Case report on Management and Complication of Peutz Jegher Syndrome

Seema kolhe1, Pratiksha Munjewar2, Aniket Pathade3
1Nursing tutor, F.N.T.C.N., Sawangi (M) Wardha, Maharashtra, India
2Department of Medical-Surgical Nursing, Smt. Radhikabai Meghe Memorial College of Nursing, Datta Meghe Institute of Medical Sciences, Sawangi, Wardha, Maharashtra.
3Research Scientist, Jawaharlal Nehru Medical College, Datta Meghe Institute of Medical Sciences, Sawangi, Wardha, Maharashtra.
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Abstract

Introduction: Infrequent and characterized by mucocutaneous pigmentation, gastrointestinal polyposis, and increased risk of malignancy, Peutz-Jeghers syndrome is an autosomal dominant genetic condition. The usual perioral macules and patches that are colored in the buccal mucosa are present in 90% of patients, and numerous but not continuously contiguous lesions, primarily in the gastrointestinal (G.I.) tract, with rarely more than 20 hamartomatous polyps, are the characteristics of Peutz-Jeghers syndrome (P.J.S.). Present complaints and Investigation:- Suddenly felt sick to my stomach and started to puke. Abdominal pain, Black stools, repeated vomiting, weight loss of 5 kg in 15 days, loss of appetite, and Intussusception were discovered during examinations in the upper small bowel. A thorough study of the gastrointestinal tract was conducted. Endoscopically, the enormous polyps of the stomach and duodenum were discovered and biopsied. Resulting of bleeding due to the G.I. polyps, she became anemic. There were polyps. Later removed endoscopically while being sedated, C.T.’s Abdomen and pelvis found multiple mildly enhanced polyoidal lesions notes attached to the pyloric part of stomach and duodenum proximal jejunal loops, similar lesions of Ascending colon, transverse colon, and advice colonoscopy. History: The patient was admitted in 2015 And Operated on 26/12/2015, the year Jejunoileal intussusception. The primary diagnosis, therapeutic intervention, and outcomes: After physical examination and investigation, Hamartomatous polyp (P-J Secondary Intussuseception ) Conclusions: The following primary complaints were the reason for the patient's hospital admission. Upon entry to the hospital, the patient's main complaint was that Peutz Jegher Syndrome was identified as the patient's condition once all investigations were complete. The patient now needs appropriate medical attention and quality nursing care.

Keywords: Polyps, hamartomatous, Peutz, Jeghers, syndrome.

INTRODUCTION

An autosomal dominant genetic condition called Peutz-Jeghers syndrome is characterized by the emergence of benign, which includes gastrointestinal polyposis, mucocutaneous pigmentation, and an increased risk of cancer. Mucocutaneous pigmentation, gastrointestinal polyposis, and a higher chance of developing cancer. The LKB1 tumor suppressor gene mutations that can induce hamartomatous polyps to form at an early age, which can lead to several consequences, such as acute intestinal blockage, anemia, and stomach pain. The Peutz-Jeghers syndrome (PJS) is an inherited gastrointestinal disease with hamartomatous polyps that are autosomal dominant and mucocutaneous pigmented lesions. Between 1 in 8300 to 1 in 280 000 people have PJS. Certain cancers are more likely to develop in PJS sufferers (gastrointestinal, pancreatic, lung, breast, uterine, ovarian, and testicular tumors) PJS prevalence is estimated to range from 1 in 8300 to 1 in 280 000 people. PJS makes patients more likely to develop certain cancers (gastrointestinal, pancreatic, lung, breast, uterine, ovarian, and testicular tumors). Patients with PJS frequently experience consequences such as bleeding, blockage, and Intussusception. The small intestine can be examined and treated using a double-balloon enteroscopy (D.B.E.). The requirement for small intestinal resection that causes short bowel syndrome and multiple urgent procedures might be avoided with polypectomy utilizing D.B.E. In PJS patients, prevention and complete small intestinal polypectomy is considered the gold standard. Before the advent of D.B.E., individuals with JPS could only receive endoscopic care by intraoperative enteroscopy (I.O.E.). Before the D.B.E. era, intraoperative enteroscopy (I.O.E.) was the only option for endoscopic treatment of patients with PJS.
Patient information: Upon admission to the female surgery ward, a 55-year-old married woman complained primarily of black spots (also known as mucocutaneous pigmentation). These spots can form on the mouth, lips, eyes, nose, hands, and feet, among other body areas. Anemia, stomach ache, pain from small bowel intussusceptions, anorexia, gastrointestinal bleeding, nausea, and vomiting

The primary concern and symptoms of the patient: Present case was seen in the O.P.D. of a hospital on the day in question and complained of abdominal pain, black feces, frequent vomiting, dark spots (also known as mucocutaneous pigmentation), and other symptoms. The mouth, lips, eyes, nose, hands, feet, anus, gastrointestinal bleeding, anemia, stomach pain, and Intussusception of small bowel pain are a few of the bodily regions where these spots may manifest.

Medical and family and psycho-social history: an individual with Peutz-Jeghers syndrome two years back. The current example is from a nuclear family and comes from a middle-class family. She had good mental health. He maintained solid interactions with family members and was the date, time, and place-oriented.

Relevant past intervention with outcomes: History of Peutz-Jeghers syndrome two years back. She had therapy for the condition for which she was hospitalized for ten days after an investigation was conducted, and his results were positive.

Physical examination and clinical findings: Unhealthy physical condition, poor build; the patient is 165 cm tall and weighs 60 kg. Her vital signs are typical, and she had excellent peripheral perfusion and a 96 beats per minute tachycardia. You're healthy if your blood pressure is 130/76 mm Hg. She only responded neurologically to pain and was sensitive to it on both sides. A critical care unit admission is necessary for reactive mydriasis to receive closer monitoring. When the temperature increases above 38°C. The epiphany In the blood chemistry, osmolality, genetic counseling, and genetic testing, the sodium level was 253 mOsm/kg, and the blood sugar level was 7.4 mmol/L. The osmolality and urine salt levels were 404 mOsm/kg and 134 mmol/L, respectively. Methamphetamine was found in the urine. The physical examination allows for the identification of melanocytic macules. Endoscopy, x-ray analysis, and wireless capsule endoscopy can all detect polyps. By microscopic examination, polyps are categorized as PJS polyps. Family background of PJS. Her immune system is compromised.

Timeline: Patient was seen in the hospital's outpatient department with the primary complaint of dark spots (also known as mucocutaneous pigmentation), which can affect the hands, feet, nose, eyes, mouth, and lips. Additional symptoms included stomach pain, gastrointestinal bleeding, anemia, nausea, vomiting, and treatment for a dietary solute deficiency. He undertook all medical procedures.

Diagnostic Assessment: During physical examination and Endoscopically, the most extensive polyps of the stomach and duodenum were discovered and biopsied. Resulting from bleeding due to the G.I. polyps, she became anemic. There were polyps. Later removed endoscopically while being sedated. C.T. Abdomen and pelvis of found to have multiple mildly enhanced polypoidal lesions notes attached to the pyloric part of stomach and duodenum proximal jejunal loops, similar lesions of Ascending colon, transverse colon, and advice colonoscopy. A doctor diagnosed a case of Peutz-Jeghers syndrome.

Diagnostic Evaluation:

Diagnosis: After a physical examination and conducting more research, a diagnosis of Peutz-Jeghers syndrome is made.

Therapeutic intervention:

There is currently no treatment for Peutz-Jeghers syndrome (PJS.). Patients are subjected to ongoing organ surveillance to detect malignancy and stop any complications from the polyps. According to specific research, closely monitored people may not require urgent small intestinal surgery and may not have cancer. To achieve this, polyps must be removed before they grow large enough to restrict airflow or develop into cancer.

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Surgical intervention: there are two primary modalities in diagnosing and treating small bowel hamartomas: intra-operative enteroscopy (I.O.E.) and double-balloon enteroscopy (D.B.E.).

Identifying and treating small bowel hamartomas can be done using two primary techniques: intra-operative enteroscopy (I.O.E.) and double-balloon enteroscopy (D.B.E.). With the help of a novel enteroscopy technique called D.B.E., practically all patients can have their jejunum and ileum examined and treated. The apparatus comprises a 145-cm over-tube and a 200-cm enteroscope with soft latex balloons attached at the tips. The endoscope can be introduced longer without creating extraneous loops of the intestine when these balloons are used to grip the intestinal wall.

Laparotomy (or laparoscopy) and endoscopy are combined in I.O.E. It enables manipulation to achieve complete small intestinal visualization and almost complete polyp removal during an endoscopic procedure.

Followup and Outcomes:

Clinical and patient assessment outcomes: patient condition was improved.

Necessary follow up diagnostic and other test results: The goal of illness prevention is to hold off on developing any signs and symptoms such as diminished mouth, lips, eyes, nose, hands, and feet, anus, gastrointestinal bleeding, anemia, and stomach pain. After a month, the doctor advised a follow-up visit and suggested blood work to see how far the condition had progressed.

Intervention adherence and tolerability: The patient regularly took all prescribed medications. He also carried out the dietician's instructions. Thiamine, multivitamins, folic acid solution, and protein-rich supplementation were recommended to the dietician. His adherence to the intervention was satisfactory.

Discussion:

The presence of a hamartoma in conjunction with two of the following three symptoms—mucocutaneous pigmentation, small-bowel polyposis, or a family history of J.P.S.—is necessary for a clinical diagnosis of PJS. Pigments often occur in the first year of life, although they sometimes fade or disappear as people age. Lip and buccal mucosal pigmentation is a crucial characteristic that aids early diagnosis. Regarding this patient, the gastrointestinal system has hamartomatous polyps and lips patches. However, a few people show mucocutaneous pigmentation, which is incomplete P.J.S. In P.J.S., polyps frequently appear in adolescence and the early stages of adulthood. It develops during the first ten years of life, and the majority of people start to exhibit symptoms between the ages of 10 and 30. Typically, polyp-related problems such as abdominal pain, bowel obstruction, Intussusception, and overt or covert gastrointestinal bleeding cause patients to appear in the first decade.

Our patient experienced sporadic upper intestine obstruction and anemia symptoms for two years. The cause of P.J.S. is uncertain. The serine/threonine kinase-coding STK11 gene, found on chromosome 8, is likely to be the source of P.J.S. Gynecomastia is a multifactorial condition that mostly depends on the ratio of free testosterone to free estradiol. Since androgen biosynthesis results in the production of androgens from steroid precursors, an increase in the substrate, aromatase activity, or both might result in more than endogenous estrogen. An increase in estrogen levels is the most common cause of imbalance. The most common causes of gynecomastia include endocrinopathies, neoplasm-producing estrogens, human chorionic gonadotropin or aromatase, hyperaromatase syndrome, and a very tiny number of idiopathic prepubertal illnesses. According to estimates, common causes include cirrhosis (8%), primary hypogonadism (8%), testicular tumors (3%), secondary hypogonadism (2%), hyperthyroidism (1.5%), and renal disease (1%). Testicular cancer, which is connected to both germ cell and non-germ cell tumors, accounts for one to one and a half percent of all male malignancies.
Conclusion:

The patient's main complaint upon admission to the hospital was. After a thorough inquiry, it was determined that the patient had Peutz-Jeghers syndrome, a common illness frequently misdiagnosed, underappreciated, and treated improperly. Patients with hypernephromatous conditions require planned interventions, which are scarce. Everything has been covered in detail.

To sum up, to raise awareness of Peutz-Jeghers syndrome, simplify treatment, and enhance prognosis P.J.S. is an autosomal dominant disorder caused by a germline mutation of the serine-threonine kinase that is characterized by mucocutaneous pigmentation and gastrointestinal hamartomatous polyps. Patients with P.J.S. are more likely to have cancer or have their benign polyps turn into malignant ones in their gastrointestinal system and other organs18-31.

REFERENCES


