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DIAGNOSIS, TREATMENT AND PREVENTION OF CROHN'S DISEASE, PEUTZ–JEGHERS SYNDROME AND ULCERATIVE COLITIS: A COMPREHENSIVE CLINICAL REVIEW

Gayrat Kurbanov,
Jamshidjon Urinboyev,
Mirziyod O'tkirov,
Nodira Toshnazarova,
Nizomiddin Baratov,
Yusup Utegenov.

Abstract: *Inflammatory bowel diseases (IBDs) and hereditary gastrointestinal syndromes represent a significant medical and social burden due to chronic progression, complications, and reduced quality of life. This article reviews three clinically important conditions: Crohn's disease, Peutz–Jeghers Syndrome (PJS), and Ulcerative Colitis (UC). The paper discusses etiological factors, pathogenesis, clinical manifestations, diagnostic protocols, modern therapeutic strategies, and prevention measures. Early recognition and multidisciplinary approaches are emphasized to reduce morbidity and improve long-term outcomes.*

Key words: *Crohn's Disease, Peutz–Jeghers Syndrome and Ulcerative Colitis*

Introduction

Chronic inflammatory and hereditary disorders affecting the gastrointestinal tract have become increasingly common worldwide. Crohn's disease and Ulcerative Colitis, classified as inflammatory bowel diseases (IBDs), are characterized by chronic immune-mediated inflammation of the gastrointestinal mucosa. Their incidence has been rising, especially in industrialized nations, suggesting environmental influences such as diet, microbiome imbalance, and lifestyle factors.

Peutz–Jeghers Syndrome (PJS), by contrast, is not an inflammatory disease but a hereditary polyposis syndrome caused by mutations of the **STK11/LKB1**

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tumor suppressor gene. It presents with mucocutaneous pigmentation and gastrointestinal hamartomatous polyps, carrying increased cancer risk. Understanding the detailed mechanisms of these disorders is crucial for developing improved treatment strategies and preventive health policies.

Etiology and Pathogenesis

Crohn's Disease

Crohn's disease involves a multifactorial etiology:

- **Genetic predisposition:** Variants in the *NOD2/CARD15* gene.
- **Environmental triggers:** Smoking, diet rich in refined sugars, low fiber intake.
- **Immune dysregulation:** Hyperactivation of T-helper cells leading to transmural granulomatous inflammation.
- **Microbiota dysbiosis:** Altered gut microbial community structure.

Pathologically, Crohn's disease can affect any gastrointestinal segment from mouth to anus, producing **skip lesions** and deep ulcerations, often resulting in **fistula formation and strictures**.

Peutz–Jeghers Syndrome (PJS)

PJS is inherited in an autosomal dominant pattern due to **mutations in the STK11/LKB1 gene**, which plays a regulatory role in cellular metabolism and DNA repair. Dysfunction leads to:

- Hamartomatous polyp formation in the gastrointestinal tract.
- Mucocutaneous hyperpigmentation (particularly on lips and oral mucosa).
- Increased lifetime risk for cancers of the pancreas, breast, colon, and reproductive organs.

Ulcerative Colitis (UC)

UC primarily affects the colon and rectum, causing **continuous mucosal inflammation**. Contributing factors include:

- Genetic susceptibility (HLA-associated).
- Abnormal immune reaction to intestinal microbiota.

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- Epithelial barrier dysfunction.

Unlike Crohn's disease, UC affects only **the mucosal layer** and does not produce deep fissures or fistulas.

Clinical Manifestations

Condition	Key Clinical Symptoms	Systemic Complications
Crohn's Disease	Chronic abdominal pain, diarrhea, weight loss, perianal fistulas	Malnutrition, anemia, bowel obstruction
Peutz–Jeghers Syndrome	Recurrent abdominal pain, gastrointestinal bleeding, mucosal pigmentation, polyp prolapse	Intussusception, increased malignancy risk
Ulcerative Colitis	Bloody diarrhea, rectal bleeding, tenesmus, abdominal cramping	Toxic megacolon, colorectal cancer

Extraintestinal symptoms common to IBDs include arthritis, uveitis, erythema nodosum, and sclerosing cholangitis.

Diagnosis

Laboratory Tests

- CBC (to assess anemia)
- ESR and CRP (inflammation markers)
- Fecal calprotectin (bowel-specific inflammation marker)

Endoscopy

- **Colonoscopy with biopsy** is essential for diagnosis.
- Crohn's disease may show patchy "cobblestone" mucosa.
- UC reveals **continuous** mucosal ulceration starting from the rectum.

Imaging

- **MR/CT enterography** is recommended to evaluate intestinal wall thickness and complications.
- In PJS, capsule endoscopy may detect small-intestinal polyps.

Genetic Testing

- STK11 gene sequencing confirms Peutz–Jeghers Syndrome.

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Treatment Strategies

Pharmacologic Therapy

Drug Class	Application	Examples
5-ASA derivatives	Mild to moderate UC	Mesalamine, Sulfasalazine
Corticosteroids	Acute flares	Prednisone, Budesonide
Immunomodulators	Steroid-sparing maintenance	Azathioprine, Methotrexate
Biologic agents	Severe, refractory disease	Infliximab, Adalimumab, Vedolizumab, Ustekinumab
Antibiotics	Fistulizing Crohn's	Metronidazole, Ciprofloxacin

Dietary and Lifestyle Management

- High-protein, high-calorie diet during flares.
- Avoid alcohol, spicy foods, and highly processed meals.
- Smoking cessation is critical in Crohn's disease.

Endoscopic and Surgical Treatment

- **Crohn's disease:** Resection for strictures or fistulas.
- **UC:** Proctocolectomy considered for refractory disease or cancer risk.
- **PJS:** Endoscopic polypectomy to prevent intussusception.

Prevention and Long-Term Management

- Regular colonoscopy surveillance (every 1–3 years depending on condition severity).
- Probiotic use may help stabilize microbiota. Genetic counseling is recommended for PJS patients.Patient education focusing on symptom monitoring and medication adherence.

Conclusion

- Crohn's disease, Peutz–Jeghers Syndrome, and Ulcerative Colitis require careful long-term clinical management. Advancements in biologic therapies, molecular genetic research, and microbiome science are contributing to more effective and personalized treatment approaches. Early recognition, regular

endoscopic monitoring, and multidisciplinary care significantly improve outcomes and quality of life.

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