Association of duodenal and biliary atresias in Martinez-Frias syndrome: A very rare case

Bircan Savran
Dumlupinar University, Turkey

Martinez-Frias Syndrome (MFS), which is characterized by duodenal atresia, extrahepatic biliary atresia, hypoplastic pancreas, intrauterine growth retardation (IUGR) with or without tracheoesophageal fistula, has been firstly described in 1992 by Martinez and Frias and it is a very rare condition with autosomal recessive inheritance. Some other abnormalities such as esophageal atresia, rectoanal atresia, cardiac disorders and hipopspadias can also be encountered in patients with MFS, but these findings may not be necessarily found. MFS has been reported five times in the literature and the total number of reported cases is nine so far, with none alive today. For this patient, the most important thing is that she is the first and still living patient of MFS among the previously reported cases.

Pediatric gastrointestinal basidiobolomycosis: Case series

Abdulaziz Mohammed Al Dakhil, Alaa Aljuaid, Mohammed Haneef, Moayyosr Karami, Walaa Almansory, Hatim Maghrabi, Abdulqader Al Rezqi and Moahmmed Sati
King Saud Bin Abdulaziz University for Health Sciences, KSA

Objectives: Basidiobolus ranarum is a fungus known to rarely cause chronic skin and subcutaneous infections and even more rarely it can present as gastrointestinal basidiobolomycosis (GIB). In this case report we describe our experience with GIB in the western region of Saudi Arabia in which we adopted a conservative approach and limited the role of surgery beyond taking the biopsy. We also describe our utilization of voriconazole that seems a promising agent in treating GIB.

Methods: Health records of five patients with GIB were retrospectively reviewed. These patients were diagnosed between January 2012 and March 2014 at King Khalid National Guard Hospital in Jeddah, Saudi Arabia.

Results: All patients were males from the southwestern region of Saudi Arabia. All of our five cases had leukocytosis, while eosinophilia was significant in four out of five patients. Four cases were managed medically with voriconazole and one case with itraconazole. None of our cases were managed surgically unless complications occurred (two out of five patients). Three cases have shown complete resolution of their symptoms. One patient that required multiple surgeries died later due to surgery complications.

Conclusion: The prognosis of GIB in pediatric patients is usually favorable if diagnosed early and aggressive surgery was avoided. To achieve early diagnosis, high index suspicion is required. We suggest fine needle biopsy instead of invasive biopsy to diagnose the disease by histopathology to minimize complications if possible. We also think that Voriconazole is an effective and promising alternative to Itraconazole in treatment of GIB.