Name of Journal: *World Journal of Clinical Cases*

Manuscript NO: 54535

Manuscript Type: CASE REPORT

Tuberous sclerosis complex presenting as primary intestinal lymphangiectasia: A case report

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Abstract

BACKGROUND

Primary intestinal lymphangiectasia (PIL) is a rare congenital protein-losing enteropathy caused by dysplasia of the small intestinal lymphatics. The cause of the disease is unknown. Through a literature review, we found that PIL and tuberous sclerosis complex (TSC) have some common symptoms and molecular pathways.

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Primary intestinal lymphangiectasia treated with rapamycin in a child with **tuberous sclerosis complex** (**TSC**) Correspondence to: Uzma Shah, M.D., Department of Pediatric Gastroenterology, Hepatology and Nutrition, **Massachusetts General Hospital**, 55 Fruit Street, Boston, MA 02114. Correspondence to:

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Tuberous sclerosis complex (**TSC**) is an autosomal dominant **disorder** that results from an inactivating mutation in one of two genes, TSC1 (on chromosome 9q34), which encodes hamartin, or TSC2 (on chromosome 16p13.3), which encodes **tuberin**. Hamartin and tuberin form a **complex** that regulates cell proliferation and differentiation . In TSC, hamartomas and neoplastic lesions form at various sites …

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Primary Intestinal Lymphangiectasia Treated With Rapamycin in a Child With **Tuberous Sclerosis Complex** (**TSC**) **Primary intestinal lymphangiectasia** (**PIL**) is a rare **protein-losing enteropathy** characterized by a **congenital malformation** of the **lymphatic** vessels of the small **intestine** causing insufficient drainage and leakage of **lymph fluid**.

Lymphangiectasia, types, causes, symptoms, diagnosis ...

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.bing.com/?FORM=Z9FD1 and colleagues 47) **reported** a **case** of **primary intestinal lymphangiectasia** in a female patient

Lymphangiectasia

Pathologic Dilation

Lymphangiectasia is a pathologic dilation of lymph vessels. When it occurs in the intestines of dogs, and more rarely humans, it causes a disease known as "intestinal lymphangiectasia". This disease is characterized by lymphatic vessel dilation, chronic diarrhea and loss of proteins such as serum albumin and globulin. It is considered to be a chronic form of protein-losing enteropathy.

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