Portuguese Journal of Dermatology and Venereology

Permanyer

Check for updates

*Port J Dermatol and Venereol.*

**CASE REPORT**

# Syringomas in a patient with Williams syndrome

## *Siringomas num doente com síndrome de Williams*

*Jorge Lindo*, Mariana Pedroso, João L. Antunes, Teresa Lemos, and Joana Calvão*

*Department of Dermatology Unidade Local de Saúde de Coimbra, Coimbra, Portugal*

## Abstract

Syringomas are benign adnexal neoplasms with predominant ductal differentiation. They are often multiple, preferentially located in the periorbital region, and may be associated with genetic syndromes, such as Down syndrome. We report the case of a 24-year-old male patient with Williams syndrome (WS) who presented to the Dermatology Clinic with multiple asymptomatic, erythematous-yellowish, millimetric, monomorphic papules on the chest, with onset at 5 years of age. An incisional biopsy revealed histopathological findings consistent with syringomas. Although syringomas are more commonly found on the periorbital region, they may also occur in other locations, including the thoracic region. A potential association between syringomas and WS is not established, highlighting the need for further studies to investigate a link between specific genetic alterations and adnexal proliferation. This case underscores the importance of considering syringoma in the differential diagnosis of papular lesions on the trunk in patients with genetic syndromes and the need to explore potential dermatological manifestations of WS.

***Keywords:*** Syringoma. Adnexal neoplasms. Williams syndrome. Dermatology.

## Resumo

Siringomas são neoplasias anexiais benignas com diferenciação predominantemente ductal. São frequentemente múltiplos, com localização preferencial na região periorbitária, podendo estar associados a síndromes genéticas, como a síndrome de Down. Um doente do sexo masculino, de 24 anos, com síndrome de Williams, recorreu à Consulta de Dermatologia por um quadro de múltiplas pápulas eritemato-amareladas, milimétricas, monomórficas e assintomáticas, na região torácica, com início aos 5 anos de idade. A biópsia incisional revelou achados histopatológicos compatíveis com siringomas. Embora os siringomas sejam mais comuns na região periorbitária, podem surgir noutras localizações, como a região torácica. Uma associação potencial com a síndrome de Williams não está estabelecida, sendo necessários estudos para investigar uma eventual relação entre determinadas alterações genéticas e proliferação anexial. Este caso reforça a importância de considerar siringomas no diagnóstico diferencial de lesões papulosas do tronco em doentes com síndromes genéticas e de explorar potenciais manifestações dermatológicas da síndrome de Williams.

***Palavras-chave:*** Siringoma. Neoplasia anexial. Síndrome de Williams. Dermatologia.

***Correspondence:**
Jorge Lindo
E-mail: lindojorgester@gmail.com

**Received:** 29-12-2025
**Accepted:** 16-01-2026
DOI: 10.24875/PJDV.25000104

Available online: 24-02-2026
*Port J Dermatol and Venereol*. (ahead of print)
www.portuguesejournalofdermatology.com

## Introduction

Syringomas are benign adnexal neoplasms characterized by distinctive ductal differentiation. They have a higher incidence in females and in Asian populations and have been associated with certain genetic syndromes (such as Down syndrome) and metabolic conditions, including diabetes mellitus. They typically present as multiple, asymptomatic, well-defined, skin-colored papules and generally do not require treatment unless for cosmetic concerns[1]. The etiology of syringomas remains controversial, with evidence supporting roles for both genetic predisposition and hormonal imbalances[2]. The risk of malignant transformation is negligible, and treatment is generally not required[1].

Williams syndrome (WS) is a rare, multisystemic genetic disorder caused by contiguous gene deletions on chromosome 7[3], including genes implicated - or possibly implicated - in cutaneous and connective-tissue features, such as *ELN* (elastin), *CLDN3* (claudin-3), *CLDN4* (claudin-4), *LIMK1,* and *NCF1*, among others. Consequently, WS may affect the skin, leading to softer skin, premature graying of the hair, increased wrinkling, and abnormal scarring[4].

To date, an association between syringomas and WS has not been reported in the literature.

## Case report

A 24-year-old male patient with WS presented to the Dermatology Clinic with multiple, asymptomatic, millimetric, monomorphic, erythematous–yellowish papular lesions on the chest, present since the age of five (Fig. 1). No triggering or aggravating factors were identified, and no other cutaneous abnormalities were noted. Dermoscopy revealed multiple rounded, erythematous-orange lesions on a homogeneous pink background, along with fine, subtle linear vessels on the surface, without a clear arborizing or glomerular pattern, or a pigment network (Fig. 2). There was no family history of similar cutaneous lesions.

A clinical hypothesis of syringomas or another type of adnexal tumor was considered, and an incisional biopsy was performed. Histopathological examination revealed superficial and mid-dermal ductal structures lined by a double layer of epithelial cells, with lumina either optically empty or filled with eosinophilic material — findings consistent with syringomas (Fig. 3).

The available therapeutic options were explained to the patient and his family; however, they opted against treatment given the benign nature of the lesions and the absence of functional or esthetic impact.

## Discussion

We report a case of thoracic syringomas in a patient with WS. Although syringomas are more commonly found in the periorbital region, they can also appear in other locations, such as the thoracic and cervical regions, as observed in our case[1].

The definitive diagnosis of syringomas relies on histopathology, which typically reveals multiple nests of cells with central ductal differentiation forming lumina lined by an eosinophilic cuticle, within a sclerotic stroma, confined to the superficial dermis, similar to our findings[1]. However, the typical clinical presentation, along with dermoscopic findings, is clinically relevant.[2] Although dermoscopic features are not yet well-defined in the literature, our observations were consistent with previously reported features, namely pinpoint and small linear vessels on a diffuse orange-pink background. Notably, white globules or pigment networks, which have also been described, were not observed in this case[5-7].

A potential association with WS has not been established. Nevertheless, other genetic syndromes have been linked to this condition, and there is evidence that some genetic alterations in WS may affect proteins involved in skin and soft-tissue homeostasis[1-4]. Down syndrome, Brooke-Spiegler syndrome, Nicolaus-Balus syndrome, Costello syndrome, Marfan syndrome and Ehlers-Danlos syndrome have all been associated with multiple syringomas[8]. In Brooke-Spiegler syndrome, *CYLD* gene mutations are known to drive adnexal tumor development[9], while in Down syndrome, trisomy 21 is associated with immune dysregulation and altered adnexal biology, which may explain some of the possible cutaneous manifestations[10]. While familial syringomas have been described[11], no similar lesions were identified in the patient's relatives, arguing against an autosomal dominant inheritance pattern.

Inflammatory, metabolic (such as diabetes) and hormonal imbalances have been proposed as contributors to syringoma development, and endocrinopathies are indeed frequent in several syndromic conditions, including WS[2,4]. However, in the present case, the lesions were noted since early childhood, suggesting that genetic factors associated with WS may play a more relevant role than cumulative hormonal or metabolic influences.

Treatment of syringomas is primarily pursued for cosmetic concerns. Even in patients with associated syndromes, these lesions do not carry a risk of malignant transformation. When desired by the patient, laser

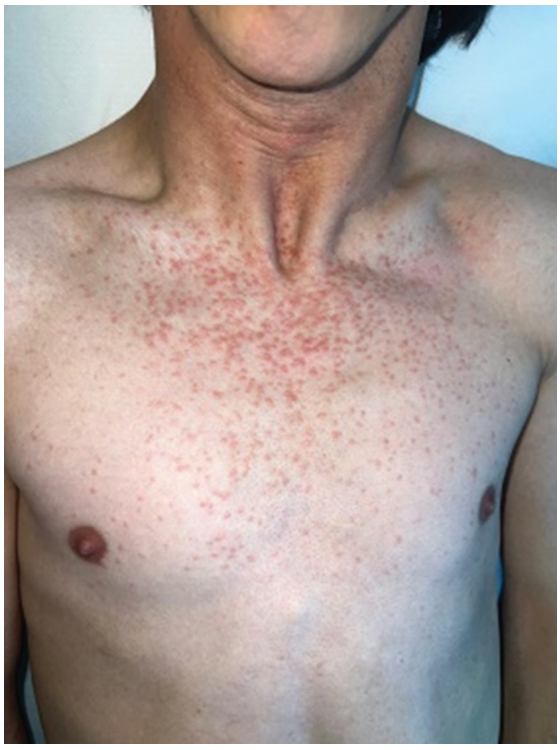

**Figure 1.** Clinical photograph of multiple eruptive syringomas in the upper thoracic region.

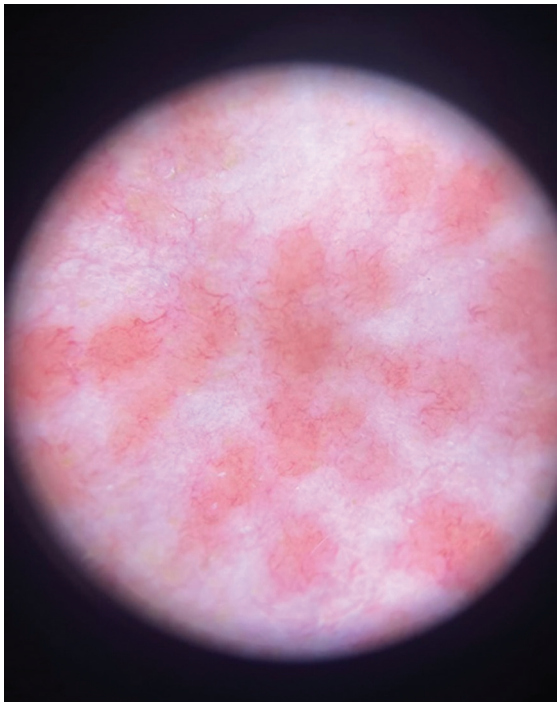

**Figure 2.** Dermoscopy showing erythematous-orange lesions on a homogeneous pink background, along with fine, subtle linear vessels on the surface.

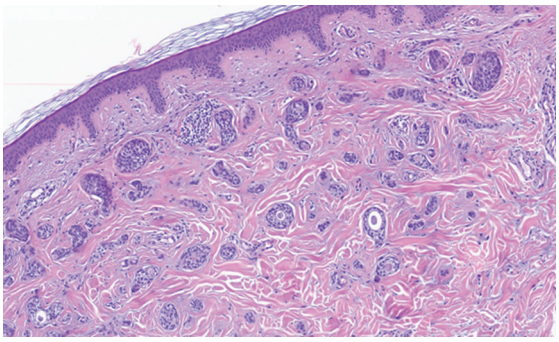

**Figure 3.** Histopathological examination showing dermal ductal structures, consistent with syringomas.

ablation may be a reasonable option, particularly in cases of eruptive syringomas[1,2].

## Conclusion

Further studies are needed to investigate a possible relationship between specific genetic alterations - namely deletion involving genes with cutaneous expression - and adnexal development and proliferation. This case highlights the importance of considering syringomas in the differential diagnosis of papular lesions in patients with genetic syndromes and of further exploring the dermatological manifestations of WS.

## Funding

None.

## Conflicts of interest

None.

## Ethical considerations

**Protection of humans and animals.** The authors declare that no experiments involving humans or animals were conducted for this research.

**Confidentiality, informed consent, and ethical approval.** The authors followed their institution's confidentiality protocols and obtained written informed consent from the patient.

**Declaration on the use of artificial intelligence.** The authors declare that no generative artificial intelligence was used in the writing of this manuscript.

## References


1. Bolognia JL, Schaffer JV Cerroni L. Dermatology. 5th ed. Philadelphia, PA: Elsevier; 2024.
2. Geist R, Crane JS. Syringoma. In: StatPearls. Treasure Island, FL: StatPearls Publishing; 2024. Available from: https://www.ncbi.nlm.nih.gov/books/nbk603740
3. Kozel BA, Barak B, Kim CA, Mervis CB, Osborn LR, Porter M, et al. Williams syndrome. Nat Rev Dis Primers. 2021;7:42.
4. Kozel BA, Bayliss SJ, Berk DR, Waxler JL, Knutsen RH, Danback JR, et al. Skin findings in Williams syndrome. Am J Med Genet A. 2014; 164A:2217-25.
5. Zaballos P, Gómez-Martín I, Martin JM, Bañul J. Dermoscopy of adnexal tumors. Dermatol Clin. 2018;36:397-412.
6. Corazza M, Borghi A, Minghetti S, Ferron P, Virgili A. Dermoscopy of isolated syringoma of the vulva. J Am Acad Dermatol. 2017;76:S37-9.
7. Lai M, Muscianese M, Piana S, Chester J, Borsari S, Paolino G, et al. Dermoscopy of cutaneous adnexal tumours: a systematic review of the literature. J Eur Acad Dermatol Venereol. 2022;36:1524-40.
8. Williams K, Shinkai K. Evaluation and management of the patient with multiple syringomas: a systematic review of the literature. J Am Acad Dermatol. 2016;74:1234-40.
9. Bowen S, Gill M, Lee DA, Fisher G, Geronemus RG, Vazquez ME, et al. Mutations in the CYLD gene in Brooke-Spiegler syndrome, familial cylindromatosis, and multiple familial trichoepithelioma: lack of genotype-phenotype correlation. J Invest Dermatol. 2005;124:919-20.
10. Rakasiwi T, Ryan C, Stein A, Vu A, Dykman M, Shah I, et al. Dermatologic conditions in down syndrome: a multi-site retrospective review of international classification of diseases codes. Pediatr Dermatol. 2024; 41:1047-52.
11. Singh SK, Rai T. Familial syringomas. Indian J Dermatol. 2013;58:412.