Crohn's Disease of the Vulva Occurring in Siblings

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Occasionally, patients with gastrointestinal Crohn's disease (CD) develop granulomatous skin lesions at sites remote from the gastrointestinal tract, separated from other ulcerations by normal skin, a phenomenon that has been referred to as metastatic cutaneous CD. Although metastatic CD of the vulval region has been often reported in English literature, we could not find such cases with family history. We report a case of vulval CD occurring in siblings. (Ann Dermatol 13(2) 129~131, 2001).

Key Words : Siblings, Vulval Crohn's disease

Crohn's disease (CD) may affect any part of the gastrointestinal tract, from the mouth to the anus. The skin can also be affected by this granulomatous disease either through a direct extension of the intestinal lesions or with manifestation of the intestinal lesions or with manifestations that are totally separated from the intestine. The latter kind of lesion is surrounded by healthy skin, is not connected to the intestine and is known in the literature as metastatic CD. We report a case of vulval CD occurring in siblings.

CASE REPORT

A 26-year-old Korean woman presented to our department for evaluation of erythematous swelling of the right labia majora. She was diagnosed 3 years previously with CD by colonoscopic study. Since the age of 13 years, she suffered from intermittent but constant diarrhea. She had undergone a fistulectomy for rectal fistulas 6 years ago; however, these lesions relapsed 3 months later. Numerous small masses developed on the perianal region since that time, and the lesions were getting worse. A painful erythematous swelling of the right labia majora developed 1 year ago. Her family history revealed that she had a younger sister who had been diagnosed with metastatic CD on the vulva. On physical examination, the patient was a woman of 35 kg (below the third percentile for her age) and 152 cm (the tenth percentile for her age). The right labia majora was affected by erythematous swelling and the perianal region showed numerous cobblestone-appearing masses (Fig 1). No enlarged lymph nodes were discovered. Laboratory studies revealed leukocytosis with a white blood cell count of 12,000/mm³ (normal range: 4,000-11,000/mm³), hemoglobin 10.1 g/dL, hematocrit 32.7%, erythrocyte sedimentation rate (ESR) 94 mm/hr (normal range: 0-20 mm/hr). Pelvic computed tomography showed spiculations and mild wall thickening of the terminal ileum, diffuse thickening of the rectal wall, and perianal fistulas. Abdominal sonography showed thickening of the distal rectal wall and rectal fistulas. Radiologic evaluation revealed no evidence of direct extension from the rectum to the vulva. Punch biopsy specimen taken from the vulval lesion showed non-caseating granulomas composed of epithelioid histiocytes and lymphocytes (Fig 2, 3).

Special stains for acid-fast bacilli, fungal, or bacterial organisms were negative. A biopsy of the perianal mass displayed mild chronic inflammatory cell infiltrations with fibrovascular stroma. Based on these findings, a diagnosis of vulval CD was made. She was treated with prednisolone 40 mg daily, mesalazine 500 mg three
Fig. 1. The right labia majora shows edema and thickening with erythematous to brownish color. The lesion revealed a firm consistency on palpation. Numerous 1 to 4 cm sized cobblestone appearing masses with fistulas are noted on the perianal region. The rectal exploration was impossible due to stenosis.

Fig. 2. Biopsy specimen from the vulva shows non-caseating granulomas. (Hematoxylin-eosin stain; original magnification, × 40).

Fig. 3. Higher power view of area in box in Fig 2. Note that granuloma is composed of epithelioid histiocytes and lymphocytes. (Hematoxylin-eosin stain; original magnification, × 400).

times daily for 1 month with marked improvement of vulval & perianal lesion.

DISCUSSION

CD is a chronic relapsing, multisystemic and granulomatous inflammatory disorder, and the genetic, infectious, psychologic, and environmental factors appear to play an etiologic role. Cutaneous involvement of CD is observed in 22-44% of cases. The dermatologic lesions of CD may be classified into four categories: (1) direct extension, in continuity with gastrointestinal disease, leading to the perineal ulceration, ischiorectal abscesses, and fistulas; (2) noncontinuous extraintestinal involvement of the vulva, submammary area, penis, abdominal wall, and/or extremities, also known as metastatic CD; (3) noncontiguous vascular reactions without the typical histologic features of CD such as erythema nodosum or pyoderma gangrenosum; (4) skin lesions associated with malabsorption such as vitamin or trace mineral deficiency. This case showed direct extension of CD such as perianal fistulas and fibrovascular masses with perifistular inflammation. In addition, the patient demonstrates involvement of the vulva by metastatic CD, because the vulva is distant from gastrointestinal tract and there was no evidence of direct extension from the rectum to the vulva on radiologic evaluation. Metastatic lesions of CD may be found on the axilla, groin, anterior vulva, retroauricular folds, submammary area, abdomen, back, forearm, lower extremities, and genitalia of male and female. Vulval CD is an uncommon extraintestinal manifestation of CD and has
been mostly reported as isolated cases in the first report of 1965. Vulval involvement with CD typically presents with erythema and edema. Other clinical manifestations of vulval CD include ulceration, labial skin tag, and labial mass. Vulval CD generally occurs in association with large bowel disease. In 25% of patients, it precedes any intestinal manifestation by up to 15 years. In this case, gastrointestinal symptom of CD preceded the vulval CD. Histopathologic findings of vulval CD have shown non-caseating granulomas with Langhans’ giant cells in most of the cases. The current case showed non-caseating granulomas without Langhans’ giant cells. This patient exhibited familial occurrence of vulval CD. The younger sister of the patient was reported in 1992. Approximately it has been reported that one sixth of patients with gastrointestinal CD have a family history. Patients with familial gastrointestinal CD are characterized by an early age onset with more extensive disease and less exclusively colonic involvement. This patient was diagnosed as CD at age 23. However, age onset of CD may be earlier, considering the history of chronic diarrhea for 13 years and perianal fistulas from 6 years ago. Her younger sister was diagnosed as CD at age 16 with a 3-year history of diarrhea. First degree relatives of patients with gastrointestinal CD, as compared with the general population, have a 10-fold increase in the risk of having the same disease as the patient. Especially, siblings with gastrointestinal CD increase the risk of developing the same disease as much as 30-fold. In one study, siblings were concordant for the type of gastrointestinal CD in 81.6% of the affected sibling pairs, for extent in 76.0%, and for extraintestinal manifestations including erythema nodosum, pyoderma gangrenosum, arthritis and uveitis in 83.6%. They were diagnosed within 10 years of each other. A precise genetic basis for CD has not yet been elucidated, but a complex multigenic hereditary pattern that predisposes individuals to inflammatory bowel disease has been proposed. We could not find reports of vulval CD with family history in spite of our extensive literature research. However, it might be presumed that familial vulval CD is not rare when considering the frequency of family history of CD.

REFERENCES