Neonatal Case of McKusick-Kaufman Syndrome Difficulty of Diagnosis and Management

Ksibi Imen1, Achour Radhouane2*, Ben Jamaa Nadia1, Bennour Wafa1, Chehour Meriem1, Ben Amara Moez1, Ayari Fayrouz1, Ben Ameur N1, Aloui Nadia1, Neji Khaled2, Masmoudi Aida1 and Kacem Samia1

1 Neonatal Intensive Care Unit, Center of Maternity and Neonatology of Tunis, University Tunis El Manar, Tunisia
2 Department of Emergency, Center of Maternity and Neonatology of Tunis, University Tunis El Manar, Tunisia
3 Department of Foetopathology, Center of Maternity and Neonatology of Tunis, University Tunis El Manar, Tunisia
4 Department of Radiology, Center of Maternity and Neonatology of Tunis, University Tunis El Manar, Tunisia

Abstract

McKusick-Kaufman syndrome (MKKS) is a rare autosomal recessive disorder. We report the case of McKusick-Kaufman syndrome in a term female neonate. Antenatal ultrasound found a large cystic abdominal mass corresponding to hydrometrocolpos with bilateral hydronephrosis. This finding was confirmed after birth and its association to polydactyly permitted us to give the diagnosis of MKKS. Exploratory laparotomy revealed vaginal atresia and suggested the association to Hirschprung disease.

MKKS is difficult to diagnose antenatally and complementary examinations should be done after birth to establish a definitive diagnosis.

Keywords: McKusick-Kaufman syndrome; Neonate; Hydronephrosis; Polydactyly; Vaginal atresia; Laparotomy; Ultrasound

Introduction

Hydrometrocolpos is caused by the accumulation of mucous secretions in the vagina and uterus due to congenital tract obstruction, such as vaginal atresia or imperforate hymen [1]. It can be seen in different syndromes such as McKusick-Kaufman syndrome (MKKS) and Bardet-Biedl syndrome (BBS). MKKS is a rare autosomal recessive disorder. We report the case of a female neonate with hydrometrocolpos, bilateral hydronephrosis and post-axial polydactyly.

Case Report

We report the case of Alaa, a female neonate born at 37 week’s gestation (WG) to a 31 year old mother, 2nd gravida 2nd para with previous history of medical interruption of pregnancy of a male fetus at 22 WG with hexadactyly and enlarged bilateral polycystic kidneys, the diagnosis of Bardet-Biedl syndrome (BBS) was highly suspected by foetopathology examination. At 34 WG, an ultrasound evaluation revealed large cystic abdominal mass as well as bilateral hydronephrosis.

Alaa was born after spontaneous labour by vaginal delivery and required neonatal resuscitation for Apgar score of 3 and 5, respectively at 3 and 5 min. Postnatal examination revealed a birth weight of 3700 g, a head circumference of 32 cm and length of 48 cm. There were facial features of trisomy 21. Alaa had respiratory distress that needed nasal Continuous Positive Airway Pressure (n CPAP).

There were facial features of trisomy 21. Alaa had respiratory distress that needed nasal Continuous Positive Airway Pressure (n CPAP). There was bilateral postaxial polydactyly with brachydactyly of the upper extremities. Abdominal ultrasonography showed a large cystic pelvic mass measuring 10 × 8 × 8 cm situated between the bladder and the rectum, corresponding to a hydrometrocolpos. Both kidneys were enlarged measuring 40 × 25 mm with bilateral hydronephrosis and thinned renal parenchyma measuring 8 × 10 mm. Transverse and left colon were moderately enlarged.

Pressure from the urinary tract, about 150 ml of an opalescent fluid was aspirated from the enlarged uterus. Culture of this fluid was negative. A Petzer probe was inserted into the uterus to release the pressure. Renal function improved after release of the compression secondary to the distended uterus, but enteral feeding was not tolerated with abdominal distension. The neonate then had an exploratory laparotomy that revealed large cystic pelvic mass measuring 10 × 8 × 8 cm situated between the bladder and the rectum, corresponding to a hydrometrocolpos. Both kidneys were enlarged measuring 40 × 25 mm with bilateral hydronephrosis and thinned renal parenchyma measuring 8 × 10 mm. Transverse and left colon were moderately enlarged.

Figure 1: Frontal view of the lower abdomen. Large cystic pelvic mass measuring 10 × 8 × 8 cm situated between the bladder and the rectum, corresponding to a hydrometrocolpos. Both kidneys were enlarged measuring 40 × 25 mm with bilateral hydronephrosis and thinned renal parenchyma measuring 8 × 10 mm. Transverse and left colon were moderately enlarged.

*Corresponding author: Achour Radhouane, Department of Emergency, Center of Maternity and Neonatology of Tunis, University Tunis El Manar, Tunisia, Tel: * 21698549398; E-mail: radhouane.a@live.com

Received September 27, 2016; Accepted October 21, 2016; Published October 28, 2016

Citation: Imen K, Radhouane A, Nadia BJ, Wafa B, Meriem C, et al. (2016) Neonatal Case of McKusick-Kaufman Syndrome Difficulty of Diagnosis and Management. J Neonatal Biol 5: 235. doi: 10.4172/2167-0897.1000235

Copyright: © 2016 Imen K, et al. This is an open-access article distributed under the terms of the Creative Commons Attribution License, which permits unrestricted use, distribution, and reproduction in any medium, provided the original author and source are credited.
revealed a disparity of calibre between a small rectum and an enlarged sigmaoide, transverse and left colon. Hirschprung disease was suspected and rectal biopsy confirmed this diagnosis. Right transverse colostomy was performed. Alaa died 5 days later of septic shock.

Discussion

McKusick-Kaufman syndrome (MKKS) (OMIM 236700) is a rare autosomal recessive inherited syndrome that was first described by McKusick in 1964 [2] in the Old Order Amish population, where it affects an estimated 1/10000 people [1,3,4]. This disease affects the development of the hands and feet, heart and reproductive system. It is characterized by a combination of three cardinal signs: postaxial polydactyly, heart defects and genital abnormalities [1,5]. Hydrometrocolpos presents in 80-95% of affected females and results either from vaginal atresia or imperforate hymen. Congenital heart defects seen in 10-20% of reported cases, includes atrioventricular canal, ventricular septal defect and hypoplastic left heart [1,6]. In our case echocardiography was normal. Slavotinek and Biesecker reviewed the most common associated features in 49 individuals with MKKS phenotype, 75% were diagnosed at birth and 98% by the age of 6 months. The most common feature were hydrometrocolpos (95%), then hydromephrosis (63%), vaginal agenesis (59%); hands were affected in only 29% of cases [11]. The diagnosis of MKKS in males is based on genital malformations (most commonly hypospadias, cryptorchidism); post-axial polydactyly and congenital heart disease [11].

Some evaluations are recommended, following initial diagnosis of MKKS, in order to establish the extent of disease. These evaluations include pelvic ultrasound examination to detect genitourinary malformations, skeletal radiographs to detect osseous polydactyly and syndactyly, echocardiogram to detect congenital heart defects [11].

Congenital vaginal atresia is a rare obstructive anomaly of the female genital tract with a reported incidence at term babies of 0.014 to 1% [14-17]. Individual surgical approaches should be considered to repair this genital anomaly, depending on the anatomical conditions. Various vaginoplasty techniques are available. The surgical method should be chosen based on the patient and the type of anomaly, such as McIndoe technique which is the most popular and preferred technique [14,18,19]. A new technique developed by Vecchietti combines surgical and conservative methods and involves epithelialization from the outer skin layer [14,20].

This disease needs multidisciplinary management and long term support. Ciccone et al. proposed project Leonardo. This project demonstrated the feasibility of incorporating care managers (specially trained nurses) into the health care system. Care managers worked directly with individual patients, helping them to make lifestyle changes, monitoring their conditions and providing the necessary information and advice to promote patient empowerment, enhance
self-management skills and achieve better compliance with care recommendations [21].

Conclusion
MKKS is a rare syndrome. Its symptoms are similar to those in BBS. The diagnosis is difficult. Continued surveillance is recommended and could later establish the diagnosis of BBS. Management is mainly surgical.

References
1. Cherian MP, Al-Sanna’a NA, Ayyat FM (2008) Hydrometrocolpos and acute renal failure: A rare neonatal presentation of Bardt-Biedl syndrome. J Pediatr Urol 4: 313-316.
2. Schaefer E, Durand M, Stoetzel C, Doray B, Viville B, et al. (2011) Molecular diagnosis reveals genetic heterogeneity for the overlapping MKKS and BBS phenotypes. Eur J Med Genet 54: 157-160.
3. McKusick V, Bauer BL, Koop CE, Scott RB (1964) Hydrometrocolpos as a simply inherited malformation. JAMA 189: 813-816.
4. Kaufman RL, Hartmann HF, McAlister WH (1972) Family studies of congenital heart disease II: A syndrome of hydrometrocolpos, postaxial polydactyly and congenital heart disease. J Pediatr 8: 85-87.
5. Lueth ET, Wood KE (2014) McKusick-Kaufman syndrome, complications arising at puberty. J Pediatr Adolesc Gynecol 27: e125-126.
6. David A, Bitoun P, Lacombe D, Lambert JC, Nivelon A, et al. (1999) Hydrometrocolpos and polydactyly: A common neonatal presentation of Bardet-Biedl and McKusick-Kaufman syndromes. J Med Genet 36: 599-603.
7. Beales PL, Ectioglu N, Woolf AS, Parker D, Flinter FA (1999) New criteria for improved diagnosis of Bardet-Biedl syndrome: Results of a population survey. J Med Genet 36: 437-446.
8. Sonmez K, Turkyilmaz Z, Karabulut R, Turan O, Onal EE, et al. (2011) Our experience with McKusick-Kaufman syndrome patients. Bratisl Lek Listy 112: 524-526.
9. Stone DL, Slavotinek A, Bouffard GG, Banerjee-Basu S, Baxevanis AD, et al. (2000) Mutation of a gene encoding a putative chaperonins causes McKusick-Kaufman syndrome. Nat Genet 25: 79-82.
10. Hirayama S, Yamazaki Y, Kitamura A, Oda Y, Morito D, et al. (2008) MKKS is a centrosome-shuttling protein degraded by disease-causing mutations via CHIP-mediated ubiquitination. Mol Biol Cell 19: 899-911.
11. Slavotinek AM (2010) McKusick-Kaufman syndrome. Gene Reviews.
12. Slavin TP, McCendless SE, LaZeinik N (2010) McKusick-Kaufman syndrome: The difficulty of establishing a prenatal diagnosis of an uncommon disorder. J Clin Ultrasound 38: 151-155.
13. Farrell SA, Davidson RG, DeMaria JE, Grant L, Toi A (1986) Abdominal distension in Kaufman-McKusick syndrome. Am J Med Genet 25: 205-210.
14. Ciftci I, Tastekin A, Annagur A, Koplay M. Early abdomino-perineal pull-through vaginoplasty. African Journal of Paediatric Surgery, p: 10.
15. Shaked O, Tepper R, Klein Z, Bezyh Y (2008) Hydrometrocolpos–diagnostic and therapeutic dilemmas. J Pediatr Adolesc Gynecol 21: 317-321.
16. Mor N, Merlob P, Reisner SH (1986) Types of hymen in the new-born infant. Eur J Obstet Gynecol Reprod Biol 22: 225-228.
17. Benson CB, Doublet PM (1996) The fetal genito-urinary system. Sonography in obstetrics and gynecology. London.
18. Wesley JR, Coran AG (1992) Intestinal vaginoplasty for congenital absence of the vagina. J Pediatr Surg 27: 885-889.
19. Lima M, Ruggeri G, Randi B, Domeni M, Gargano T, et al. (2010) Vaginal replacement in the pediatric age group: A 34 year experience of intestinal vaginoplasty in children and young girls. J Pediatr Surg 45: 2087-2091.
20. Bonuto F, Chases CH, Chervenak FA, Fedele L (1999) The Vecchietti procedure for surgical treatment of vaginal agenesis: Comparison of laparoscopy and laparotomy. Int J Gynaecol Obstet 64: 153-158.
21. Ciccone MM, Aquilino A, Corteese F, Sicchitano P, Sassara M, et al. (2010) Feasibility and effectiveness of a disease and care management model in the primary health care system for patients with heart failure and diabetes (Project Leonardo).Vasc Health Risk Manag 6: 297-305.