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Unveiling the complexity of Schimmelpenning–Feuerstein–Mims syndrome: A comprehensive case study

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ABSTRACT

Purpose: The purpose of this study was to report the comprehensively examined patient exhibiting oculocutaneous clinical features of Schimmelpenning–Feuerstein–Mims syndrome (SFMS).

Background: Schimmelpenning–Feuerstein–Mims syndrome (SFMS) is a rare phakomatosis characterized by the presence of sebaceous hamartomas on the skin along with extracutaneous abnormalities involving various neuroectodermal systems. The syndrome is typically sporadic and can originate from postzygotic mutations in genes implicated in the RAS signaling pathway (RAS proteins and their downstream pathways play pivotal roles in cell proliferation, differentiation, survival, and cell death): *HRAS* (11p15), *NRAS* (1p13), and *KRAS* (12p12). This case report involves a comprehensively examined patient exhibiting oculocutaneous clinical features of SFM, without neurological or involvement in other areas. Clinical and molecular diagnoses enable tailored monitoring of potentially affected organs and systems, involving a multidisciplinary approach by various medical specialists.

Conclusion and importance: The SFM is attributed to a pathogenic variant in *KRAS* gene. The molecular analysis in individuals suspected of SFMS involves identifying the somatic mutation in affected tissues and comparing it with non-affected tissues, such as mucosa or blood. Early detection and appropriate treatment of ophthalmological abnormalities associated with SFM are crucial to improving the quality of life and visual prognosis of affected individuals.

1. Introduction

Schimmelpenning–Feuerstein–Mims syndrome (SFMS, or linear nevus sebaceous of Jadassohn), is a rare phakomatosis characterized by the presence of sebaceous hamartomas on the skin along with extracutaneous abnormalities affecting the central nervous, ocular, and skeletal systems. Neurological features include epilepsy, cognitive delays, and brain malformations, while ocular manifestations range from eyelid colobomas and epibulbar dermoids to optic nerve hypoplasia and corneal opacities, often causing significant visual impairment. Skeletal anomalies such as hemihypertrophy and craniofacial asymmetries are also common.[1–4]

Ophthalmological involvement is frequent and varied, often contributing to visual deficiency in affected individuals.[5] One of the most common ophthalmological manifestations in SFM is the presence of sebaceous nevi in the periocular region,[2,6,7] which may involve the eyelids and surrounding areas. These nevi can be associated with structural abnormalities, such as microphthalmia, choristomas, and colobomas of the iris or optic nerve.[8] Additionally, alterations in the orbital region, including ptosis or ocular misalignment, may impact visual function.

The syndrome is typically sporadic,[9] and originated by postzygotic (mosaic) mutations in genes implicated in the RAS signaling pathway (RAS signaling pathway, plays pivotal roles in cell proliferation,

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differentiation, survival, and cell death): *HRAS* (11p15), *NRAS* (1p13), and *KRAS* (12p12).[7,10] Samples for genetic study must be obtained from affected organs. The timing of the mutation establishes that germ cells are spared of damage, so affected individuals will not pass the mutation to their offspring.[11] This intriguing and complex syndrome exemplifies the dynamic interplay between genetic factors and embryonic development, culminating in the distinctive clinical features observed in affected individuals. SFMS is part of the mosaic RASopathies spectrum, which includes oculoectodermal syndrome (OES) and encephalocraniocutaneous lipomatosis (ECCL).[10,12] The condition's multifaceted nature highlights the importance of further research to unravel its underlying genetic mechanisms and potential therapeutic interventions.

A comprehensive ophthalmological evaluation of diagnosed patients is essential since SFM is a complex syndrome affecting multiple systems. Early detection and appropriate treatment of ophthalmological abnormalities associated with SFM are crucial to improving the quality of life and visual prognosis of affected individuals. We present a case involving a comprehensively examined patient exhibiting oculocutaneous clinical features of SFMS, without neurological or involvement in other areas.

2. Case presentation

A 19-year-old male patient presented to our cornea service for evaluation of corneal opacification of the left eye since birth, associated with low vision and aesthetic discomfort. No previous diagnosis and treatment were given before referral to us. The patient was born to healthy parents after a normal pregnancy via cesarean section, with no complications recorded during prenatal care. The patient is the only child of non-consanguineous parents from Mexico City. His family history was unremarkable. He exhibits normal intellectual and physical development. Up to this point, he has not developed seizures, although he does complain of recurrent headaches that have shown improvement with paracetamol.

Physical exam showed areas of alopecia in the left temporal region, horizontal eyelid fissures, and prominent nasal bridge. Skin findings showed a well-circumscribed brownish-yellow sebaceous plaque with irregular borders and a rough, elevated surface. The main plaque was located on the left temporal region extending to the root of the lower jaw. Another plaque with similar characteristics was found in the midline of the neck. Overall, comedogenic lesions appeared throughout the surface of the face (Fig. 1). No other abnormalities were found.

Ophthalmological examination revealed best corrected visual acuity (BCVA) in the Right eye (RE) 20/50 (Snellen) and counting fingers from 4 m in the left eye (LE). Per-rotational nystagmus was present in both eyes (OU). On the RE he presented superficial corneal vascularization in the 9 and 12 o'clock meridians and clear cornea. On gonioscopy, the RE

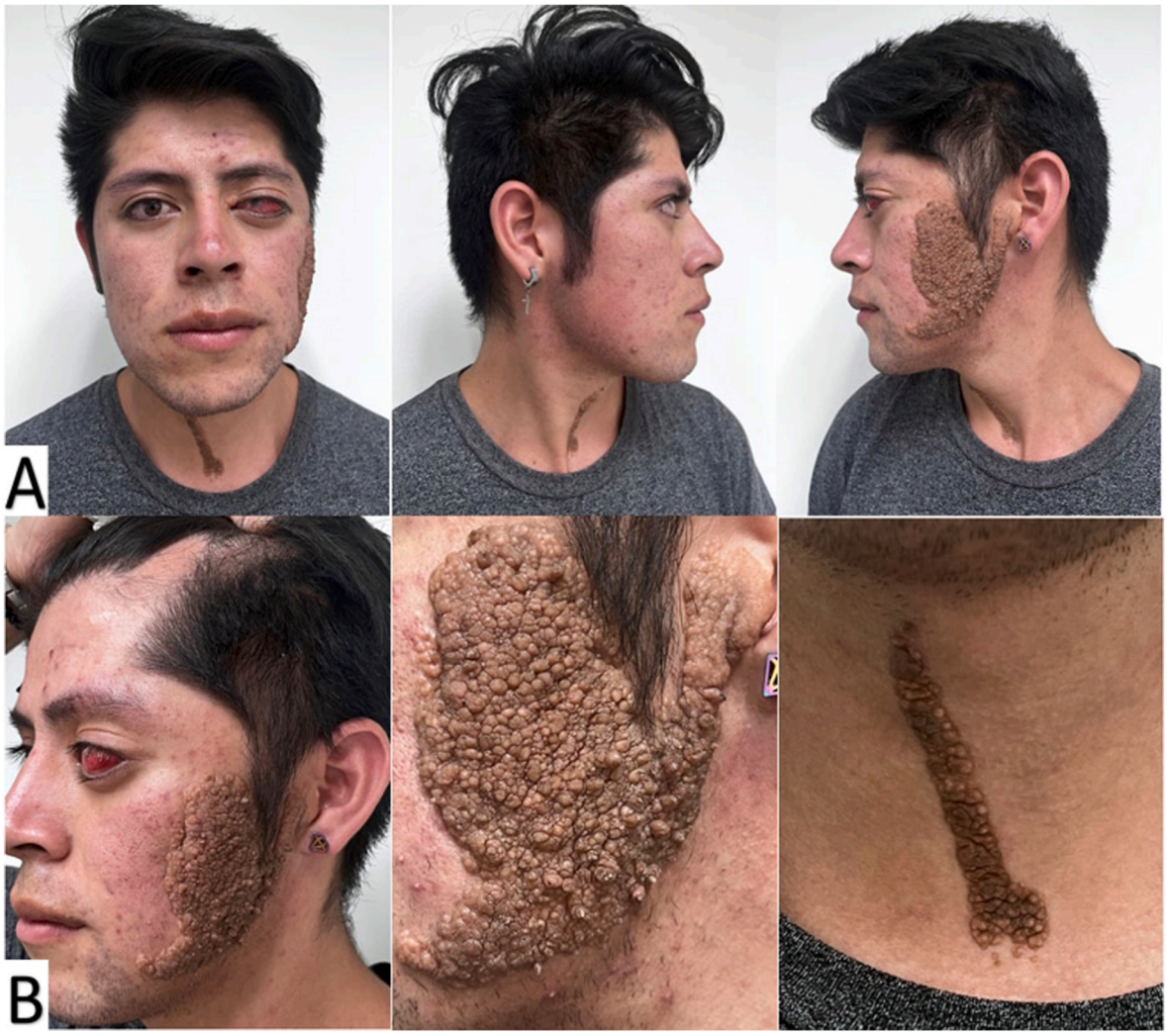

Fig. 1. A. Clinical characteristics of the patient. Frontal and lateral view showing a nevus sebaceous on the left side of the face. **B.** Nevus sebaceous located on the left cheek, below the eye line and above the jawline. Mild alopecia is noticeable on the affected area, showcasing the subtle hair loss that can be associated with this condition. Another linear nevus sebaceous is observed on the midline of the neck.

had a low insertion of iris root, the rest of the anterior segment had no alterations. Fundoscopic examination of the RE revealed an excavated optic disc, associated with a peripapillary staphyloma and chorioretinal atrophy. No other anomalies were found on RE. In the LE he presented a firm, flat, pink mass localized in the temporal bulbar conjunctiva that trespassed the corneal limbus and covered superficially 80 % of the cornea, which corresponded to an epibulbar dermoid, a sector in nasal cornea was spared where vascularization appeared in a spiral pattern. The anterior chamber was formed and corectopia was observed. Other intraocular structures could not be evaluated (Fig. 2). Ocular ultrasonography of RE revealed an optic nerve staphyloma and the LE revealed optic nerve staphyloma and a calcified plaque in the upper nasal region that resulted in a posterior sonic shadow, corresponding to a choroidal osteoma. A spectral domain optical coherence tomography (SD-OCT) of the macula was performed which revealed the presence of an optic nerve staphyloma accompanied by peripapillary subretinal fluid. The presence of subretinal fluid may be attributed to an optic nerve pit, the identification of which proved challenging due to the suboptimal quality of the images, that were caused mainly by the presence of irregular surface within the area of the staphyloma or because of poor fixation due to the nystagmus (Fig. 3).

Due to the multisystem involvement typically associated with SFMS, which was suspected in our patient, he was approached for neurological, cardiovascular, and abdominal evaluations, all of which did not reveal extracutaneous findings. The patient was referred to the medical genetics service, where, based on the clinical findings, the diagnosis of Schimmelpenning-Feuerstein-Mims syndrome was suspected. According to previous reports of molecular diagnosis in individuals suspected of SFMS in the Mexican population, molecular testing of the *KRAS* gene was recommended.

3. Materials and methods

The study was approved by Institutional Review Board and Ethics committees and was conducted according to the principles expressed in the Declaration of Helsinki. Patient signed a written informed consent for clinical and molecular studies and for tissue biopsies. A complete ophthalmologic and dermatologic examination was performed. A small biopsy was obtained from the sebaceous nevi. Buccal mucosa and peripheral leukocytes were also obtained. Tubes containing skin biopsies were placed in a TissueLyser Adapter Set for disruption and homogenization of tissues, following the manufacturer's recommendations (TissueLyser II system, Qiagen, Hilden, Germany). Genomic DNA from biopsy tissues, peripheral blood leukocytes, and buccal mucosa cells was isolated using the QIAamp DNA Mini Kit (Qiagen, Hilden, Germany), and Gentra Puregene Buccal Cell Kit (Qiagen, Hilden, Germany), following the manufacturer's instructions. The coding regions of *KRAS* and its adjacent intronic sequences were amplified by PCR using pairs of primers corresponding to Ensembl reference sequences (*KRAS*, ENST00000311936.7, NM_004985.5). Direct automated sequencing of exons of *KRAS* was performed with the BigDye Terminator 3.1 Cycle Sequencing kit (Applied Biosystems, Foster City, CA). All samples were analyzed by ABI3130 Genetic Analyzer (Applied Biosystems) and sequences were compared against the respective reference sequences.

4. Results

Sanger sequencing analysis revealed the *KRAS* heterozygous pathogenic variant c.35G > T in the affected biopsy skin; this variant corresponds to a transversion of guanine to thymine at nucleotide 35, which at the protein level predicts a change from glycine to valine at amino acid 12 (p.Gly12Val). This variant has been reported as pathogenic previously in the literature.[13] Sanger analysis in DNA from both blood and oral mucosa cells did not identify the c.35G > T *KRAS* (Fig. 4).

5. Discussion

SFMS is part of the mosaic RASopathies spectrum, which includes oculoectodermal syndrome (OES) and encephalocraniocutaneous lipomatosis (ECCL). These disorders are caused by somatic mutations in the

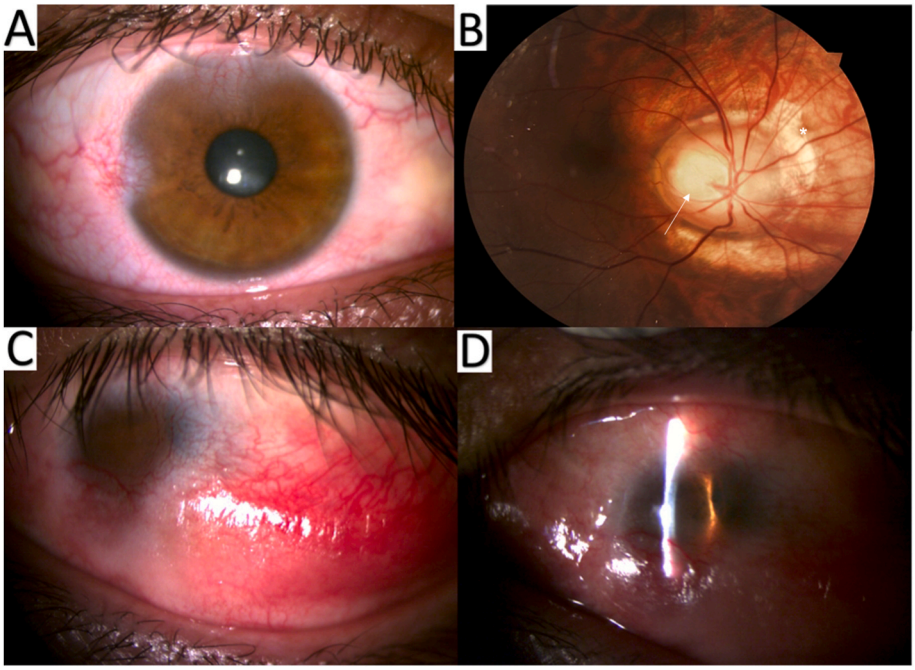

Fig. 2. A. The Right eye (RE) of the patient showing conjunctival hyperemia, accompanied by peripheral pannus at 9 and 12 o'clock positions. The rest of the cornea is clear. B. Fundus photograph of the right eye showing an excavated optic disc (arrow), associated with a peripapillary staphyloma and chorioretinal atrophy (asterisk). The macula with no abnormalities. **C.** Left Eye (LE) of the patient showing a pink-red mass on the bulbar conjunctiva, associated with keratinization on its surface. Conjunctivalization and vascularization of the cornea, leaving only the central region optically clear. **D.** LE of the patient showing corneal transparency at the center, and a formed anterior chamber.

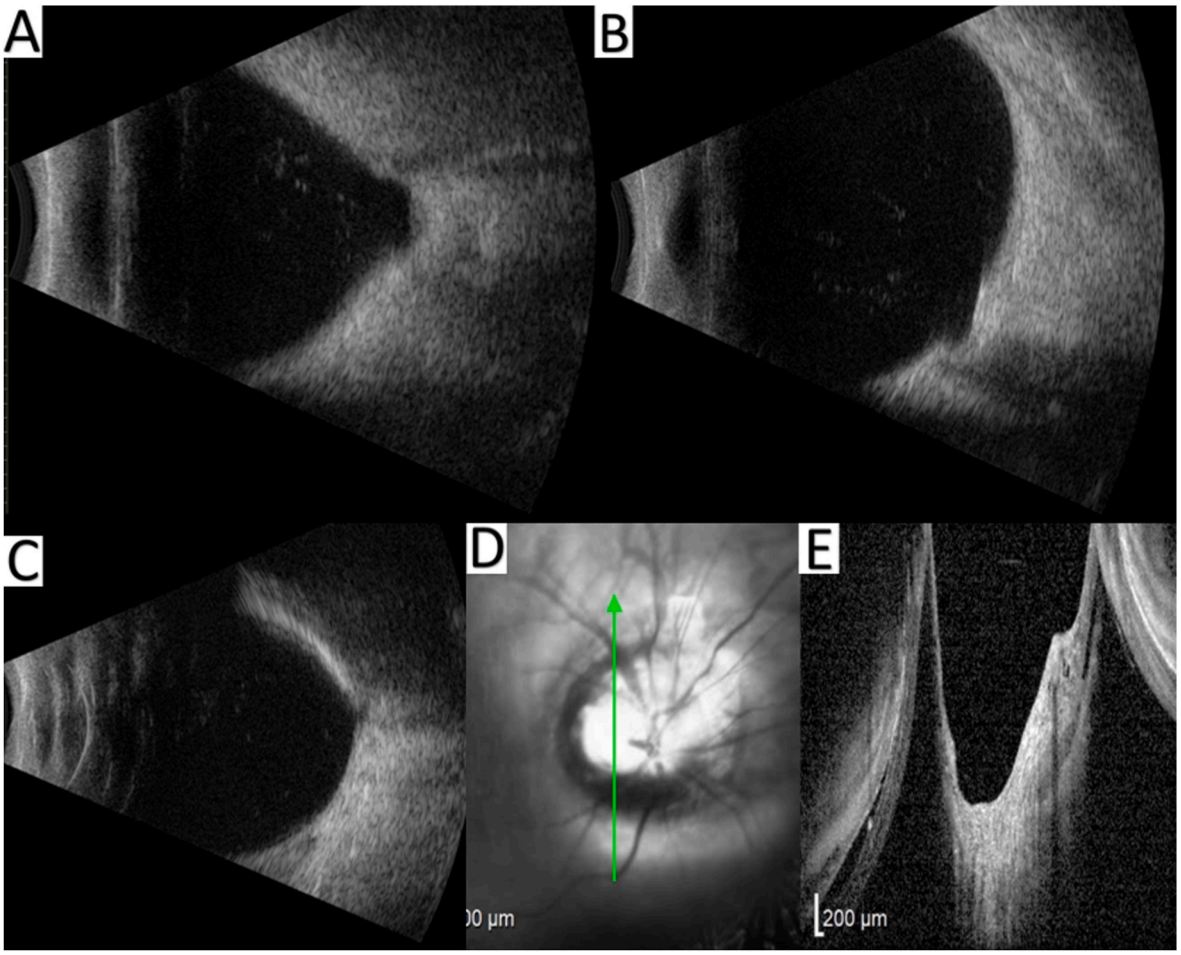

Fig. 3. **A.** Ultrasound B-scan of the RE showing the optic nerve staphyloma. **B.** Ultrasound B-scan of the LE showing the optic nerve staphyloma. **C.** Ultrasound B-scan of the LE showing an hyperreflective flat lesion superonasal to the optic nerve head with shadowing behind, confirming the diagnosis of osteoma. **D.** SD-OCT of the RE confirming optic nerve staphyloma accompanied by peripapillary subretinal fluid.

RAS/MAPK pathway and exhibit a wide range of clinical manifestations due to mosaicism. SFMS is a rare multisystem disorder characterized by nevus sebaceous (NS) associated with extracutaneous anomalies. Nevus sebaceous is a hallmark of Schimmelpenning syndrome,[6,14] and a classic triad described in this syndrome is the presence of sebaceous nevi, epileptic seizures, and intellectual disability.[15,16] However, recent reports have shown that the phenotypic spectrum is very variable. This could be mainly due to the affected cell line, the timing of the post-zygotic mutation, and other mechanisms that are not fully understood. While the nevus is a defining feature, it represents just one aspect of a broader syndrome. Similarly, OES is defined by congenital scalp lesions and ocular dermoid, often accompanied by non-ossifying fibromas and jaw giant cell granulomas typically emerging in the first decade of life.[17] In contrast, ECCL is characterized by key features including nevus psiloliparus, subcutaneous lipomas, skin aplasia, and patchy alopecia.[18]

The scalp is the most common location of nevi (59.3 %), followed by the face (32.6 %), the neck (3.2 %), and in locations away from the head and neck (1.3 %).[6,14] Our patient has it located on the scalp and neck. Secondary malignancies within the nevus commonly arise post-puberty and occasionally before[19,20]; this emphasizes the importance of continuous monitoring.

Ophthalmological abnormalities are the second most common manifestation of SFMS, following cutaneous conditions and central nervous system impairments. The incidence of these ocular manifestations is about 59–68 %.[21,22] Major ocular anomalies include colobomas and choristomas.[6,8] Involvement of other organ systems occurs in 61 % of the cases.[21] Neurological abnormalities are reported to occur in 66 % of cases,[6,21] often presenting as epilepsy, intellectual disability, or structural brain anomalies. In this case, the patient presents multiple ophthalmologic anomalies, including epibulbar dermoid, optic nerve staphyloma, corneal opacity, and choroidal osteoma, along with skin manifestations such as alopecia and sebaceous plaque. The relevance of this report lies in the diversity of predominant ocular and skin alterations and the absence of abnormalities in other areas (cardiovascular, urinary, skeletal, and neurological) when compared to previously documented cases. Choroidal osteoma is a rare and benign ossifying tumor involving the choroid.[23] Optic disc pits are a rare congenital anomaly that belongs to the group of excavated optic disc anomalies.[24] The prevalence of these anomalies remains unreported; however, reports indicate the necessity for vigilant follow-up due to potential complications, such as choroidal neovascularization and retinal detachment.[3,25,26] In our case, diminished visual acuity of the LE is attributed to various factors, ranging from corneal opacity, choroidal osteoma, and optic nerve staphyloma.

Syndromes with overlapping clinical findings in the eye, heart, skin, and hair have been recently shown to result from somatic mosaicism for mutations in the genes involved in the RAS pathway. These conditions include oculoectodermal syndrome (OES, OMIM #600268), Schimmelpenning-Feuerstein-Mims syndrome (SFMS, OMIM #163200), and encephalocraniocutaneous lipomatosis (ECCL, OMIM #613001); are characterized by a range of interconnected anomalies such as scalp lesions, epilepsy, epibulbar dermoid, cloudy cornea, eyelid coloboma, aorta coarctation, and variations in skin pigmentation.[10,17,27,28] Because

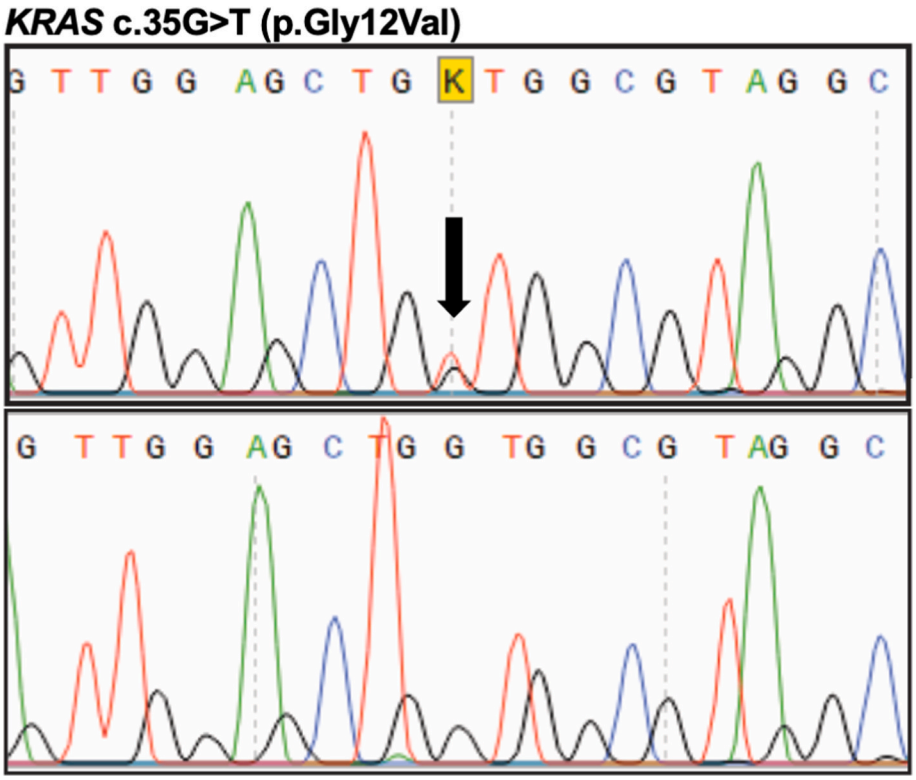

Fig. 4. Molecular analysis of *KRAS*. Sanger sequencing on proband showed a heterozygous mutation c.35G > T (p.Gly12Val) in the lesional tissue (top). Sanger sequencing in the gDNA of the buccal mucosa sample showed the wild-type sequence (bottom).

clinical features are diverse, molecular confirmation is necessary to be certain of the diagnosis. Our patient underwent biopsies of the sebaceous nevi and a buccal swab sample from the ipsilateral side, which confirmed the diagnosis of SFMS.

Based on the existing evidence, there is no necessity to perform the removal of SN as a preventive measure, as the likelihood of malignant neoplasia development is less than previously thought.[29–31] In essence, when dealing with a Jadassohn's sebaceous nevus, a comprehensive examination of various systems should be carried out. Analysis should involve EEG, CT, or MRI imaging, assessment of liver and kidney, and measurement of calcium and phosphate concentrations in both the bloodstream and urine.

In conclusion, we present a case of a patient diagnosed with Schimmelpenning-Feuerstein-Mims syndrome, which is attributed to the pathogenic variant c.35G > T, p.Gly12Val in *KRAS* gene. The molecular analysis in these individuals involves identifying the somatic mutation in affected tissues and comparing it with non-affected tissues, such as mucosa or blood. Literature reports on this condition are limited, underscoring the significance of comprehensive clinical characterization to expand the described phenotypic spectrum. Variability in expression is observed, depending on the specific cell lineage affected. Our patient, for example, exhibits more pronounced ocular and cutaneous manifestations. Both clinical and molecular diagnoses enable tailored monitoring of potentially affected organs and systems, involving a multidisciplinary approach by various medical specialists. Given the phenotypic variability and the risk of progressive complications, early and accurate diagnosis of SFMS is crucial. Multidisciplinary follow-up, integrating ophthalmology, dermatology, neurology, and genetics, ensures timely intervention for associated abnormalities and optimizes patient outcomes. Regular monitoring of ocular, neurological, and systemic manifestations is essential for reducing the long-term morbidity associated with this condition.

CRediT authorship contribution statement

Guillermo Raul Vera-Duarte: Writing – review & editing, Writing – original draft, Visualization, Validation, Resources, Investigation, Conceptualization. **Ruth Eskenazi-Betech:** Writing – original draft, Visualization, Validation, Project administration, Data curation, Conceptualization. **Isabel De la Fuente-Batta:** Writing – original draft, Methodology, Investigation, Data curation, Conceptualization. **David Carreño-Bolaños:** Writing – review & editing, Writing – original draft, Visualization, Validation, Project administration, Investigation. **Oscar F. Chacón-Camacho:** Writing – review & editing, Writing – original draft, Visualization, Validation, Supervision, Project administration, Investigation. **Juan C. Zenteno:** Writing – review & editing, Writing – original draft, Visualization, Validation, Supervision, Project administration, Methodology, Investigation, Data curation. **Enrique O. Graue-Hernandez:** Writing – review & editing, Writing – original draft, Visualization, Validation, Supervision, Methodology, Investigation, Data curation, Conceptualization.

Patient consent

The patient provided written consent regarding the publication of the present case report.

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Conflicts of interest
None.
All authors have no financial disclosure.
All authors attest that they meet the current ICMJE criteria for Authorship.

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Declaration of competing interest

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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