

Hidradenitis suppurativa in a long-lived patient with trisomy 13



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Key words: hidradenitis suppurativa; trisomy 13.

INTRODUCTION

Hidradenitis suppurativa (HS) is a chronic, recurrent, purulent skin disease that usually affects the axillary, inguinal, and/or anogenital regions. HS typically presents as inflammatory or noninflammatory nodules, fistulae, abscesses, and scars. The detailed causes of HS have not been elucidated, but both genetic and environmental factors are thought to be involved.

Trisomy 13 (T13) is a congenital anomaly syndrome based on the full or partial duplication of chromosome 13. Patients with T13 exhibit multiple severe malformations, including anomalies of the central nervous, cardiovascular, and urogenital systems. The 5-year survival rate of patients with T13 is 9.7%, and the median survival is 5 days.¹ However, there are a few reported cases of patients with T13 achieving long-term survival and reaching puberty. This case report describes a long-lived patient with T13 and HS.

CASE REPORT

The patient was a 12-year-old Japanese boy who was born by natural delivery at 34 weeks and 2 days of gestation. At birth, he weighed 1918 g, was 42.5 cm long, and had a head circumference of 32.1 cm. He had multiple anomalies, including a scalp defect, low pinnae, cleft lip and palate, polydactyly, and patent ductus arteriosus. The patent ductus arteriosus closed naturally at 3 days of age, and no other cardiac malformations were identified. Chromosome analysis performed by G-banding found that the chromosomal karyotype was full T13. At 1 year of age, the patient had bilateral cryptorchidism and vesicoureteral reflux; orchiopexy was performed, and intermittent urethral catheterization was started. At the age of 2 years and

Abbreviations used:

HS: hidradenitis suppurativa
T13: trisomy 13

7 months, he had sudden heart failure, and dilated cardiomyopathy was diagnosed. Tracheostomy was performed, and home oxygen therapy was commenced. At 11 years and 8 months of age, laryngotracheal separation was performed to prevent repeated aspiration pneumonia. There have been no recurrent systemic infections suggestive of immunodeficiency after laryngotracheal separation.

From approximately 10 years of age, the patient had multiple abscesses that discharged hemorrhagic purulent discharge in the axillae (Fig 1), inguinal folds, lower abdomen (Fig 2), perineum, buttocks (Fig 3), neck, and periauricular region (Fig 4). The abscesses on the right side of the neck were connected to the upper back, whereas those on the right axilla spread to the anterior chest. The fistulae were filled with soft, immature, granulation tissue. Atrophic and hypertrophic scars were also present, but there were no comedones or acne. A bacterial culture from the purulent discharge was negative, and systemic antibiotic therapy was ineffective. The formation of abscesses and fistulae gradually progressed for 2 years, producing hemorrhagic purulent discharge without distinct flares. The patient has received repeated treatment with surgical or manual drainage and local injection of corticosteroids into the granulation tissue.

DISCUSSION

A literature search found only 4 reported cases of T13 with extensive comedones, severe acne, and

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Fig 1. The right axillary region has multiple abscesses with purulent discharge. Soft granulation tissue is also present.



Fig 2. Fistulae are present from the lower abdomen to the periphery of the penis, and there is purulent discharge from several places.

abscess formation consistent with HS; 3 of these patients had full trisomy, whereas one had partial



Fig 3. There is an abscess on the perineum, with hypertrophic scarring and fistulae extending to the buttocks.



Fig 4. An abscess with atrophic scarring and fistulae under the ear.

trisomy.²⁻⁵ One of the patients with full trisomy was a 2-year-old girl with numerous cystic and comedone-like lesions on the cheeks, chin, and perineum, and age-appropriate levels of steroid and sex hormones.³ The other 2 patients with full trisomy were a 17-year old boy⁴ and a 15-year-old boy.⁵ The patient with partial trisomy was a 20-year-old man.³ In the 4 cases involving males, including our case, the skin lesions worsened at the age of puberty.

In the previous case reports, of the 2 boys with full trisomy, abscess and fistula formation occurred on the face, posterior auricular region, neck, axillae, chest, groin, and abdominal wall.^{4,5} Furthermore, they both had comedones with cysts on their faces.^{4,5} These findings suggest that these were cases of

severe acne that developed with HS. In contrast, our patient has had no facial lesions or comedones, and therefore seems to have T13 with HS without acne.

Inflammatory lesions reminiscent of HS have also been reported in 2 patients with mosaic T13.^{6,7} In one case, cytogenetic analysis of blood lymphocytes showed a normal karyotype, whereas a karyotype 47, XX +mar. rev. ish enh (13)(q22qter) was found in fibroblasts from a lesional skin biopsy.⁶ In the other case, fibroblasts derived from lesional skin showed a 13q21.33-q34 duplication.⁷

The pathogenesis of HS has not been fully clarified. However, the development of HS is thought to be affected by genetic, hormonal, bacterial, and environmental factors. Regarding the genetic factors involved in HS, several mutations in γ -secretase genes have been identified in some affected families.⁸ However, mutations in γ -secretase genes are only present in 29% of familial cases,⁹ and it is likely that not all genetic factors have been identified yet. In addition, a mutation of the gap junction β gene on chromosome 13 that encodes connexin 26 associated with keratitis-ichthyosis-deafness syndrome might contribute to the follicular occlusion triad consisting of HS, severe cystic acne, and dissecting cellulitis.¹⁰

Our case and previous reports of HS associated with T13 suggest that unknown abnormalities on chromosome 13 may be involved in the development of HS. As long-term survival of patients with T13 is rare, the accumulation of case data is extremely difficult. Therefore, our case adds

important information that may help to elucidate the genetic etiologies of HS.

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