

## Turner Syndrome and Alopecia: What Relation?

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### Case Report

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**Abstract:** Turner syndrome is a rare genetic condition characterized by total or partial loss of one of the X chromosomes. Affecting approximately 1/2500 newborn females, it almost consistently associates short stature, premature ovarian failure and infertility. A variety of additional symptoms can occur including abnormalities of the eyes and ears, skeletal malformations, heart anomalies, kidney abnormalities and skin manifestations. We report the case of a patient followed for alopecia, referred for short stature and whose explorations concluded to the diagnosis of Turner syndrome.

**Keywords:** Turner syndrome – alopecia – autoimmune disease.

### INTRODUCTION

Turner syndrome was first described by Henry Turner in 1938 [1]. It represents one of the most common chromosomal disorders and maybe the most frequent genetic disorder of females. Turner's symptoms vary from one patient to another. Many features of this syndrome are nonspecific and others may occur slowly over time or can be subtle. Several cutaneous findings are observed in Turner syndrome, some of them are common such as congenital lymphedema, shield chest, wide nipples, micrognathia and low posterior hairline, while other stigmata are of unknown or increased frequency such as alopecia, psoriasis and vitiligo [2].

### CASE REPORT

A 23 years girl, followed 8 years ago for alopecia put under minoxidil 2%, ten sprays twice a day during 3 months without any improvement, she was sent to our department by her dermatologist who noticed a small size and primary amenorrhea.

On general examination her pulse rate was 70 / min, her blood pressure was 120/80 mmHg, her weight was 39 kg (between -2 and -3 SD), her height was 136 cm (less than - 4 SD), capillary blood glucose was 0.86 g/l, Tanner scale was at stage II for breast and stage I for hair, we also noticed a dysmorphic triangular face, a broad chest with widely spaced nipples, on thyroid examination she had homogeneous palpable thyroid, slightly increased by volume. Her hand X-ray showed a

bone age at 13 years, her thyroid ultrasound noted an aspect of thyroiditis. On blood examination she had low estradiol level with high FSH and LH levels, normal thyroid profile with positive anti TPO antibodies, her karyotype showed a Turner syndrome at 46 homogeneous state, i,X,(Xq). The patient was put under estrogenic substitution and sent to dermatology for further management or she received boluses of solumedrol with good progress and hair growth.



**Fig-1: alopecia in a girl with Turner syndrome**



**Fig-2: evolution after 2 bolus of solumedrol**

## DISCUSSION

Women with Turner syndrome are at excess risk of autoimmune diseases [3, 4], mainly thyroid diseases [5] such as Hashimoto's thyroiditis and Graves' disease, antithyroid antibodies are present in 50% of patients with only 15 to 37% developing hypothyroidism and 3% thyrotoxicosis, other autoimmune disease are noticed including celiac disease, ulcerative colitis, Crohn's disease, idiopathic thrombocytopenic purpura, autoimmune hepatitis, juvenile rheumatoid arthritis and type 1 diabetes mellitus [3].

Although dermatologic autoimmune disorders such as psoriasis and vitiligo are well known in turner syndrome, only a few associated cases of alopecia have been reported [6, 7]. Alopecia areata is a chronic disorder characterized by non-scarring hair loss from some or all areas of the body which mostly results in a few bald spots on the scalp [8], sometimes it's more extensive and all the hair on the scalp or all body hair is lost, it may also go into remission for a time, or may be permanent. It is common in children.

Alopecia areata is an autoimmune disorder mediated by T-lymphocyte, in which inflammatory

cells concentrate in the bulbar region of hair follicles, leading to premature arrest of the anagen phase and abnormal major histocompatibility complex expression in the area of the follicular epithelium [9], its pathogenesis associates an interplay of genetic and environmental factors. At the present time and even if numerous treatment options are now available such as corticosteroid, methotrexate, cyclosporine A and azathioprine, there is no cure for alopecia areata, the course of this disorder varies among individuals and is difficult to predict. Regarding the influence of karyotype on clinical features, several studies reported that the frequency of autoimmune diseases was higher in Turner patients with X isochromosome I (X) (p10) karyotype [10-12]. This frequency is probably due to X-chromosome genes haploinsufficiency that may be responsible for a decrease in auto-antigen exposure in the thymus and leakage of self-reactive T cells, thus predisposing to autoimmunity.

In our patient, turner syndrome with 46 homogeneous state i,X, (Xq) was associated with autoimmune thyroid disease and alopecia areata with good clinical response to corticosteroid treatment.

## CONCLUSION

Turner syndrome may be associated with autoimmune disease such as Hashimoto's thyroiditis or alopecia hence the need to search for autoimmune disorders in patients with a Turner syndrome especially those with X isochromosome.

## REFERENCES

1. Turner syndrome. Available from <https://rarediseases.org/rare-diseases/turner-syndrome/>.
2. Lowenstein EJ, Kim KH, Glick SA. Turner's syndrome in dermatology. *Journal of the American Academy of Dermatology*. 2004 May 1;50(5):767-76.
3. Bianchi I, Lleo A, Gershwin ME, Invernizzi P. The X chromosome and immune associated genes. *Journal of autoimmunity*. 2012 May 1;38(2-3):J187-92.
4. Jorgensen KT, Rostgaard K, Bache I, Biggar RJ, Nielsen NM, Tommerup N, Frisch M. Autoimmune diseases in women with Turner's syndrome. *Arthritis Rheum* 2010 Mar;62(3):658-66
5. Grossi A, Crino A, Luciano R, Lombardo A, Cappa M, Fierabracci A. *Italian Journal of Pediatrics* 2013, 39:79
6. Tebbe B, Gollnick H, Muller R, Reupke HJ, Orfanos CE. Alopecia areata and diffuse hypotrichosis associated with Ullrich-Turner Syndrome. Presentation of 4 patients. *Hautarzt* 1993;44:647-652. PMID:8225974
7. Gianfaldoni S, Tchernev G, Wollina U, Lotti T. A Case of Alopecia Areata in a Patient with Turner Syndrome. *Open Access Maced J Med Sci*. 2017 Jul 25; 5(4):493-496.
8. Alopecia areata. NORD (national organization for rare disorders). Available from <https://rarediseases.org/rare-diseases/alopecia-areata/>
9. Pratt CH, King LE Jr, Messenger AG, Christiano AM, Sundberg JP. Alopecia areata. *Nat Rev Dis Primers* 2017;3:17011.
10. Sparkes RS, Motulsky AG. The Turner syndrome with isochromosome X and Hashimoto's thyroiditis. *Ann Intern Med* 1967, 67:132-144.
11. Hamza RT, Raof NA, Abdallah KO. Prevalence of multiple forms of autoimmunity in Egyptian patients with Turner syndrome: relation to karyotype. *J Pediatr Endocrinol Metab*. 2013, 26:545-550.
12. De Kerdanet M, Lucas J, Lemee F, Lecornu M. Turner's syndrome with X-isochromosome and Hashimoto's thyroiditis. *Clin Endocrinol (Oxf)* 1994, 41:673-676.