A case of cystic fibrosis presenting as a distended bowel in third trimester

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Objective
Cystic fibrosis (CF) is the most common severe autosomal recessive disease that affects white children, with an incidence between 1/2500 to 1/5000 (carrier rate 1/25 to 1/35). The overall incidence of CF when prenatal diagnosis of intestinal anomaly is made is approximately 13%, more than 300 times higher than the estimated risk in the general population. An hyperechoic pattern with dilated bowel is associated with almost 100 percent specificity for CF. Prenatal screening for CF should be indicated in all pregnancies with US patterns of specific intestinal disorders. CF is caused by a mutation in a gene which encodes a protein called CFTR (cystic fibrosis transmembrane conductance regulator).

Methods
Case report: A 32yrs old, G4P1A1 had previous 3 full term normal deliveries. She had unremarkable past medical/surgical history and no other risk factors noted in the history including no family history of CF. A 24 weeks anomaly scan was normal. The patient was referred at 36 +3 weeks with an US diagnosis of abnormally dilated bowel loops. Scan findings were confirmed in the hospital and had suggested enlarged distended bowel (27-28mm) with hyper echogenic walls, abnormal peristalsis and mild ascites. These findings were highly suspicious of mechanical obstruction like volvulus or atresia. The findings were discussed with the Pediatric Surgeon, parents and NICU. The plan was to evaluate the neonate after delivery. Delivery was uneventful. A live female baby was delivered, wt 2.57kg with good Apgar score. Post-partum examination revealed a distended abdomen and patent anus. Abdominal X-ray showed dilated stomach and small bowel. Water soluble contrast meal suggested an isolated air-filled bowel loop seen with linear lucencies within the wall (suggestive of pneumatosis intestinalis). Features were suggestive of intestinal obstruction due to atresia. The baby was operated at the age of 4 hours and finding consisted of dilated ileum loops filled with sticky meconium pallets. Ileostomy was performed. No ischemic bowel was noted that might suggest volvulus or atresia. Stool microscopy was highly suggestive of CF. CFTR genetic mutation was found. A homozygous mutation c.4242=1>c genotype in HGVS: c 4242 G>C. This mutation has never been reported before this case.

Results
Meconium ileus (MI) is typically reported as a manifestation of CF. CF is diagnosed in almost 100% of neonates with MI, 30% of these are diagnosed antenatal as a result of an intestinal disorder. Genetic studies allow diagnosis of CF during pregnancy, but not all mutations responsible for the disease have yet been identified. MI could be detected during the 2nd trimester of gestation when diagnosis is suggested by either dilated bowel or hyperechoic bowel. Fetal echogenic bowel is associated with an increased risk of CF. Initially, prenatal testing was only offered to selected population. Prenatal diagnosis of bowel echogenicity, that suggestive of CF, can raise the first suspicion for the disease.

Conclusion
The discovery of more than 1000 different mutations in the CFTR gene makes it impossible to detect all mutations by simple routine screening. Furthermore, in general, prenatal testing consists of analysis of a limited number of known mutations, and the mutation detection rate varies according to the molecular technique used, the proportion of the gene screened, and the ethnic origin of the population tested.