Uncommon causes of diarrhoea in children and adults

Joint Session Digestive Diseases Pathology / Paediatric and Perinatal Pathology

European Society of Pathology

30th European Congress of Pathology

Pathology: Path to Precision medicine

8 – 12 September 2018, Euskalduna Conference Centre, Bilbao, Spain

Disclosure Information

I hereby declare that I have had business or personal interests in the following industrial enterprises since 1 September 2017:

Name of the enterprise / Nature of the interest

Enterprise | Interest
None

Case 1

- A 2-year-old male of nonconsanguineous healthy parents,
- Growth retardation, chronic diarrhoea, abdominal distention, gradually progressive swellings of face, upper limbs and calves, since 4 months of age.
- CT scan: bilateral pleural effusion, moderate pericardial effusion, mild ascites and a few slightly enlarged mesenteric lymph nodes.

Laboratory tests

- Stool and urine routine examination, liver, renal and thyroid function, coagulation and blood lipid profiles and electrolytes were all unremarkable.
- Complete blood count revealed a markedly low total lymphocyte count (560 cells/mL with a reference range 1100-3200 cells/uL).
- Total serum protein was **4.00** g/dL (reference range is 6.50–8.50 g/dL), albumin **2.12** g/dL (reference range is 4.00–5.50 g/dL), globulin 1.88 g/dL (reference range is 2.00–4.00 g/dL), alpha-1-antitrypsin was within the reference range.

Your diagnosis?

Primary intestinal lymphangiectasia (Waldmann's disease)

Primary intestinal lymphangiectasia (PIL)

A rare disorder characterized by dilated intestinal lacteals resulting in lymph leakage into the small bowel lumen and responsible for protein-losing enteropathy leading to lymphopenia,hypoalbuminemia and hypogammaglobulinemia.

The prevalence of clinically overt PIL is unknown. It primarily affects children - generally diagnosed before 3 years of age - and young adults

Malabsorption + Fatigue, abdominal pain, nausea, vomiting and weight loss, inability to gain weight and growth retardation

Fat-soluble vitamin deficiencies and hypocalcemia may lead to convulsions

Etiopathogenesis

Several genes, such as VEGFR3 (vascular endothelial growth factor receptor 3), prospero-related homeobox-transcriptional factor PROX1, forkhead transcriptional factor FOXC2 and SOX18 are implicated in the development of the lymphatic system.

Changed expressions of regulatory molecules for lymphangiogenesis in the duodenal mucosa of PIL patients were reported

Treatment: A high-protein, low-fat diet supplemented with medium chain triglycerides (MCT)

Small bowel resection in the rare cases in which intestinal lymphangiectasia is segmental and localized


details matter