International Journal of Academic Health and Medical Research (IJAHMR)
ISSN: 2643-9824
Vol. 7 Issue 3, March - 2023, Pages: 76-77

Hyperechoic Fetal Colon as Prenatal Sign of Cystinuria

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***Abstract**: Cystinuria is an autosomal recessive metabolic disorder responsible for lithiasis in children and adolescents. Isolated fetal colon hyperechoicity detected prenatally appears to be an early prenatal ultrasound sign of this disorder. We report a case of a 23 year old patient, gravida 2, para1, who presented to the maternity in labor for a full-term pregnancy whose prenatal ultrasound showed fetal colon hyperechoicity, with birth of a girl with a normal physical examination, urine analysis confirmed excessive excretion of cystinuria.*

Keywords: prenatal diagnosis, cystinuria, hyperechoic colon, lithiasis.

Introduction

Cystinuria accounts for 6% of the etiologies of kidney stones in children, the clinical manifestation often occurs after the first decade of life. The main complications of the disease are recurrent urinary tract infections and kidney failure. Early diagnosis of this disease can help to preserve kidney function [1].

Case report

A 23 year old female, with no pathological history, gravida 2, para 1, who has a live child delivered by caesarean section, , admitted to maternity in labor with a pregnancy of 39 weeks without follow-up, the prenatal ultrasound showed a hyperchogenic colon (figures 1, 2) with no other morphological abnormalities, the patient gave birth vaginally after a normal scanopelvimetry, of a female baby with birth weight at 3400g, apgar 10/10. With a normal physical examination. The urine analysis confirmed excess excretion of cystinuria.

Discussion

Cystinuria is an autosomal recessive disease with a prevalence of 1/7000 [2]. It is a metabolic disorder characterized by cystine reabsorption during renal filtration and responsible for lithiasis. It has no known gastrointestinal manifestations [3]. Two main genes have been identified as being responsible for this disease (CLS3A1 and CLS7A9) [4].
Nrmally the fetal colon on obstetrical ultrasound appears hypoechoic or isoechoic relative to adjacent abdominal structures. A hyperechoic colon in the third trimester of pregnancy is rare but has long been considered a normal variant [5,6]. In 2006, Brasseur-Daudruy et al [7] reported three cases of kidney stones, due to cystinuria, in children with a hyperechoic colon in prenatal care. Another study performed by Amat et al [8] on 16 cases with prenatal detection of a hyperechoic colon confirms the association between hyperechoic colon and cystinuria when the hyperechoic colon is observed before 36 weeks of gestation. Maturation and closure of the anal sphincter starting at 22 weeks of gestation leads to a progressive accumulation of cystine in the colon. At high concentrations, this cystine precipitates and forms radiopaque stones that appear as hyperechoicity of the colon on ultrasound [8].
If cystinuria is suspected, amniocentesis can confirm the prenatal diagnosis [9], but it is not indicated because the prenatal diagnosis will have no impact on the outcome of the pregnancy. Post-natal detection of cystinuria on urine analysis can lead to diagnosis and therefore early therapeutic management by hygienic measures and medical treatment to prevent recurrence and progression to renal failure [10].

Conclusion

Prenatal detection of isolated fetal colon hyperechogenicity must be taken seriously in order to eliminate cystinuria in the newborn, especially in the postnatal period, thus preserving renal parenchyma and avoiding progression to renal failure in children with this metabolic disorder.

Conflicts of interest: The authors do not declare any conflict of interest

References

[1]. Buisson, P., Haraux, E., Hamzy, M., Ricard, J., Canarelli, J.-P., Boudailliez, B., & Braun, K. (2011). Cystinurie chez l'enfant : à propos de 4 observations. Archives de Pédiatrie, 18(5),553–557.

[2]. Buxmann, H., Eggermann, T., Lorenz, D., Sewell, A., Böhles, H., Schlößer, R., & Merz, E. (2014). Cystinuria in a Girl Presenting with a Hyperechogenic Colon Detected by Prenatal Ultrasonography and a New SLC3A1 Gene Mutation (p.Phe278Ser). Ultraschall in Der Medizin- European Journal of Ultrasound, 35(05), 473–474.

[3]. Asatoor AM, Crouchman MR, Harrison AR, Light FW, Loughridge LW, Milne MD, Richards AJ. Intestinal absorption of oligopeptides in cystinuria. Clin Sci 1971; 41: 23–33.

[4]. Botzenhart E, Vester U, Schmidt C, et al. 2002. Cystinuria in children: distribution and frequencies of mutations in the SLC3A1 and SLC7A9 genes. Kidney Int 62(4): 1136–1142.
[5]. Paulson EK, Hertzberg BS. Hyperechoic meconium in the third trimester fetus: an uncommon normal variant. J Ultrasound Med 1991; 10: 677–680.
[6]. Fung AS, Wilson S, Toi A, Johnson JA. Echogenic colonic meconium in the third trimester: a normal sonographic finding. J Ultrasound Med 1992; 11: 676–678.
[7]. Brasseur-Daudruy M, Garel C, Brossard V, Broux F, Heckettsweiler B, Eurin D. Hyper-echogenic colon: a prenatal sign of cystinuria? Prenat Diagnosis 2006; 26: 1254–1255.
[8]. Amat, S., Czerkiewicz, I., Benoist, J.-F., Eurin, D., Fontanges, M., & Muller, F. (2011). Isolated hyperechoic fetal colon before 36 weeks' gestation reveals cystinuria. Ultrasound in Obstetrics & Gynecology, 38(5), 543–547.
[9]. Benoist JF, Imbard A, Dreux S, Garel C, Haddad G, Hoffet M, Biou D, Muller F. Antenatal biochemical expression of cystinuria and relation to fetal hyperechogenic colon. Clin Chem 2007; 53: 149–150.
[10]. Assimos DG, Leslie SW, Ng C, Streem SB, Hart LJ. The impact of cystinuria on renal function. J Urol 2002; 168: 27–30.

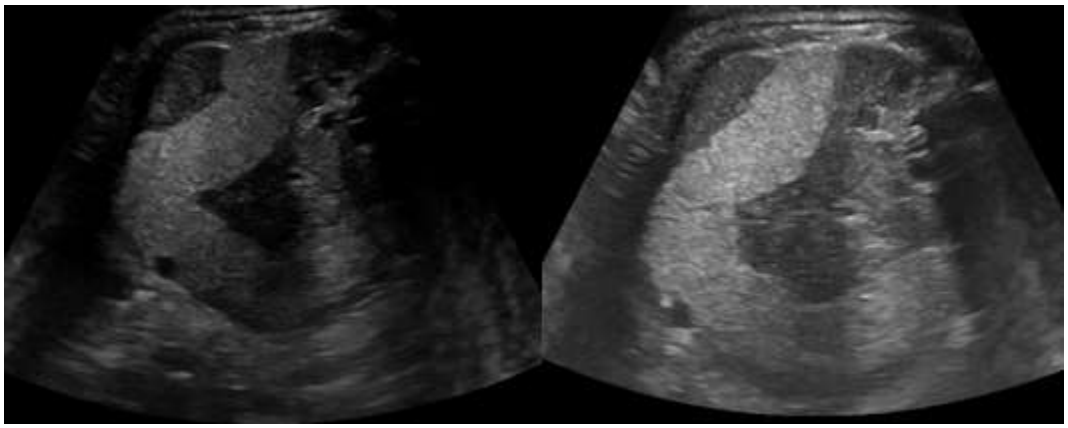

Figure 1 : Axial ultrasound image of the abdomen showing a hyperechoic fetal colon.

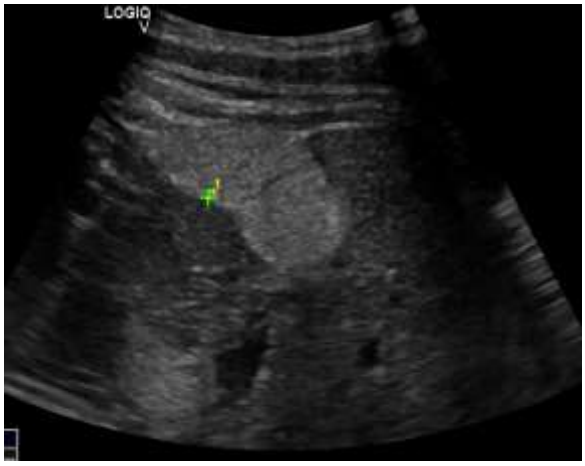

Figure 2 : Coronal ultrasound image of the abdomen showing hyperechoicity of the fetal colon.