infants showed significantly elevated plasma tyrosine levels when compared to the infants who were fed a whey predominant formula.[2] In another study of preterm infants, metabolic acidosis was more evident in infant's fed a casein predominant high protein formulation.[3] Although long term neurological complications of elevated plasma tyrosine have been described in literature, transient elevations may not have the same effect other than falsely affecting results of a newborn screen.[4,5]

Higher protein content present in undiluted goat's milk contributes to increased protein breakdown into acidic phosphates and sulphates. Similarly, the concentrations of chloride, potassium and phosphorus are substantially higher than in human milk. The former two minerals may worsen acidosis by decreasing concentrations of ammonia and bicarbonate. In infants, mechanisms to normalize blood pH in the setting of acidosis such as bicarbonate reabsorption in the proximal tubule, phosphate buffering and ammonia formation is decreased, leading to a decreased ability to combat acidosis.[6]

Within recent years, there has been a shift in the commercial demand for goat milk based infant formulas. Because of this, new products and formulations have been introduced to meet this demand and has since created greater avenues for biomedical research. These formulations usually require modifications to mimic that of human milk. They may undergo dilution, the addition of vitamins and minerals and even protein modification.[1,7]

When evaluated for ileal protein digestion and quality in simulated infant gut studies against human milk formulations, results were similar.[8] In fact, it was hypothesized that both goat and human milk protein may be digested faster.[8] In a double blinded randomized control trial of 200 term infants in Australia, comparisons of goat, cow and human milk formulations were investigated. Both the cow and goat milk formulations were modified to that of a low protein formula, however the whey to casein ratio was kept at 20:80 for the goat milk formulation. As seen in similar trials, the level of plasma valine and phenylalanine were higher in infants fed goat formula compared to that of breast and cow milk formulations.[9]

It is important for pediatricians to engage guardians/parents on topics of alternative nutritional infant feeds and carefully listen to their opinions. With regard to goat milk feeds, discussions of increased infection risk with unpasteurized formulations, inadequate levels of folate (B9), cobalamin (B12) or vitamin E, reports of increased bloody stool and possible allergic reactions must be ongoing with each visit.[1,9]

Despite adverse effects, goat milk possesses some functional and compositional properties that may be of benefit in treatment and prevention of malnutrition. Because goat milk fatty acids are comprised of more medium and branched chain fatty acids, it was proposed as an alternative high energy source after simple modifications in the treatment of undernourished children in Madagascar.[1,10]

CONCLUSION

In summary, clinicians should always discuss infant feeding practices at visits. Although we advocate for breast milk, care must be taken when addressing questions or concerns related to substitute milk formulations.

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