Primary Intestinal Lymphangiectasia (Genetic Predisposition and Diagnosis)

1. CHAPLE syndrome uncovers the primary role of complement in a familial form of Waldmann's disease

Author(s): Ozen A.

Source: Immunological Reviews; Jan 2019; vol. 287 (no. 1); p. 20-32

Publication Date: Jan 2019

Publication Type(s): Review

PubMedID: 30565236

Available at Immunological Reviews - from Wiley Online Library Science, Technology and Medicine Collection 2017

Abstract: Primary intestinal lymphangiectasia (PIL) or Waldmann's disease was described in 1961 as an important cause of protein-losing enteropathy (PLE). PIL can be the sole finding in rare individuals or occur as part of a multisystemic genetic syndrome. Although genetic etiologies of many lymphatic dysplasia syndromes associated with PIL have been identified, the pathogenesis of isolated PIL (with no associated syndromic features) remains unknown. Familial cases and occurrence at birth suggest genetic etiologies in certain cases. Recently, CD55 deficiency with hyperactivation of complement, angiopathic thrombosis, and PLE (the CHAPLE syndrome) has been identified as a monogenic form of PIL. Surprisingly, loss of CD55, a key regulator of complement system leads to a predominantly gut condition. Similarly to other complement disorders, namely paroxysmal nocturnal and hemoglobinuria (PNH) and atypical hemolytic uremic syndrome (aHUS), CHAPLE disease involves pathogenic cross-activation of the coagulation system, predisposing individuals to severe thrombosis. The observation that complement system is overly active in CHAPLE disease introduced a novel concept into the management of PLE; anti-complement therapy. While CD55 deficiency constitutes a treatable subgroup in the larger pool of patients with isolated PIL, the etiology remains to be identified in the remaining patients with intact CD55. Copyright © 2018 John Wiley & Sons A/S. Published by John Wiley & Sons Ltd

Database: EMBASE

Author(s): Valakada, Jineesh; Madhusudhan, Kumble S; Ranjan, Gyan; Garg, Pramod Kumar; Sharma, Raju; Gupta, Arun Kumar

Source: Current problems in diagnostic radiology; 2018; vol. 47 (no. 3); p. 200-202

Publication Date: 2018

Publication Type(s): Case Reports Journal Article

PubMedID: 28554788

Database: Medline

3. CT Lymphangiography (CTL) in Primary Intestinal Lymphangiectasia (PIL): A Comparative Study with Intraoperative Enteroscopy (IOE)

Author(s): Dong J.; Wen T.; Chen X.; Wang R.; Xin J.; Shen W.; Sun Y.

Source: Academic Radiology; 2018

Publication Date: 2018

Publication Type(s): Article In Press

Abstract: Rationale and Objectives: To investigate the clinical feasibility of CT lymphangiography (CTL) in primary intestinal lymphangiectasia (PIL) by comparison with intraoperative enteroscopy (IOE) during exploratory laparotomy. Materials and Methods: Eleven PIL patients (F/M, two/nine, age range 10-37 years) were recruited in this study, and they were performed IOE during exploratory laparotomy for suspected serious lymphatic-intestinal leakages. All the patients were performed CTL before surgery, and the imaging data were reviewed by two radiologists separately. CTL assessments included intestinal lesions, edematous lesions, intestinal and mesenteric lymphangiectasia, lymphatic-abdominal leakages, lymph fluid reflux, lymphangioma and abnormal lymphatics in other area. The intestinal lymphangiectasia and lymphatic-intestinal leakages were confirmed by histology and IOE. Results: For CTL, (1) nine intestinal wall thickening; (2) eight ascites, complicated with four pleural effusions, (3) eight intestinal and mesenteric lymphangiectasia, (4) six lymph fluid reflux (5) one lymphatic-abdominal leakage, (6) two lymphangioma. While for IOE, intestinal lymphangiectasia has been confirmed in all patients, including five segmental and six diffusive lesions in intestinal mucosa. Besides, one lymphatic-intestinal fistula, one lymphatic-abdominal leakage was confirmed. Compared to IOE and histology, the accuracy of CTL was 72.7% in detecting intestinal lymphangiectasia. Conclusion: Compared to IOE, CTL demonstrates feasibility in detection of intestinal lymphangiectasia and other abnormalities in whole lymphatic circulation for PIL. Combination of CTL with IOE accommodates guidance for preoperative evaluation and therapeutic management for PIL.

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Database: EMBASE
4. Fetal intestinal lymphangiectasia as a causative factor of mirror syndrome

Author(s): Khor A.

Source: Australian and New Zealand Journal of Obstetrics and Gynaecology; Sep 2018; vol. 58; p. 60

Publication Date: Sep 2018

Publication Type(s): Conference Abstract

Available at Australian and New Zealand Journal of Obstetrics and Gynaecology - from Wiley Online Library Science, Technology and Medicine Collection 2017

Abstract: Background: Mirror syndrome was first described by John Ballantyne in 1892. ZA is a 31-year-old female, G2P0 who was managed in a tertiary centre in NSW for fetal ascites and severe maternal oedema, and a clinical picture consistent with mirror syndrome. Case: ZA had an unremarkable pregnancy up to 24 weeks gestation. An ultrasound at 24 weeks revealed gross fetal ascites. ZA developed severe oedema of the upper and lower limbs at 28 weeks. ZA tested negative for cytomegalovirus, herpes simplex, parvovirus and toxoplasma and displayed no clinical or biochemical signs of pre eclampsia. There were no fetal structural abnormalities found on antenatal ultrasound. The fetal ascites was drained at 30 weeks. There were no significant findings in the cytology and infectious screen conducted on the ascitic fluid. ZA's membranes ruptured spontaneously at 31 weeks. She went into labour the next day and had a forceps assisted vaginal delivery of a female fetus. At birth, baby of ZA was found to have ascites, skin oedema and a chylothorax. The ascitic fluid was chylous on microscopy and biochemical investigations. Baby of ZA was diagnosed with intestinal lymphangiectasia. ZA's oedema resolved completely one week after delivery. Discussion: Mirror syndrome has been associated with a variety of causes of fetal hydrops including rhesus isoimmunisation, multiple pregnancies, viral infections, and fetal malformations. Our case is unique as it is the first case reported in literature where fetal intestinal lymphangiectasia resulted in fetal ascites and subsequently mirror syndrome.

Database: EMBASE

5. Small bowel high grade B-cell lymphoma in an adolescent girl with primary intestinal lymphangiectasia—feared association of a rare disease

Author(s): Teo Y.-M.

Source: Pediatric Radiology; May 2018; vol. 48 (no. 1)

Publication Date: May 2018

Publication Type(s): Conference Abstract

Available at Pediatric Radiology - from SpringerLink

Abstract: Purpose or Case Report: Intestinal lymphangiectasia is a rare disease characterised by dilated intestinal lacteals causing loss of lymph into the small bowel lumen and resultant hypoproteinemia, hypogammaglobulinemia, hypoalbuminemia and lymphopenia. The disease may occur as a primary/congenital form (primary idiopathic intestinal lymphangiectasia/Waldmann disease) or as a secondary form resulting from causes of lymphatic obstruction, such as tumor or fibrosis. Our case report describes the workup of an adolescent girl with known Primary Intestinal Lymphangiectasia (Waldmann disease) presenting with an acute history of abdominal pain and vomiting. We present her prior imaging leading to her initial diagnosis of Waldmann disease, as well as at the time of acute presentation, with imaging features raising suspicion for, and eventually leading to histo-pathologic confirmation of small bowel B-cell lymphoma. Via this case report poster, we hope to create awareness of this rare disease, as well as its feared association of lymphoma. Typical imaging features are presented and discussed.

Database: EMBASE
6. An approach to familial lymphoedema.

**Author(s):** Jones, Gabriela E; Mansour, Sahar

**Source:** Clinical medicine (London, England); Dec 2017; vol. 17 (no. 6); p. 552-557

**Publication Date:** Dec 2017

**Publication Type(s):** Journal Article Review

**PubMedID:** 29196357

Available at Clinical Medicine - from ProQuest (Hospital Premium Collection) - NHS Version

Available at Clinical Medicine - from Unpaywall

**Abstract:** Lymphoedema is the build-up of lymphatic fluid leading to swelling in the tissues. Most commonly it affects the peripheries. Diagnosis is based on clinical assessment and imaging with lymphoscintigraphy. Treatment is supportive with compression garments, massage, good skin hygiene and prompt use of antibiotics to avoid the complication of cellulitis. Most commonly, lymphoedema occurs as a result of damage to the lymphatic system following surgery, trauma, radiation or infection. However, it can be primary, often associated with a genetic defect that causes disruption to the development of the lymphatic system. Common genetic conditions associated with lymphoedema include Turner syndrome and Noonan syndrome; however, there are numerous others that can be classified based on their clinical presentation and associated features. Herein we discuss how to diagnose and classify the known primary lymphoedema conditions and how best to investigate and manage this group of patients.

**Database:** Medline

7. Primary intestinal lymphangiectasia: Multiple detector computed tomography findings after direct lymphangiography.

**Author(s):** Sun, Xiaoli; Shen, Wenbin; Chen, Xiaobai; Wen, Tingguo; Duan, Yongli; Wang, Rengui

**Source:** Journal of medical imaging and radiation oncology; Oct 2017; vol. 61 (no. 5); p. 607-613

**Publication Date:** Oct 2017

**Publication Type(s):** Journal Article

**PubMedID:** 28345300

Available at Journal of medical imaging and radiation oncology - from Wiley Online Library Science, Technology and Medicine Collection 2017

**Abstract:**

**INTRODUCTION** To analyse the findings of multiple detector computed tomography (MDCT) after direct lymphangiography in primary intestinal lymphangiectasia (PIL). **METHODS** Fifty-five patients with PIL were retrospectively reviewed. All patients underwent MDCT after direct lymphangiography. The pathologies of 16 patients were confirmed by surgery and the remaining 39 patients were confirmed by gastroendoscopy and/or capsule endoscopy. **RESULTS** After direct lymphangiography, MDCT found intra- and extraintestinal as well as lymphatic vessel abnormalities. Among the intra- and extraintestinal disorders, 49 patients had varying degrees of intestinal dilatation, 46 had small bowel wall thickening, 9 had pleural and pericardial effusions, 21 had ascites, 41 had mesenteric oedema, 20 had mesenteric nodules and 9 had abdominal lymphatic cysts. Features of lymphatic vessel abnormalities included intestinal trunk reflux (43.6%, n = 24), lumbar trunk reflux (89.1%, n = 49), pleural and pulmonary lymph reflux (14.5%, n = 8), pericardial and mediastinal lymph reflux (16.4%, n = 9), mediastinal and pulmonary lymph reflux (18.2%, n = 10), and thoracic duct outlet obstruction (90.9%, n = 50). **CONCLUSIONS** Multiple detector computed tomography after direct lymphangiography provides a safe and accurate examination method and is an excellent tool for the diagnosis of PIL.

**Author(s):** Martins, Cláudio Rodrigues; Gagnaire, Alice; Rostain, Florian; Lepage, Come

**Source:** Revista espanola de enfermedades digestivas : organo oficial de la Sociedad Espanola de Patologia Digestiva; May 2017; vol. 109 (no. 5); p. 385-388

**Publication Date:** May 2017

**Publication Type(s):** Case Reports Journal Article

**PubMedID:** 28376626

**Abstract:** Primary intestinal lymphangiectasia or Waldmann's disease is an uncommon cause of protein losing enteropathy with an unknown etiology and is usually diagnosed during childhood. It is characterized by dilation and leakage of intestinal lymph vessels leading to hypoalbuminemia, hypogammaglobulinemia and lymphopenia. Differential diagnosis should include erosive and non-erosive gastrointestinal disorders, conditions involving mesenteric lymphatic obstruction and cardiovascular disorders that increase central venous pressure. Since there are no accurate serological or radiological available tests, enteroscopy with histopathological examination based on intestinal biopsy specimens is currently the gold standard diagnostic modality of intestinal lymphangiectasia. We report a rare case of a primary intestinal lymphangiectasia in a 60-year-old Caucasian female who presented with asymptomatic hypoalbuminemia and hypogammaglobulinemia. After the diagnosis of a protein losing enteropathy, the patient underwent an enteroscopy and biopsies were taken, whose histological examination confirmed dilated intestinal lymphatics with broadened villi of the small bowel. Secondary causes of intestinal lymphangiectasia were excluded and the diagnosis of Waldmann's disease was recorded. The patient was put on a high-protein and low-fat diet with medium-chain triglyceride supplementation with improvement.

**Database:** Medline

9. Primary intestinal lymphangiectasia

**Author(s):** Siddeswari R.; Manohar S.; Reddy T.; Suryanarayana B.; Abhilash T.; Devender

**Source:** Journal, Indian Academy of Clinical Medicine; 2016; vol. 17 (no. 1); p. 60-63

**Publication Date:** 2016

**Publication Type(s):** Article

**Abstract:** Primary intestinal lymphangiectasia (PIL) is a rare disorder characterised by dilated intestinal lacteals resulting in lymph leakage into the small bowel lumen and responsible for protein-losing enteropathy leading to lymphopenia, hypoalbuminaemia and hypogammaglobulinaemia. PIL is generally diagnosed before 3 years of age but may be diagnosed in older patients. Prevalence is unknown. The main symptom is predominantly bilateral lower limb oedema. Oedema may be moderate-to-severe with anasarca and includes pleural effusion, pericarditis, or chylous ascites. This case of primary intestinal lymphangectasia was diagnosed at Osmania General Hospital, Afzalgunj, Hyderabad.Copyright © 2016, Indian Academy of Clinical Medicine. All rights reserved.

**Database:** EMBASE
10. Protein-losing enteropathy with intestinal lymphangiectasia in skeletal dysplasia with Lys650Met mutation.

**Author(s):** Yang, Chen; Dehner, Louis P

**Source:** American journal of medical genetics. Part A; Nov 2016; vol. 170 (no. 11); p. 2993-2997

**Publication Date:** Nov 2016

**Publication Type(s):** Case Reports Journal Article

**PubMedID:** 27214123

Available at [American journal of medical genetics. Part A](https://onlinelibrary.wiley.com/doi/10.1002/ajmg.a.36934) from Wiley Online Library Science, Technology and Medicine Collection 2017

**Abstract:** Protein-losing enteropathy is a primary or secondary manifestation of a group of conditions, and etiologies which are broadly divisible into those with mucosal injury on the basis of inflammatory and ulcerative conditions, mucosal injury without erosions or ulcerations, and lymphatic abnormalities. We describe the first case of protein-losing enteropathy in a pediatric patient, with severe skeletal dysplasia consistent with thanatophoric dysplasia type I and DNA analysis that revealed a c.1949A>T (p.Lys650Met) in exon 15 of the FGFR3 gene. She presented with protein-losing enteropathy in her 6th month. Post-mortem examination revealed lymphangiectasia in the small intestine. To our knowledge, this is the first report of intestinal lymphangiectasia as a complication of skeletal dysplasia resulting in severe protein-losing enteropathy. © 2016 Wiley Periodicals, Inc.

**Database:** Medline


**Author(s):** Alshikho, Mohamad J; Talas, Joud M; Noureldine, Salem I; Zazou, Saf; Addas, Aladdin; Kurabi, Haitham; Nasser, Mahmoud

**Source:** The American journal of case reports; Jul 2016; vol. 17; p. 512-522

**Publication Date:** Jul 2016

**Publication Type(s):** Case Reports Journal Article Review

**PubMedID:** 27440277

Available at [American Journal of Case Reports](https://www.americanjournalofcasereports.com) - from Europe PubMed Central - Open Access

**Abstract:** BACKGROUND Intestinal lymphangiectasia (IL) is a rare disease characterized by a dilatation of the intestinal lymphatics and loss of lymph fluid into the gastrointestinal tract leading to hypoproteinemia, edema, lymphocytopenia, hypogammaglobulinemia, and immunological abnormalities. Iron, calcium, and other serum components (e.g., lipids, fat soluble vitamins) may also be depleted. A literature search revealed more than 200 reported cases of IL. Herein, we report our observations of a patient diagnosed with IL; we also present our conclusion for our review of the published literature. CASE REPORT A 24-year-old male was admitted to Aleppo University Hospital with the complaints of abdominal pain, headache, arthralgia, fever, and rigoRs. His past medical history was remarkable for frequent episodes of diarrhea, recurrent infections, and swelling in the lower limbs. In addition, he had been hospitalized several times in non-academic hospitals due to edema in his legs, cellulitis, and recurrent infections. In the emergency department, a physical examination revealed a patient in distress. He was weak, dehydrated, pale, and had a high-grade fever. His lower extremities were edematous, swollen, and extremely tender to touch. The overlying skin was erythematous and warm. Moreover, the patient was tachycardic, tachypneic, and moderately hypotensive. The patient was resuscitated with IV fluids, and Tylenol was administered to bring the temperature down. Blood tests showed anemia and high levels of inflammatory markers. The patient’s white blood cell count was elevated with an obvious left shift. However,
subsequent investigations showed that the patient had IL. Suitable diet modification plans were applied as a long-term management plan. CONCLUSIONS IL is a rare disease of challenging nature due to its systematic effects and lack of comprehensive studies that can evaluate the effectiveness of specific treatments in a large cohort of patients. MCT (medium-chain triglyceride) oils and diet modification strategies are effective in reducing the loss of body proteins and in maintaining near-normal blood levels of immunoglobulins. However, octreotide and MCT oils had no proven role in shrinking edema in our patient.

**Database**: Medline

### 12. The lymphatic phenotype in Noonan and Cardiofaciocutaneous syndrome

**Author(s)**: Joyce S.; Brice G.; Short J.; Moore S.; Mansour S.; Gordon K.; Ostergaard P.; Mortimer P.; Nagaraja R.

**Source**: European Journal of Human Genetics; May 2016; vol. 24 (no. 5); p. 690-696

**Publication Date**: May 2016

**Publication Type(s)**: Article

**PubMedID**: 26242988

Available at [European Journal of Human Genetics](https://www.ncbi.nlm.nih.gov/pubmed/26242988) - from ProQuest (Hospital Premium Collection) - NHS Version

Available at [European Journal of Human Genetics](https://www.ncbi.nlm.nih.gov/pubmed/26242988) - from Europe PubMed Central - Open Access

**Abstract**: The RASopathies, which include Noonan syndrome (NS) and Cardiofaciocutaneous syndrome (CFC), are autosomal dominant disorders with genetic heterogeneity associated with germline mutations of genes in the Ras/mitogen-activated protein kinase (MAPK; RAS-MAP kinase) pathway. The conditions overlap and are characterised by facial dysmorphism, short stature and congenital heart disease. NS and CFC, in particular, are known to be associated with lymphatic problems, but this has not been well characterised to date. We describe 11 patients with Noonan or CFC syndrome with significant, persistent and progressive lymphatic dysplasia. The lymphatic disorders in Noonan and CFC syndrome are rare, but have a characteristic pattern with bilateral lower limb lymphoedema, genital swelling with chylous reflux and frequent systemic involvement, including intestinal lymphangiectasia and chylothoraces, which may be progressive. Lymphoscintigraphy demonstrates reflux and/or rerouting of lymphatic drainage associated with incompetent veins on the venous duplex scans.

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**Database**: EMBASE
A novel CCBE1 mutation leading to a mild form of Hennekam syndrome: Case report and review of the literature

Author(s): Frosk P.; Chodirker B.; El-Matary W.; Hanlon-Dearman A.; Rockman-Greenberg C.; Simard L.; Schwartzentruber J.; Majewski J.; Boycott K.; Friedman J.; Michaud J.; Bernier F.; Brudno M.; Fernandez B.; Knoppers B.; Samuels M.; Scherer S.

Source: BMC Medical Genetics; Apr 2015; vol. 16 (no. 1)

Publication Date: Apr 2015

Publication Type(s): Article

PubMedID: 25925991

Abstract: Background: Mutations in CCBE1 have been found to be responsible for a subset of families with autosomal recessive Hennekam syndrome. Hennekam syndrome is defined as the combination of generalized lymphatic dysplasia (i.e. lymphedema and lymphangiectasia), variable intellectual disability and characteristic dysmorphic features. The patient we describe here has a lymphatic dysplasia without intellectual disability or dysmorphism caused by mutation in CCBE1, highlighting the phenotypic variability that can be seen with abnormalities in this gene. Case presentation: Our patient is a 5 week old child of Pakistani descent who presented to our center with generalized edema, ascites, and hypoalbuminemia. She was diagnosed with a protein losing enteropathy secondary to segmental primary intestinal lymphangiectasia. As the generalized edema resolved, it became clear that she had mild persistent lymphedema in her hands and feet. No other abnormalities were noted on examination and development was unremarkable at 27 months of age. Given the suspected genetic etiology and the consanguinity in the family, we used a combination of SNP genotyping and exome sequencing to identify the underlying cause of her disease. We identified several large stretches of homozygosity in the patient that allowed us to sort the variants found in the patient’s exome to identify p.C98W in CCBE1 as the likely pathogenic variant. Conclusions: CCBE1 mutation analysis should be considered in all patients with unexplained lymphatic dysplasia even without the other features of classic Hennekam syndrome.

Database: EMBASE

Author(s): Ingle, Sachin B; Hinge Ingle, Chitra R

Source: World journal of clinical cases; Oct 2014; vol. 2 (no. 10); p. 528-533

Publication Date: Oct 2014

Publication Type(s): Journal Article Review

PubMedID: 25325063

Available at World journal of clinical cases - from Europe PubMed Central - Open Access

Abstract: Primary idiopathic intestinal lymphangiectasia is an unusual disease featured by the presence of dilated lymphatic channels which are located in the mucosa, submucosa or subserosa leading to protein loosing enteropathy. Most often affected were children and generally diagnosed before third year of life but may be rarely seen in adults too. Bilateral pitting oedema of lower limb is the main clinical manifestation mimicking the systemic disease and posing a real diagnostic dilemma to the clinicians to differentiate it from other common systemic diseases like Congestive cardiac failure, Nephrotic Syndrome, Protein Energy Malnutrition, etc. Diagnosis can be made on capsule endoscopy which can localise the lesion but unable to take biopsy samples. Thus, recently double-balloon enteroscopy and biopsy in combination can be used as an effective diagnostic tool to hit the correct diagnosis. Patients respond dramatically to diet constituting low long chain triglycerides and high protein content with supplements of medium chain triglyceride. So early diagnosis is important to prevent untoward complications related to disease or treatment for the sake of accurate pathological diagnosis.

Database: Medline

15. The lymphoscintigraphic manifestation of (99m)Tc-dextran lymphatic imaging in primary intestinal lymphangiectasia.

Author(s): Wen, Zhe; Tong, Guansheng; Liu, Yong; Meeks, Jacqui K; Ma, Daqing; Yang, Jigang

Source: Nuclear medicine communications; May 2014; vol. 35 (no. 5); p. 493-500

Publication Date: May 2014

Publication Type(s): Research Support, Non-u.s. Gov't Journal Article

PubMedID: 24488065

Available at Nuclear medicine communications - from Ovid (LWW Total Access Collection 2015 - Q1 with Neurology)

Abstract: PURPOSE The aim of this study was to analyze the imaging characteristics of (99m)Tc-dextran ((99m)Tc-DX) lymphatic imaging in the diagnosis of primary intestinal lymphangiectasia (PIL). MATERIALS AND METHODS Forty-one PIL patients were diagnosed as having PIL with the diagnosis being subsequently confirmed by laparotomy, endoscopy, biopsy, or capsule colonoscopy. Nineteen patients were male and 22 were female. A whole-body (99m)Tc-DX scan was performed at 10 min, 1 h, 3 h, and 6 h intervals after injection. The 10 min and 1 h postinjection intervals were considered the early phase, the 3 h postinjection interval was considered the middle phase, and the 6 h postinjection interval was considered the delayed phase. RESULTS The imaging characteristics of (99m)Tc-DX lymphatic imaging in PIL were of five different types: (i) presence of dynamic radioactivity in the intestine, associated with radioactivity moving from the small intestine to the ascending and transverse colon; (ii) presence of delayed dynamic radioactivity in the intestine, no radioactivity or little radioactivity distributing in the intestine in the early phase, or significant radioactivity distributing in the intestine in the delayed phase; (iii) radioactivity distributing in the intestine and abdominal cavity; (iv) radioactivity distributing only in the abdominal cavity with no radioactivity in the intestines; and (v) no radioactivity distributing in the intestine and abdominal
activity. CONCLUSION (99m)Tc-DX lymphatic imaging in PIL showed different imaging characteristics. Caution should be exercised in the diagnosis of PIL using lymphoscintigraphy. Lymphoscintigraphy is a safe and accurate examination method and is a significant diagnostic tool in the diagnosis of PIL.

Database: Medline

16. Primary intestinal lymphangiectasia: A rare cause of diarrhea in adults diagnosed by capsule endoscopy and double balloon enteroscopy

Author(s): Gupta V.; Ranjan P.; Kumar M.; Sachdeva M.
Source: Journal of Digestive Endoscopy; Apr 2014; vol. 5 (no. 2); p. 71-74
Publication Date: Apr 2014
Publication Type(s): Article
Available at Journal of Digestive Endoscopy - from Unpaywall

Abstract: Primary intestinal lymphangiectasia (PIL) or Waldmann's disease is a rare protein-losing enteropathy presenting with diarrhea. The etiology and prevalence of PIL remain unknown. <200 cases have been reported in the literature so far. Diagnosis of intestinal lymphangiectasia is difficult as there are no serological or radiological tests available. Small bowel imaging modalities like capsule endoscopy and double balloon enteroscopy have increased the chances of diagnosis of PIL due to direct visualization of small bowel. Diagnosis is confirmed by characteristic histopathological finding, which includes dilated intestinal lymphatics with broadened villi of the small bowel. We report a case of a patient with chronic diarrhea who was extensively worked up before he was finally diagnosed to have PIL involving the small bowel by performing balloon enteroscopy-guided biopsy.

Database: EMBASE

17. Ileal polypoid lymphangiectasia bleeding diagnosed and treated by double balloon enteroscopy.

Author(s): Park, Min Seon; Lee, Beom Jae; Gu, Dae Hoe; Pyo, Jeung-Hui; Kim, Kyeong Jin; Lee, Yun Ho; Joo, Moon Kyung; Park, Jong-Jae; Kim, Jae Seon; Bak, Young-Tae
Source: World journal of gastroenterology; Dec 2013; vol. 19 (no. 45); p. 8440-8444
Publication Date: Dec 2013
Publication Type(s): Case Reports Journal Article Review
PubMedID: 24363538
Available at World journal of gastroenterology - from Europe PubMed Central - Open Access

Abstract: Intestinal lymphangiectasia is a rare disease characterized by focal or diffuse dilated enteric lymphatics with impaired lymph drainage. It causes protein-losing enteropathy and may lead to gastrointestinal bleeding. Commonly, lymphangiectasia presents as whitish spots or specks. To our knowledge, small bowel bleeding resulting from polypoid intestinal lymphangiectasia has not been reported. Here, we report a rare case of active bleeding from the small bowel caused by polypoid lymphangiectasia with a review of the relevant literature. An 80-year-old woman was hospitalized for melena. Esophagogastroduodenoscopy could not identify the source of bleeding. Subsequent colonoscopy showed fresh bloody material gushing from the small bowel. An abdominal-pelvic contrast-enhanced computed tomography scan did not reveal any abnormal findings. Video capsule endoscopy showed evidence of active and recent bleeding in the ileum. To localize the bleeding site, we performed double balloon enteroscopy by the anal approach. A small, bleeding, polypoid lesion was found in the distal ileum and was successfully removed using endoscopic snare electrocautery.
A 10-year-old girl presented with recurrent small bowel diarrhea with features of malabsorption of fats and protein and growth retardation from the age of 3 years. This illness was not associated with fever, vomiting, abdominal pain, rash, arthralgia or urinary symptoms. She was second born to nonconsanguineous parents, with a birth weight of 3.5 kg and was diagnosed to have congenital lymphedema right upper limb in the newborn period. On examination she had gum hypertrophy, teeth overcrowding and prominent maxilla; lymphedema of right upper limb and bilateral pitting pedal edema. She had height 124 cm and weight 21 kg (both <3rd centile) and BMI 13.65 kg/m². She had hepatomegaly and shifting dullness. Other systems were normal. Hemoglobin 13 g/dL, WBC count - 8000 cells/cu.mm, differential count P83L14E3, ESR-10 mm/1st hour, platelet count 3.1 lakh/ cu.mm.; 24 hour proteinuria-nil; stool fat 10/hpf; LFT: bilirubin-1/0.4, OT/PT-22/18 IU/L, ALP-160, total protein/serum albumin-3/1.4 g/dL, INR-1.02, blood urea-18 mg/dL, creatinine-0.5 mg/dL; fasting lipid profile: total cholesterol-75 mg/dL, triglyceride-60 mg/dL; HDL-14 mg/dL and LDL-47mg/dL. Lymphoscintigraphy showed obstructive pattern of right upper limb lymphatic flow. USG abdomen showed ascites and edematous bowel loops. BMFT showed diffuse thickening of mucosal folds involving jejunum and ileum. The characteristic snowflake appearance of the duodenum was seen on upper gastrointestinal endoscopy. Duodenal biopsy showed dilated lymphatics which clinched the diagnosis of primary intestinal lymphangiectasia. She was given medium chain triglyceride based diet and albumin transfusions.

**Author(s):** Lai, Yu; Yu, Tao; Qiao, Xiao-Yu; Zhao, Li-Na; Chen, Qi-Kui

**Source:** Journal of medical case reports; Jan 2013; vol. 7 ; p. 19

**Publication Date:** Jan 2013

**PubMedID:** 23316917

**Abstract:**
UNLABELED
INTRODUCTION
Primary intestinal lymphangiectasia is a disorder characterized by exudative enteropathy resulting from morphologic abnormalities of the intestinal lymphatics. Intestinal lymphangiectasia can be primary or secondary, so the diagnosis of primary intestinal lymphangiectasia must first exclude the possibility of secondary intestinal lymphangiectasia. A double-balloon enteroscopy and biopsy, as well as the pathology can be used to confirm the diagnosis of intestinal lymphangiectasia. A polymeric diet containing medium-chain triglycerides and total parenteral nutrition may be a useful therapy.

CASE PRESENTATION
A 17-year-old girl of Mongoloid ethnicity was admitted to our hospital with a history of diarrhea and edema. She was diagnosed with protein-losing enteropathy caused by intestinal lymphangiectasia. This was confirmed by a double-balloon enteroscopy and multi-dot biopsy. After treatment with total parenteral nutrition in hospital, which was followed by a low-fat and medium-chain triglyceride diet at home, she was totally relieved of her symptoms.

CONCLUSION
Intestinal lymphangiectasia can be diagnosed with a double-balloon enteroscopy and multi-dot biopsy, as well as the pathology of small intestinal tissue showing edema of the submucosa and lymphangiectasia. Because intestinal lymphangiectasia can be primary or secondary, the diagnosis of primary intestinal lymphangiectasia must first exclude the possibility of secondary intestinal lymphangiectasia. A positive clinical response to the special diet therapy, namely a low-fat and medium-chain triglyceride diet, can further confirm the diagnosis of primary intestinal lymphangiectasia.

**Database:** Medline


**Author(s):** Alreheili, Khalid; AlMehaidib, Ali; Alsaleem, Khalid; Banemi, Mohammad; Aldekhail, Wajeeh; Al-Mayouf, Sulaiman M

**Source:** Annals of Saudi medicine; 2012; vol. 32 (no. 2); p. 206-208

**Publication Date:** 2012

**PubMedID:** 22366835

**Abstract:**
Infantile systemic hyalinosis (ISH) is a rare autosomal recessive disease. Typically, ISH patients present with progressive painful joint contractures, intractable diarrhea, hyperpigmented skin lesions, and peri-anal fleshy nodules. We report a case of a 19-month-old male child with atypical ISH presentation. His main clinical finding was protein-losing enteropathy due to intestinal lymphangiectasia. This report is intended to enhance awareness about the gastrointestinal tract presentation of ISH.

**Database:** Medline
21. Octreotide in Hennekam syndrome-associated intestinal lymphangiectasia

**Author(s):** Al Sinani S.; Al Rawahi Y.; Abdoon H.

**Source:** World Journal of Gastroenterology; 2012; vol. 18 (no. 43); p. 6333-6337

**Publication Date:** 2012

**Publication Type(s):** Article

**PubMedID:** 23180957

Available at [World journal of gastroenterology](http://worldjournal.com) - from Europe PubMed Central - Open Access

**Abstract:** A number of disorders have been described to cause protein losing enteropathy (PLE) in children. Primary intestinal lymphangiectasia (PIL) is one mechanism leading to PLE. Few syndromes are associated with PIL; Hennekam syndrome (HS) is one of them. The principal treatment for PIL is a high protein, low fat diet with medium chain triglycerides supplementation. Supportive therapy includes albumin infusion. Few publications have supported the use of octreotide to diminish protein loss and minimize hypoalbuminemia seen in PIL. There are no publications on the treatment of PIL with octreotide in patients with HS. We report two children with HS and PLE in which we used octreotide to decrease intestinal protein loss. In one patient, octreotide increased serum albumin to an acceptable level without further need for albumin infusions. The other patient responded more dramatically with near normal serum albumin levels and cessation of albumin infusions. In achieving a good response to octreotide in both patients, we add to the publications supporting the use of octreotide in PIL and suggest that octreotide should be tried in patients with PIL secondary to HS. To the best of our knowledge, this is the first case report on the use of octreotide in HS associated PIL.

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**Database:** EMBASE

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22. Primary intestinal lymphangiectasia diagnosed by capsule endoscopy and double balloon enteroscopy.

**Author(s):** Oh, Tak Geun; Chung, Joo Won; Kim, Hee Man; Han, Seok-Joo; Lee, Jin Sung; Park, Jung Yeob; Song, Si Young

**Source:** World journal of gastrointestinal endoscopy; Nov 2011; vol. 3 (no. 11); p. 235-240

**Publication Date:** Nov 2011

**Publication Type(s):** Journal Article

**PubMedID:** 22110841

Available at [World journal of gastrointestinal endoscopy](http://worldjournal.com) - from Europe PubMed Central - Open Access

**Abstract:** Primary intestinal lymphangiectasia (PIL) is a rare disorder characterized by dilated intestinal lymphatics and the development of protein-losing enteropathy. Patients with PIL develop hypoalbuminemia, hypocalcemia, lymphopenia and hypogammaglobulinemia, and present with bilateral lower limb edema, fatigue, abdominal pain and diarrhea. Endoscopy reveals diffusely elongated, circumferential and polypoid mucosae covered with whitish enlarged villi, all of which indicate intestinal lymphangiectasia. Diagnosis is confirmed by characteristic tissue pathology, which includes dilated intestinal lymphatics with diffusely swollen mucosa and enlarged villi. The prevalence of PIL has increased since the introduction of capsule endoscopy. The etiology and prevalence of PIL remain unknown. Some studies have reported that several genes and regulatory molecules for lymphangiogenesis are related to PIL. We report the case of a patient with PIL involving the entire small bowel that was confirmed by capsule endoscopy and double-balloon enteroscopy-guided tissue pathology who carried a deletion on chromosome 4q25. The relationship between this deletion on chromosome 4 and PIL remains to be investigated.
23. Intestinal lymphangiectasia in a patient with autoimmune polyglandular syndrome Type III
Author(s): Choudhury B.K.; Saikia U.K.; Sarma D.; Choudhury B.N.; Choudhury S.D.; Saharia D.; Saikia M.
Source: Journal of Association of Physicians of India; Nov 2011; vol. 59 (no. 11); p. 729-731
Publication Date: Nov 2011
Publication Type(s): Article
PubMedID: 22616341
Abstract: Autoimmune polyglandular syndromes (APS) comprise a wide clinical spectrum of autoimmune disorders. APS is divided into Type I, Type II, Type III and Type IV depending upon the pattern of disease combination. Chronic diarrhoea is one of the many manifestations of APS and many aetiological factors have been suggested for it. Apart from the established aetiological factors, intestinal lymphangiectasia may be responsible for chronic diarrhea in some cases. Intestinal lymphangiectasia has been reported in Type I APS. We report a case of Type III APS with hypocalcaemia and hypothyroidism who had chronic diarrhea of long duration and was finally diagnosed to have intestinal lymphangiectasia. © JAPI.

24. Long-Standing Intestinal Lymphangiectasia Detected by Double-Balloon Enteroscopy
Author(s): Imbesi V.; Ciccocioppo R.; Corazza G.R.
Source: Clinical Gastroenterology and Hepatology; Sep 2011; vol. 9 (no. 9)
Publication Date: Sep 2011
Publication Type(s): Article
PubMedID: 21596158
Database: EMBASE

25. Intestinal lymphangiectasia associated with recurrence of histiocytosis X
Author(s): Hui C.K.
Source: Singapore Medical Journal; Sep 2011; vol. 52 (no. 9)
Publication Date: Sep 2011
Publication Type(s): Article
PubMedID: 21947159
Abstract: Intestinal lymphangiectasia may occur as a primary congenital disorder or a secondary disorder. Secondary lymphangiectasia could be associated with diseases such as abdominal carcinoma, retroperitoneal fibrosis or chronic pancreatitis. This is the first reported case of intestinal lymphangiectasia associated with recurrent histiocytosis X. This case report illustrates the need for more prospective, well-designed studies to determine the natural history and outcome of intestinal lymphangiectasia in the duodenum. Hopefully, these studies will also help clinicians identify which group of patients with intestinal lymphangiectasia in the duodenum is more likely to have a secondary cause.
Database: EMBASE
26. Primary intestinal lymphangiectasia: four case reports and a review of the literature.

Author(s): Wen, Jie; Tang, Qingya; Wu, Jiang; Wang, Ying; Cai, Wei

Source: Digestive diseases and sciences; Dec 2010; vol. 55 (no. 12); p. 3466-3472

Publication Date: Dec 2010

Publication Type(s): Case Reports Journal Article Review

PubMedID: 20198428

Available at Digestive diseases and sciences - from ProQuest (Hospital Premium Collection) - NHS Version

Available at Digestive diseases and sciences - from SpringerLink

Abstract:BACKGROUND Primary intestinal lymphangiectasia (PIL) is a rare digestive disease and most articles on this condition are isolated case reports. AIM Our purpose is to investigate the clinical characteristics, therapeutic management, and outcome of PIL through case studies. METHODS We conducted a retrospective analysis and obtained detailed clinical information for four PIL patients treated at our institution. A MEDLINE database search was also performed using the search term "intestinal lymphangiectasia" and all pertinent literature was carefully reviewed. RESULTS Four children treated in our department showed elevated IgE and a good response to diet intervention. After reviewing the literature, we conducted statistical analysis on the basis of all the cases, with a total of 84 cases. Thirty-eight cases have been reported with diet treatment, 24 (63%) of whom showed apparent improvement in clinical symptoms and laboratory parameters. Four cases (5%) had a malignant transformation of lymphoma, and the average time from PIL onset to lymphoma diagnosis was 31 years (range, 19-45 years). No difference was observed regarding the presence of major clinical manifestations among children and adults. Diet intervention in children was more effective than that in adults. CONCLUSIONS Diet intervention is the cornerstone of PIL medical management, which was found to be more effective in children than in adults. Early diagnosis and treatment of IL is of great importance for effective diet therapy. An elevated IgE level should be monitored periodically since it could be an indication of malignant transformation-lymphoma.

Database: Medline

27. Castleman's disease associated with a cerebellar chordoid meningioma and intestinal lymphangiectasia


Source: Child's Nervous System; Nov 2010; vol. 26 (no. 11); p. 1647-1652

Publication Date: Nov 2010

Publication Type(s): Article

PubMedID: 20567835

Available at Child's nervous system : ChNS : official journal of the International Society for Pediatric Neurosurgery - from SpringerLink

Abstract: Castleman's disease (CD) is a rare nonneoplastic lymphoproliferative disorder of unknown etiology. It is characterized by enlarged hyperplastic lymph nodes, usually presenting as a localized mass. Although an intracranial location is very uncommon, it should be considered in the differential diagnosis of a chordoid meningioma. We describe a pediatric case of CD with a cerebellar chordoid meningioma and intestinal lymphangiectasia. © 2010 Springer-Verlag.

Database: EMBASE
28. Primary intestinal lymphangiectasia (Waldmann's disease).

**Author(s):** Vignes, Stéphane; Bellanger, Jérôme

**Source:** Orphanet journal of rare diseases; Feb 2008; vol. 3 ; p. 5

**Publication Date:** Feb 2008

**Publication Type(s):** Journal Article Review

**PubMedID:** 18294365

Available at [Orphanet journal of rare diseases](https://orphanetjournalofrarediseases.biomedcentral.com) - from BioMed Central

**Abstract:** Primary intestinal lymphangiectasia (PIL) is 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. PIL is generally diagnosed before 3 years of age but may be diagnosed in older patients. Prevalence is unknown. The main symptom is predominantly bilateral lower limb edema. Edema may be moderate to severe with anasarca and includes pleural effusion, pericarditis or chylous ascites. Fatigue, abdominal pain, weight loss, inability to gain weight, moderate diarrhea or fat-soluble vitamin deficiencies due to malabsorption may also be present. In some patients, limb lymphedema is associated with PIL and is difficult to distinguish lymphedema from edema. Exudative enteropathy is confirmed by the elevated 24-h stool alpha1-antitrypsin clearance. Etiology remains unknown. Very rare familial cases of PIL have been reported. Diagnosis is confirmed by endoscopic observation of intestinal lymphangiectasia with the corresponding histology of intestinal biopsy specimens. Videocapsule endoscopy may be useful when endoscopic findings are not contributive. Differential diagnosis includes constrictive pericarditis, intestinal lymphoma, Whipple's disease, Crohn's disease, intestinal tuberculosis, sarcoidosis or systemic sclerosis. Several B-cell lymphomas confined to the gastrointestinal tract (stomach, jejunum, midgut, ileum) or with extra-intestinal localizations were reported in PIL patients. A low-fat diet associated with medium-chain triglyceride supplementation is the cornerstone of PIL medical management. The absence of fat in the diet prevents chyle engorgement of the intestinal lymphatic vessels thereby preventing their rupture with its ensuing lymph loss. Medium-chain triglycerides are absorbed directly into the portal venous circulation and avoid lacteal overloading. Other inconsistently effective treatments have been proposed for PIL patients, such as antiplasmin, octreotide or corticosteroids. Surgical small-bowel resection is useful in the rare cases with segmental and localized intestinal lymphangiectasia. The need for dietary control appears to be permanent, because clinical and biochemical findings reappear after low-fat diet withdrawal. PIL outcome may be severe even life-threatening when malignant complications or serous effusion(s) occur.

**Database:** Medline
29. Primary intestinal lymphangiectasia as a component of autoimmune polyglandular syndrome type I: A report of 2 cases

**Author(s):** Makharia G.K.; Tandon R.K.; Tandon N.; Stephen N.D.J.R.; Gupta S.D.

**Source:** Indian Journal of Gastroenterology; 2007; vol. 26 (no. 6); p. 293-295

**Publication Date:** 2007

**Publication Type(s):** Article

**PubMedID:** 18431016

**Abstract:** Chronic diarrhea and steatorrhea occur frequently in patients with autoimmune polyglandular syndrome (APS) type I. Intestinal lymphangiectasia has been reported earlier as a cause of steatorrhea in a young girl with APS Type I. We describe 2 patients with APS Type I who were found to have intestinal lymphangiectasia, one of whom had symptomatic protein-losing enteropathy.

**Database:** EMBASE

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30. Intestinal lymphangiectasia with protein-losing enteropathy in Waldenstrom macroglobulinemia.

**Author(s):** Pratz, Keith W; Dingli, David; Smyrk, Thomas C; Lust, John A

**Source:** Medicine; Jul 2007; vol. 86 (no. 4); p. 210-214

**Publication Date:** Jul 2007

**Publication Type(s):** Case Reports Journal Article Review

**PubMedID:** 17632262

Available at Medicine - from Ovid (Journals @ Ovid) - Remote Access

Available at Medicine - from Unpaywall

**Abstract:** Gastrointestinal complications of Waldenstrom macroglobulinemia (WM) are unusual but often treatable. We report a case of WM associated with significant gastrointestinal involvement manifest as chronic diarrhea with protein-losing enteropathy and recurrent venous thromboses. Small bowel biopsy was negative for amyloidosis but revealed intestinal lymphangiectasia with deposition of monoclonal IgM. The patient was treated with cyclophosphamide, vincristine, and prednisone with rapid and complete resolution of the peripheral edema and diarrhea. We follow the case report with a retrospective analysis of patients with WM and gastrointestinal symptoms seen at our institution, and review the available literature on this unusual association. An increased awareness of the gastrointestinal manifestations of WM may help to explain and to treat the chronic, debilitating, and potentially life-threatening symptoms in patients with this lymphoproliferative disorder.

**Database:** Medline
31. A primary intestinal lymphangiectasia patient diagnosed by capsule endoscopy and confirmed at surgery: A case report

Author(s): Fang Y.-H.; Zhang B.-L.; Chen C.-X.; Wu J.-G.

Source: World Journal of Gastroenterology; Apr 2007; vol. 13 (no. 15); p. 2263-2265

Publication Date: Apr 2007

Publication Type(s): Article

PubMedID: 17465517

Abstract: Intestinal lymphangiectasia (IL) is a rare disease characterized by dilated lymphatic vessels in the intestinal wall and small bowel mesentery which induce loss of protein and lymphocytes into bowel lumen. Because it most often occurs in the intestine and cannot be detected by upper gastroendoscopy or colonoscopy, and the value of common image examinations such as X-ray and computerized tomography (CT) are limited, the diagnosis of IL is difficult, usually needing the help of surgery. Capsule endoscopy is useful in diagnosing intestinal diseases, such as IL. We here report a case of IL in a female patient who was admitted for the complaint of recurrent edema accompanied with diarrhea and abdominal pain over the last twenty years, and aggravated ten days ago. She was diagnosed by M2A capsule endoscopy as a primary IL and confirmed by surgical and pathological examination. © 2007 The WJG Press. All rights reserved.

Database: EMBASE

32. Use of capsule endoscopy in children with primary intestinal lymphangiectasia

Author(s): Rivet C.; Dumortier J.; Budin C.; Bouvier R.; Ponchon T.; Lachaux A.; Lapalus M.-G.; Le Gall C.

Source: Gastrointestinal Endoscopy; Oct 2006; vol. 64 (no. 4); p. 649-650

Publication Date: Oct 2006

Publication Type(s): Article

PubMedID: 16996365

Database: EMBASE
33. Videocapsule endoscopy is useful for the diagnosis of intestinal lymphangiectasia.

**Author(s):** Chamouard, P; Nehme-Schuster, H; Simler, J-M; Finck, G; Baumann, R; Pasquali, J-L

**Source:** Digestive and liver disease : official journal of the Italian Society of Gastroenterology and the Italian Association for the Study of the Liver; Sep 2006; vol. 38 (no. 9); p. 699-703

**Publication Date:** Sep 2006

**Publication Type(s):** Case Reports Journal Article

**PubMedID:** 16527553

**Abstract:** We study two authentic cases of protein-losing enteropathy, the diagnosis of which was facilitated using Given M2A videocapsule endoscopy. The first case corresponded to a primary intestinal lymphangiectasia confirmed by jejunal biopsies and the second one to a protein-losing enteropathy with lymphatic abnormalities secondary to a chronic constrictive pericarditis. In the first case, the mucosa of jejunal presented with a diffuse oedematous aspect, whitish villi, white curved lines probably related to submucosal dilated lymphatics and lacteal juice. In the second case, capsule endoscopy showed oedematous aspect of jejunal mucosa associated with white curved lines similar to those observed in the first case. Videocapsule endoscopy is useful in cases of protein-losing enteropathy to identify presence of intestinal lymphangiectasia and to specify their localisation after ruling out other disorders liable to induce protein-losing gastrointestinal syndrome.

**Database:** Medline

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34. Capsule endoscopy revealing small-intestinal lymphangiectasia and GI stromal tumor polyps in neurofibromatosis type 1

**Author(s):** Calabrese C.; Pironi L.; Di Febo G.

**Source:** Gastrointestinal Endoscopy; Jul 2006; vol. 64 (no. 1); p. 130-131

**Publication Date:** Jul 2006

**Publication Type(s):** Article

**PubMedID:** 16813823

**Database:** EMBASE
35. Familial pachydermoperiostosis in association with protein-losing enteropathy

**Author(s):** Sethuraman G.; Malhotra A.K.; Khaitan B.K.; Sharma V.K.; Kumar R.; Makharia G.K.; Vinod B.N.; Sharma S.K.; Goswami R.; Bandhu S.

**Source:** Clinical and Experimental Dermatology; Jun 2006; vol. 31 (no. 4); p. 531-534

**Publication Date:** Jun 2006

**Publication Type(s):** Article

**PubMedID:** 16716156

Available at Clinical and Experimental Dermatology - from Wiley Online Library Science, Technology and Medicine Collection 2017

**Abstract:** In this report we describe a rare association of pachydermoperiostosis with protein-losing enteropathy (PLE) in a family of three brothers. The first brother had the complete form of pachydermoperiostosis along with PLE. The second brother had the 'forme fruste' of pachydermoperiostosis, with minimal skin changes, bony abnormalities and PLE, which was due to intestinal lymphangiectasia. The third brother had an incomplete form of pachydermoperiostosis without evidence of PLE. To our knowledge, the association of pachydermoperiostosis with PLE due to intestinal lymphangiectasia has not been reported previously. © 2006 Blackwell Publishing Ltd.

**Database:** EMBASE

36. Capsule endoscopy in the management of a patient with a rare syndrome

**Author(s):** Danielsson A.; Toth E.; Thorlacius H.

**Source:** Gut; Feb 2006; vol. 55 (no. 2); p. 196

**Publication Date:** Feb 2006

**Publication Type(s):** Article

**PubMedID:** 16407384

Available at Gut - from BMJ Journals - NHS

**Database:** EMBASE

37. Noonan syndrome and related disorders

**Author(s):** Noonan J.A.

**Source:** Progress in Pediatric Cardiology; Jul 2005; vol. 20 (no. 2); p. 177-185

**Publication Date:** Jul 2005

**Publication Type(s):** Article

**Abstract:** Noonan syndrome is a common multiple malformation syndrome seen in children with congenital heart disease. Recently, a mutation in the PTPN11 gene was found to be present in about 50% of individuals with Noonan syndrome. Over 80% of these patients have some form of congenital heart disease with pulmonary stenosis often associated with a dysplastic valve being, by far, the most common lesion. Hypertrophic cardiomyopathy occurs in 20-30%. Characteristic facies, chest deformity, short stature, undescended testes in the male and learning disabilities comprise the Noonan phenotype but there is wide phenotypic variation and a changing phenotype with age. This phenotype is noted in several other syndromes which share similar cardiac defects. These include LEOPARD, neurofibromatosis, Noonan syndrome, cardio-facio-cutaneous syndrome and Costello syndrome. A definitive diagnosis is particularly difficult in infancy. © 2005 Elsevier Ireland Ltd. All rights reserved.
38. Different patterns of lymphoscintigraphic findings in patients with intestinal lymphangiectasia

**Author(s):** So Y.; Chung J.-K.; Lee D.S.; Lee M.C.; Seo J.K.; Ko J.S.; Kim J.Y.

**Source:** Nuclear Medicine Communications; 2001; vol. 22 (no. 11); p. 1249-1254

**Publication Date:** 2001

**Publication Type(s):** Article

**PubMedID:** 11606892

Available at [Nuclear medicine communications](https://journals.lww.com/nuclearmedcommunications) - from Ovid (Journals @ Ovid) - Remote Access

**Abstract:** Technetium-99m antimony sulfide colloid lymphoscintigraphy conveniently demonstrates intestinal leakage of lymph in patients with intestinal lymphangiectasia. However, we found no intestinal radioactivity in some patients. We evaluated lymphoscintigraphic findings and compared them with clinical data. Technetium-99m antimony colloid lymphoscintigraphy was performed in 12 patients (age, 8.9 +/- 6.4 years; male : female = 8 : 4) with histologically proven intestinal lymphangiectasia. After subcutaneous injection of 103.6 MBq of technetium-99m antimony colloid into the webs of both feet, sequential abdominal images were obtained up to 24 h post-injection. Four patients underwent technetium-99m methylene diphosphonate bone scintigraphy. Patients were divided into two groups according to the presence or absence of intestinal radioactivity. Five showed intestinal activity (Group 1), but seven did not (Group 2). No Group I patient had a history of ascites, while all Group 2 patients had ascites as the initial manifestation. Serum total protein and albumin levels were significantly lower in Group I patients than in Group 2 patients. In three Group I patients, technetium-99m methylene diphosphonate bone scintigraphy revealed intestinal radioactivity, while in one Group 2 patient this was not found. We observed two types of lymphoscintigraphic pattern in patients with intestinal lymphangiectasia. To clarify the exact pathophysiology, further study is required. © 2001 Lippincott Williams & Wilkins.

**Database:** EMBASE

**Author(s):** van Haelst, M M; Hoogeboom, J; Galjaard, R J; Kleijer, W J; den Hollander, N S; de Krijger, R R; Hennekam, R C; Niermeijer, M F

**Source:** American journal of medical genetics; Nov 2001; vol. 104 (no. 1); p. 65-68

**Publication Date:** Nov 2001

**Publication Type(s):** Case Reports Journal Article

**PubMedID:** 11746030

Available at [American journal of medical genetics](https://onlinelibrary.wiley.com/doi/abs/10.1002/ajmg.20421) - from Wiley Online Library Science, Technology and Medicine Collection 2017

**Abstract:** We report on a sibship with protein-losing enteropathy related to intestinal lymphangiectasia, a peculiar face, and genital anomalies. The parents are distantly related and from Dutch ancestry. The first patient was born with a protein-losing enteropathy, craniofacial anomalies, and renal defects. At 1 year of age, she died of severe complications of the protein-losing enteropathy and respiratory distress. Her brother was a cytogenetically normal male fetus identified by prenatal ultrasound at 19 weeks with similar anomalies. The pregnancy was terminated at 20 weeks. Autopsy showed müllerian duct remnants. These cases seem to confirm the Urioste syndrome [Urioste et al., 1993: Am J Med Genet 47:494-503]. Although it was previously only reported in 46,XY individuals, this report of a consanguineous family with an affected sibship of both sexes suggests it to be an autosomal recessive entity.

**Database:** Medline

40. Primary intestinal lymphangiectasia: a rare disease in the differential diagnosis of acute abdomen.

**Author(s):** Uğuralp, S; Mutus, M; Kutlu, O; Cetin, S; Baysal, T; Mizrak, B

**Source:** Journal of pediatric gastroenterology and nutrition; Oct 2001; vol. 33 (no. 4); p. 508-510

**Publication Date:** Oct 2001

**Publication Type(s):** Case Reports Journal Article

**PubMedID:** 11698774

Available at [Journal of Pediatric Gastroenterology and Nutrition](https://www.journals.ovid.com/doi/10.1097/00005123-200110000-00011) - from Free Medical Journals . com

Available at [Journal of Pediatric Gastroenterology and Nutrition](https://www.ovid.com/doi/10.1097/00005123-200110000-00011) - from Ovid (LWW Total Access Collection 2015 - Q1 with Neurology)

**Database:** Medline
41. Computed tomography after lymphangiography in the diagnosis of intestinal lymphangiectasia with protein-losing enteropathy in Noonan's syndrome

**Author(s):** Keberle M.; Jenett M.; Hahn D.; Mork H.; Scheurlen M.

**Source:** European Radiology; 2000; vol. 10 (no. 10); p. 1591-1593

**Publication Date:** 2000

**Publication Type(s):** Article

**PubMedID:** 11044930

Available at [European radiology](http://www.european-radiology.com) - from ProQuest (Hospital Premium Collection) - NHS Version

Available at [European radiology](http://www.european-radiology.com) - from SpringerLink

**Abstract:** Noonan's syndrome is a rare congenital disorder that may be associated with abnormalities in the lymphatic drainage. In this case of a 21-year-old man CT after bipedal lymphangiography confirmed the diagnosis of intestinal lymphangiectasy causing protein-losing enteropathy in Noonan's syndrome by showing contrast-enhanced abnormal lymphatic vessels in the mesentery and the intestinal wall. Because of the benefit of diet in case of intestinal involvement, we recommend a thorough documentation of the lymphatic drainage with lymphangiography followed by CT, if clinical signs of lymphatic dysplasia, such as pleural effusions, lymphedema, or hypoproteinemia are present.

**Database:** EMBASE

42. Early death in two sisters with Hennekam syndrome

**Author(s):** Scarcella A.; De Lucia A.; Pasquariello M.B.; Gambardella P.

**Source:** American Journal of Medical Genetics; Jun 2000; vol. 93 (no. 3); p. 181-183

**Publication Date:** Jun 2000

**Publication Type(s):** Article

**PubMedID:** 10925377

Available at [American journal of medical genetics](http://www.americanjournalofmedicalgenetics.com) - from Wiley Online Library Science, Technology and Medicine Collection 2017

**Abstract:** We report on two sisters with facial anomalies, protein-losing enteropathy, and intestinal lymphangiectasia consistent with the diagnosis of Hennekam syndrome. Both patients had a number of other anomalies not previously described in this autosomal recessive disorder, i.e., primary hypothyroidism, hypertrophic pyloric stenosis, and an early fatal outcome. These cases support the autosomal recessive transmission and the expansion of the phenotype of the Hennekam syndrome.

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**Database:** EMBASE
43. Characteristic endoscopic features of intestinal lymphangiectasia: correlation with histological findings.

**Author(s):** Aoyagi, K; Iida, M; Yao, T; Matsui, T; Okada, M; Oh, K; Fujishima, M

**Source:** Hepato-gastroenterology; 1997; vol. 44 (no. 13); p. 133-138

**Publication Date:** 1997

**Publication Type(s):** Journal Article

**PubMedID:** 9058131

**Abstract:**
Six patients with histologically proven intestinal lymphangiectasia were evaluated for the endoscopic features. White villi and/or spots, previously reported as endoscopic findings, were seen. Furthermore, white nodules and submucosal elevations with or without white mucosa were observed. All patients demonstrated the appearance of submucosal elevations. The four characteristic endoscopic features were correlated with histological findings. It is considered that these endoscopic features are of value for the diagnosis of intestinal lymphangiectasia in patients with protein-losing enteropathy.

**Database:** Medline

44. The CT halo sign: A new finding in intestinal lymphangiectasia

**Author(s):** Stevens R.L.; Jones B.; Fishman E.K.

**Source:** Journal of Computer Assisted Tomography; 1997; vol. 21 (no. 6); p. 1005-1007

**Publication Date:** 1997

**Publication Type(s):** Article

**PubMedID:** 9386299

Available at [Journal of computer assisted tomography](#) - from Ovid (LWW Total Access Collection 2015 - Q1 with Neurology)

**Abstract:**
A 'halo sign' has been described in patients with Crohn disease, ulcerative colitis, radiation enteritis, ischemic colitis, and pseudomembranous colitis. This sign is characterized by an inner ring of low CT attenuation surrounded by a higher attenuation outer ring. We present a patient with primary intestinal lymphangiectasia in whom CT demonstrated a halo sign correlated with mucosal biopsy.

**Database:** EMBASE
45. Intestinal lymphangiectasia: Value of Tc-99m dextran lymphoscintigraphy

**Author(s):** Yueh T.-C.; Pui M.H.; Zeng S.-Q.

**Source:** Clinical Nuclear Medicine; 1997; vol. 22 (no. 10); p. 695-696

**Publication Date:** 1997

**Publication Type(s):** Article

**PubMedID:** 9343726

Abstract: Intestinal lymphangiectasia is a common cause of protein-losing enteropathy characterized by diarrhea, generalized edema, enteric protein loss, hypoproteinemia, and lymphopenia. Diagnosis is based on demonstration of enteric protein loss and characteristic small bowel mucosal histology. Various imaging modalities including barium studies, computed tomography, and lymphangiography have had limited clinical use. The authors report a case of intestinal lymphangiectasia in which Tc-99m dextran lymphoscintigraphy played a significant role in the patient management.

**Database:** EMBASE

46. Intestinal lymphangiectasia in a patient with Zellweger cerebrohepatorenal syndrome

**Author(s):** Erdem G.; Oran O.; Kotiloglu E.; Kale G.; Topcu M.; Renda Y.

**Source:** American Journal of Medical Genetics; 1995; vol. 58 (no. 2); p. 152-154

**Publication Date:** 1995

**Publication Type(s):** Article

**PubMedID:** 8533807

Abstract: Zellweger cerebrohepatorenal syndrome (ZWCHRS) is an autosomal-recessive disease, characterized by the absence or profound deficiency of peroxisomes. We report a case of ZWCHRS with intestinal lymphangiectasia, observed as an autopsy finding. This combination is previously unreported.

**Database:** EMBASE

47. Endoscopic features of long-standing primary intestinal lymphangiectasia.

**Author(s):** Salomons, H A; Kramer, P; Nikulasson, S; Schroy, P C

**Source:** Gastrointestinal endoscopy; May 1995; vol. 41 (no. 5); p. 516-518

**Publication Date:** May 1995

**Publication Type(s):** Case Reports Journal Article

**PubMedID:** 7615236

**Database:** Medline
48. Intestinal lymphangiectasia in a patient with autoimmune polyglandular disease type I and steatorrhea

Author(s): Bereket A.; Lowenheim M.; Blethen S.L.; Wilson T.A.; Kane P.
Source: Journal of Clinical Endocrinology and Metabolism; Mar 1995; vol. 80 (no. 3); p. 933-935
Publication Date: Mar 1995
Publication Type(s): Article
PubMedID: 7883852

Abstract: Steatorrhea is seen in 18-24% of patients with autoimmune polyglandular disease (APD) type 1. The etiology and pathophysiology of the steatorrhea in this disease are unknown. We present a patient with APD type 1 and steatorrhea in whom biopsies revealed intestinal lymphangiectasia. This association has not been previously described. Intestinal lymphangiectasia may explain the steatorrhea in some patients with APD type 1. As blind intestinal biopsies may miss areas of intestinal lymphangiectasia, endoscopically directed intestinal biopsies should be included in the evaluation of steatorrhea in APD type 1.

Database: EMBASE

49. Intestinal lymphangiectasia: value of double-contrast radiographic study.

Author(s): Aoyagi, K; Iida, M; Yao, T; Matsui, T; Okada, M; Fujishima, M
Source: Clinical radiology; Nov 1994; vol. 49 (no. 11); p. 814-819
Publication Date: Nov 1994
Publication Type(s): Journal Article
PubMedID: 7955851

Abstract: Intestinal lymphangiectasia is a disorder presenting as enteric protein loss through the dilated lymphatics without mucosal ulceration. To determine the double-contrast radiographic features and to assess the significance of them, five patients with intestinal lymphangiectasia were examined using single- and double-contrast small intestinal studies. The double-contrast examinations demonstrated clearly the main radiographic findings of smooth nodular protrusions, thickening of the mucosal folds, with no evidence of mucosal ulceration. Compared with the single-contrast study, smooth nodular protrusions were seen more often and in more widespread segments, particularly in the duodenum, on double-contrast study. Thickening of the mucosal folds was revealed similarly by both methods. Double-contrast study appears to be worthwhile to demonstrate the characteristic radiographic findings of this disease.

Database: Medline
Intestinal lymphangiectasia: evaluation by CT and scintigraphy.

Author(s): Puri, A S; Aggarwal, R; Gupta, R K; Sewatkar, A B; Gambhir, S; Tandon, P; Choudhuri, G

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Abstract: Intestinal lymphangiectasia caused severe diarrhea and generalized edema in a 40-year-old man. The diagnosis was established by clinical, laboratory, and duodenal biopsy findings. The abnormalities detected on computed tomography (CT) and scintigraphy using 99mTc human serum albumin are herein described and pertinent literature is briefly reviewed.

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