

## Alopecia Areata and Vitiligo in a Female with Alfi Syndrome (9p Deletion Syndrome)

Raquel Rozner, BS; Danilo C Delcampo, MD<sup>\*</sup>; Marian Russo, MD

**\*Danilo C Delcampo, MD**

Department of Dermatology, New York Medical College, USA

Email: danilo.delcampo@gmail.com

### Abstract

Alfi Syndrome (also known as 9p deletion syndrome or 9-minus-P syndrome) is a rare chromosomal disorder that involves the deletion of the short arm of chromosome 9, resulting in a loss of genetic material. This syndrome is characterized by clinical findings such as developmental delay, malformations of craniofacial region, structural malformations of heart and abdomen, bony abnormalities, and other characteristic phenotypic features. This case highlights the unique presentation of vitiligo and alopecia areata in a young girl with Alfi Syndrome that has not been previously described in the literature.

### Keywords

genetic syndrome, chromosomal disorder, alopecia areata, vitiligo, 9p

### Introduction

Alfi Syndrome also known as 9p deletion syndrome or 9-minus-P syndrome) is a rare chromosomal disorder. A “9p deletion” indicates that the short arm of chromosome 9s has broken. The breakpoint of the deletion can be almost anywhere in the short arm, but majority have been described in the region called 9p22. People diagnosed a gene deletion in this region are said to have Alfi syndrome, which is named after the scientist who first identified a group of people with similar features and this genetic defect [1].

Alfi syndrome is characterized by developmental delay (primarily speech and language delay), “developmental delay, malformations of craniofacial region, and bony abnormalities [1]. Other features of affected people include globalized hypotonia, seizures, GERD, constipation, cardiac defects such as VSD and ASD, elongated digits, precocious puberty, and frequent ear infections [2, 4].

This case highlights the unique presentation of vitiligo and alopecia areata that have not been previously described with this syndrome. To date, there have been over 150 reports of Alfi syndrome [2]. However, to our knowledge, there are no other reports of alopecia areata and vitiligo described in the literature of patients with Alfi syndrome.

### Case Presentation

An 11-year-old female with past medical history of deafness since birth, juvenile glaucoma, repaired atrial septal defect, and congenital umbilical hernia presented to dermatology clinic with

3-month history of patches of hair loss on scalp and 1-year history of white patches on the body. On physical examination, the scalp had alopecic patches in the occipital central region (Figure 1). Further examination included bilateral multiple depigmented patches on the knees, achilles tendons, thorax, abdomen, and ankles (Figure 2). Extensive other clinical findings included extreme scoliosis, ocular hypertelorism, gingival hyperplasia, and developmental delay (Figure 3 and Figure 4). Of importance, the patient has no family history for vitiligo, diabetes, alopecia areata, thyroid disorders, or other autoimmune conditions. As a result of her extensive pathology, the patient was sent for genetic testing.

Genetic testing via whole blood confirmed the diagnosis of Alfi syndrome. Her karyotype was determined as 46 XX del (9) (p22p24.3), with a deletion at the critical region on the 9<sup>th</sup> chromosome. Due to financial constraints, further genetic testing was not feasible at the time.

Biopsies from the depigmented patches on the patient's knees and thorax were sent for histopathology. The biopsy revealed absence of melanocytes consistent with the diagnosis of vitiligo. The alopecic patches on her scalp were identified as alopecia areata due to clinical findings of exclamation-mark hair shafts noted on dermoscopy.

The patient was treated for alopecia areata with monthly intralesional corticosteroid injections and tacrolimus ointment. The alopecic patches regrew normal terminal hairs within 3 months of therapy. The patient was also treated for vitiligo with topical tacrolimus ointment to her face and body as well as corticosteroid treatment on the depigmented patches. The patient was advised to wear sun block when outdoors and to receive annual full body skin screenings. The patient's alopecia areata and vitiligo were improving over the course of 6 months. The patient continues to follow dermatology clinic for observation and regular full body skin examinations.

## Discussion

Alfi syndrome presents as a constellation of clinical findings supported with the genetic testing of particular genetic deletions. Numerous reports have described the association between partial 9p monosomy deletions and phenotypic changes including developmental delay (primarily speech and language delay), “keel shaped” forehead, choanal atresia, ocular hypertelorism, flattened nasal bridge, and micrognathia [1, 2, 3, 4]. Other features of affected people include globalized hypotonia, seizures, GERD, constipation, cardiac defects such as VSD and ASD, umbilical herniation, elongated digits, precocious puberty, and frequent ear infections.[2, 4] In light of no associated autoimmune conditions in the patient's family history, this case highlights the unusual and new findings of alopecia areata and vitiligo in a patient with Alfi syndrome. This is the first case report to our knowledge demonstrating alopecia areata and vitiligo in a patient with Alfi syndrome.

The basis of Alfi syndrome is a deletion in a portion of the 9<sup>th</sup> chromosome, notably the “critical region” at the 9p22-3 region [3]. In most cases, Alfi syndrome appears to be caused by spontaneous (de novo) errors very early in embryonic development that occur sporadically [2]. In such instances, the parents of the affected child usually have normal chromosomes and a relatively low risk of having another child with the chromosomal abnormality. In other cases, Alfi syndrome may result from a “balanced translocation” in one of the parents when portions of certain chromosomes break off and are rearranged, resulting in shifting of genetic material and an altered set of chromosomes [3]. If a chromosomal

rearrangement is balanced, meaning that it consists of an altered but balanced set of chromosomes, it is usually harmless to the carrier. However, such a chromosomal rearrangement may be associated with an increased risk of abnormal chromosomal development in the carrier's offspring [2]. Chromosomal analysis and genetic counseling are typically recommended for parents of an affected child to help confirm or exclude the presence of a balanced translocation or other chromosomal rearrangement involving chromosome 9 in one of the parents [5]. Diagnosis of this syndrome can be achieved prenatally via amniocentesis or chorionic villus sampling. Diagnosis can also be done postnatally based on characteristic phenotypic findings, chromosomal analysis, or through clinical examinations such as an electrocardiogram or echocardiogram [5].

Treatment of this disorder is usually medical management of symptoms only, with the exception of surgical treatments for malformations that are refractory to medical therapy. For the other possible disorders faced by the Alfi Syndrome patient, these are treated medically.

## Conclusion

Alfi syndrome is a rare chromosomal disorder that warrants further research and investigation. There is a lack of research in this disease, given its significant symptoms and phenotypic characteristics. This is the first case report demonstrating alopecia areata and vitiligo in a patient with Alfi syndrome. This case highlights the importance of monitoring the development of dermatologic conditions and expanding the knowledge base for this rare syndrome. It is imperative that clinicians be able to recognize and treat the myriad of symptoms and traits that go along with Alfi syndrome. Tracking the progression of the patient's condition can lead clinicians to further characterize this disease and lead to a better understanding of its impact on the patient and their families.

## Figures



**Figure 1:** alopectic patches on occipital scalp





**Figure 2:** well-demarcated depigmented patches on extremities



**Figure 4:** scoliosis with depigmented patch at central spine



**Figure 3:** Pronounced hypertelorism, upslanting lateral palpebral fissures, and abnormal dentition.

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**Authors Information:** Raquel Rozner, BS; Danilo C Delcampo, MD<sup>\*</sup>; Marian Russo, MD  
Department of Dermatology, New York Medical College, USA

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