

World Journal of *Clinical Cases*

World J Clin Cases 2022 May 16; 10(14): 4327-4712



OPINION REVIEW

- 4327 Emerging role of biosimilars in the clinical care of inflammatory bowel disease patients
Najeeb H, Yasmin F, Surani S

MINIREVIEWS

- 4334 Practical insights into chronic management of hepatic Wilson's disease
Lynch EN, Campani C, Innocenti T, Dragoni G, Forte P, Galli A
- 4348 Adipose-derived stem cells in the treatment of hepatobiliary diseases and sepsis
Satilmis B, Cicek GS, Cicek E, Akbulut S, Sahin TT, Yilmaz S

ORIGINAL ARTICLE**Clinical and Translational Research**

- 4357 Learning curve for a surgeon in robotic pancreaticoduodenectomy through a "G"-shaped approach: A cumulative sum analysis
Wei ZG, Liang CJ, Du Y, Zhang YP, Liu Y
- 4368 Clinical and prognostic significance of expression of phosphoglycerate mutase family member 5 and Parkin in advanced colorectal cancer
Wu C, Feng ML, Jiao TW, Sun MJ

Case Control Study

- 4380 Significance of preoperative peripheral blood neutrophil-lymphocyte ratio in predicting postoperative survival in patients with multiple myeloma bone disease
Xu ZY, Yao XC, Shi XJ, Du XR

Retrospective Study

- 4395 Association between depression and malnutrition in pulmonary tuberculosis patients: A cross-sectional study
Fang XE, Chen DP, Tang LL, Mao YJ
- 4404 Pancreatic cancer incidence and mortality patterns in 2006-2015 and prediction of the epidemiological trend to 2025 in China
Yin MY, Xi LT, Liu L, Zhu JZ, Qian LJ, Xu CF
- 4414 Evaluation of short- and medium-term efficacy and complications of ultrasound-guided ablation for small liver cancer
Zhong H, Hu R, Jiang YS

- 4425 Hematopoiesis reconstitution and anti-tumor effectiveness of Pai-Neng-Da capsule in acute leukemia patients with haploidentical hematopoietic stem cell transplantation

Yuan JJ, Lu Y, Cao JJ, Pei RZ, Gao RL

- 4436 Oral and maxillofacial pain as the first sign of metastasis of an occult primary tumour: A fifteen-year retrospective study

Shan S, Liu S, Yang ZY, Wang TM, Lin ZT, Feng YL, Pakezhati S, Huang XF, Zhang L, Sun GW

- 4446 Reduced serum high-density lipoprotein cholesterol levels and aberrantly expressed cholesterol metabolism genes in colorectal cancer

Tao JH, Wang XT, Yuan W, Chen JN, Wang ZJ, Ma YB, Zhao FQ, Zhang LY, Ma J, Liu Q

Observational Study

- 4460 Correlation of pressure gradient in three hepatic veins with portal pressure gradient

Wang HY, Song QK, Yue ZD, Wang L, Fan ZH, Wu YF, Dong CB, Zhang Y, Meng MM, Zhang K, Jiang L, Ding HG, Zhang YN, Yang YP, Liu FQ

- 4470 Multi-slice spiral computed tomography in diagnosing unstable pelvic fractures in elderly and effect of less invasive stabilization

Huang JG, Zhang ZY, Li L, Liu GB, Li X

SYSTEMATIC REVIEWS

- 4480 Distribution and changes in hepatitis C virus genotype in China from 2010 to 2020

Yang J, Liu HX, Su YY, Liang ZS, Rao HY

CASE REPORT

- 4494 Bow hunter's syndrome successfully treated with a posterior surgical decompression approach: A case report and review of literature

Orlandi N, Cavallieri F, Grisendi I, Romano A, Ghadirpour R, Napoli M, Moratti C, Zanichelli M, Pascarella R, Valzania F, Zedde M

- 4502 Histological remission of eosinophilic esophagitis under asthma therapy with IL-5 receptor monoclonal antibody: A case report

Huguenot M, Bruhm AC, Essig M

- 4509 Cutaneous mucosa-associated lymphoid tissue lymphoma complicating Sjögren's syndrome: A case report and review of literature

Liu Y, Zhu J, Huang YH, Zhang QR, Zhao LL, Yu RH

- 4519 Plexiform neurofibroma of the cauda equina with follow-up of 10 years: A case report

Chomanskis Z, Jusky R, Cepkus S, Dulko J, Hendrixson V, Ruksenas O, Rocka S

- 4528 Mixed porokeratosis with a novel mevalonate kinase gene mutation: A case report

Xu HJ, Wen GD

- 4535 Isolated pancreatic injury caused by abdominal massage: A case report

Sun BL, Zhang LL, Yu WM, Tuo HF

- 4541** Bronchiolar adenoma with unusual presentation: Two case reports
Du Y, Wang ZY, Zheng Z, Li YX, Wang XY, Du R
- 4550** Periodontal-orthodontic interdisciplinary management of a “periodontally hopeless” maxillary central incisor with severe mobility: A case report and review of literature
Jiang K, Jiang LS, Li HX, Lei L
- 4563** Anesthesia management for cesarean section in a pregnant woman with odontogenic infection: A case report
Ren YL, Ma YS
- 4569** Convulsive-like movements as the first symptom of basilar artery occlusive brainstem infarction: A case report
Wang TL, Wu G, Liu SZ
- 4574** Globe luxation may prevent myopia in a child: A case report
Li Q, Xu YX
- 4580** Computer tomography-guided negative pressure drainage treatment of intrathoracic esophagojejunal anastomotic leakage: A case report
Jiang ZY, Tao GQ, Zhu YF
- 4586** Primary or metastatic lung cancer? Sebaceous carcinoma of the thigh: A case report
Wei XL, Liu Q, Zeng QL, Zhou H
- 4594** Perianesthesia emergency repair of a cut endotracheal tube’s inflatable tube: A case report
Wang TT, Wang J, Sun TT, Hou YT, Lu Y, Chen SG
- 4601** Diagnosis of cytomegalovirus encephalitis using metagenomic next-generation sequencing of blood and cerebrospinal fluid: A case report
Xu CQ, Chen XL, Zhang DS, Wang JW, Yuan H, Chen WF, Xia H, Zhang ZY, Peng FH
- 4608** Primary sigmoid squamous cell carcinoma with liver metastasis: A case report
Li XY, Teng G, Zhao X, Zhu CM
- 4617** Acute recurrent cerebral infarction caused by moyamoya disease complicated with adenomyosis: A case report
Zhang S, Zhao LM, Xue BQ, Liang H, Guo GC, Liu Y, Wu RY, Li CY
- 4625** Serum-negative Sjogren's syndrome with minimal lesion nephropathy as the initial presentation: A case report
Li CY, Li YM, Tian M
- 4632** Successful individualized endodontic treatment of severely curved root canals in a mandibular second molar: A case report
Xu LJ, Zhang JY, Huang ZH, Wang XZ

- 4640 Successful treatment in one myelodysplastic syndrome patient with primary thrombocytopenia and secondary deep vein thrombosis: A case report
Liu WB, Ma JX, Tong HX
- 4648 Diagnosis of an extremely rare case of malignant adenomyoepithelioma in pleomorphic adenoma: A case report
Zhang WT, Wang YB, Ang Y, Wang HZ, Li YX
- 4654 Management about intravesical histological transformation of prostatic mucinous carcinoma after radical prostatectomy: A case report
Bai SJ, Ma L, Luo M, Xu H, Yang L
- 4661 Hepatopulmonary metastases from papillary thyroid microcarcinoma: A case report
Yang CY, Chen XW, Tang D, Yang WJ, Mi XX, Shi JP, Du WD
- 4669 PD-1 inhibitor in combination with fruquintinib therapy for initial unresectable colorectal cancer: A case report
Zhang HQ, Huang CZ, Wu JY, Wang ZL, Shao Y, Fu Z
- 4676 Cutaneous metastasis from esophageal squamous cell carcinoma: A case report
Zhang RY, Zhu SJ, Xue P, He SQ
- 4684 Rare pattern of Maisonneuve fracture: A case report
Zhao B, Li N, Cao HB, Wang GX, He JQ
- 4691 Suprasellar cistern tuberculoma presenting as unilateral ocular motility disorder and ptosis: A case report
Zhao BB, Tian C, Fu LJ, Zhang XB
- 4698 Development of plasma cell dyscrasias in a patient with chronic myeloid leukemia: A case report
Zhang N, Jiang TD, Yi SH
- 4704 Ovarian growing teratoma syndrome with multiple metastases in the abdominal cavity and liver: A case report
Hu X, Jia Z, Zhou LX, Kakongoma N

LETTER TO THE EDITOR

- 4709 Perfectionism and mental health problems: Limitations and directions for future research
Nazari N

ABOUT COVER

Editorial Board Member of *World Journal of Clinical Cases*, Jamir Pitton Rissardo, MD, Academic Research, Adjunct Associate Professor, Research Associate, Department of Medicine, Federal University of Santa Maria, Santa Maria 97105110, Brazil. jamirrissardo@gmail.com

AIMS AND SCOPE

The primary aim of *World Journal of Clinical Cases* (*WJCC*, *World J Clin Cases*) is to provide scholars and readers from various fields of clinical medicine with a platform to publish high-quality clinical research articles and communicate their research findings online.

WJCC mainly publishes articles reporting research results and findings obtained in the field of clinical medicine and covering a wide range of topics, including case control studies, retrospective cohort studies, retrospective studies, clinical trials studies, observational studies, prospective studies, randomized controlled trials, randomized clinical trials, systematic reviews, meta-analysis, and case reports.

INDEXING/ABSTRACTING

The *WJCC* is now indexed in Science Citation Index Expanded (also known as SciSearch®), Journal Citation Reports/Science Edition, Scopus, PubMed, and PubMed Central. The 2021 Edition of Journal Citation Reports® cites the 2020 impact factor (IF) for *WJCC* as 1.337; IF without journal self cites: 1.301; 5-year IF: 1.742; Journal Citation Indicator: 0.33; Ranking: 119 among 169 journals in medicine, general and internal; and Quartile category: Q3. The *WJCC*'s CiteScore for 2020 is 0.8 and Scopus CiteScore rank 2020: General Medicine is 493/793.

RESPONSIBLE EDITORS FOR THIS ISSUE

Production Editor: *Hua-Ge Yan*, Production Department Director: *Xu Guo*, Editorial Office Director: *Jin-Lei Wang*.

NAME OF JOURNAL

World Journal of Clinical Cases

ISSN

ISSN 2307-8960 (online)

LAUNCH DATE

April 16, 2013

FREQUENCY

Thrice Monthly

EDITORS-IN-CHIEF

Bao-Gan Peng, Jerzy Tadeusz Chudek, George Kontogeorgos, Maurizio Serati, Ja Hyeon Ku

EDITORIAL BOARD MEMBERS

<https://www.wjgnet.com/2307-8960/editorialboard.htm>

PUBLICATION DATE

May 16, 2022

COPYRIGHT

© 2022 Baishideng Publishing Group Inc

INSTRUCTIONS TO AUTHORS

<https://www.wjgnet.com/bpg/gerinfo/204>

GUIDELINES FOR ETHICS DOCUMENTS

<https://www.wjgnet.com/bpg/GerInfo/287>

GUIDELINES FOR NON-NATIVE SPEAKERS OF ENGLISH

<https://www.wjgnet.com/bpg/gerinfo/240>

PUBLICATION ETHICS

<https://www.wjgnet.com/bpg/GerInfo/288>

PUBLICATION MISCONDUCT

<https://www.wjgnet.com/bpg/gerinfo/208>

ARTICLE PROCESSING CHARGE

<https://www.wjgnet.com/bpg/gerinfo/242>

STEPS FOR SUBMITTING MANUSCRIPTS

<https://www.wjgnet.com/bpg/GerInfo/239>

ONLINE SUBMISSION

<https://www.f6publishing.com>

Mixed porokeratosis with a novel mevalonate kinase gene mutation: A case report

Hong-Jun Xu, Guang-Dong Wen

Specialty type: Dermatology

Provenance and peer review:

Unsolicited article; Externally peer reviewed.

Peer-review model: Single blind

Peer-review report's scientific quality classification

Grade A (Excellent): 0

Grade B (Very good): 0

Grade C (Good): C

Grade D (Fair): 0

Grade E (Poor): 0

P-Reviewer: Sridharan G, India

Received: July 30, 2021

Peer-review started: July 30, 2021

First decision: December 1, 2021

Revised: December 11, 2021

Accepted: March 25, 2022

Article in press: March 25, 2022

Published online: May 16, 2022



Hong-Jun Xu, Department of Dermatology, Beijing Friendship Hospital, Capital Medical University, Beijing 100050, China

Guang-Dong Wen, Department of Dermatology, Peking University People's Hospital, Peking University, Beijing 100044, China

Corresponding author: Hong-Jun Xu, MD, Chief Doctor, Department of Dermatology, Beijing Friendship Hospital, Capital Medical University, No. 95 Yong'an Road, Xicheng District, Beijing 100050, China. ink-008@163.com

Abstract

BACKGROUND

Porokeratosis is a rare, acquired, or inherited disorder of keratinization. There are numerous clinical types of porokeratosis and they can coexist in one patient and multiple members of an affected family. However, coexistence of disseminated superficial actinic porokeratosis (DSAP) and porokeratosis ptychotropica (Ppt) is rare.

CASE SUMMARY

A 45-year-old man presented with long-standing skin lesions. Physical examination identified numerous small, brown 2-mm to 4-mm patches on his face and several hyperkeratotic, verrucous plaques on his trunk and extremities. His father and one of his brothers also had similar lesions for years. Skin biopsies indicated a cornoid lamella in the epidermis. We identified c.155G>A mutation in the mevalonate kinase (MVK) gene, which converted a serine residue to asparagine (p.Ser52Asn) and was causative for porokeratosis in this family. A clinicopathologic diagnosis of DSAP and Ppt with a novel MVK gene mutation was made. The hyperkeratotic plaques on the patient's scrotum were completely removed more than 10 times using a microwave knife.

CONCLUSION

An unusual case of DSAP coexisting with Ppt harbored a novel MVK gene mutation also present in the patient's family.

Key Words: Disseminated superficial actinic porokeratosis; Porokeratosis ptychotropica; Mevalonate kinase gene; Gene mutation; Microwave knife; Case report

©The Author(s) 2022. Published by Baishideng Publishing Group Inc. All rights reserved.

Core Tip: Porokeratosis is a rare, acquired, or inherited disorder of keratinization. The coexistence of disseminated superficial actinic porokeratosis (DSAP) and porokeratosis ptychotropica (Ppt) is rare. We present an unusual case of DSAP coexisting with Ppt and identified a novel mevalonate kinase (MVK) gene mutation in this patient's family. To date, only four cases of DSAP coexisting with Ppt have been reported in the English literature. One case also had an MVK gene mutation. Thus, mutation of phosphomevalonate kinase pathway genes, especially the MVK gene, may have an important role in the pathogenesis of DSAP coexisting with Ppt.

Citation: Xu HJ, Wen GD. Mixed porokeratosis with a novel mevalonate kinase gene mutation: A case report. *World J Clin Cases* 2022; 10(14): 4528-4534

URL: <https://www.wjgnet.com/2307-8960/full/v10/i14/4528.htm>

DOI: <https://dx.doi.org/10.12998/wjcc.v10.i14.4528>

INTRODUCTION

Porokeratosis is a rare, acquired, or inherited disorder of keratinization, which presents as a keratotic papule or plaque with an annular ridge-like border[1]. Its main histological characteristic is a cornoid lamella, a thin column of parakeratosis leading to a ridge-like hyperkeratotic border. Numerous types of porokeratosis have been described and there are reports of more than one type of porokeratosis developing in the same patient and in multiple members of an affected family[2]. Inherited or sporadic genetic defects have an important role in porokeratosis. Correlations between gene mutations and clinical phenotypes of porokeratosis have been reported previously[3]. Porokeratosis lesions may alter a patient's appearance or function. Furthermore, some cases develop squamous cell carcinoma within the porokeratosis lesions. Currently, no therapeutic interventions with good efficacy are available although various topical, surgical, destructive, and systemic therapies appear to be effective in some patients. Here, we report a rare case of disseminated superficial actinic porokeratosis (DSAP) and porokeratosis ptychotropica (Ppt) associated with a mevalonate kinase (MVK) gene mutation which was successfully treated by surgery.

CASE PRESENTATION

Chief complaints

A 45-year-old man complained of long-standing skin lesions.

History of present illness

The lesions started as brown patches on his face and gradually spread to the trunk and extremities with verrucous plaques 30 years ago. Involvement of the scrotum occurred 20 years ago.

History of past illness

The patient had no medical history and reported no history of ultraviolet exposure, immunosuppression, or immunodeficiency caused by human immunodeficiency virus (HIV) infection, tumors, or drugs.

Personal and family history

The patient's father and one of his brothers had similar lesions for years.

Physical examination

Physical examination identified numerous small, brown 2-mm to 4-mm patches on his face (Figure 1A) and several hyperkeratotic, verrucous plaques on his trunk (Figure 1B) and extremities (Figure 1C). The widespread verrucous plaques with erosions and crust on his scrotum (Figure 1D) affected his normal life.

Laboratory examinations

Laboratory investigations including routine blood examination, hepatic and renal function, HIV antibody, *Treponema pallidum* hemagglutination test, rapid plasma reagin test, human papillomavirus, hepatitis B and C, tumor markers, and autoimmune screens were all negative.

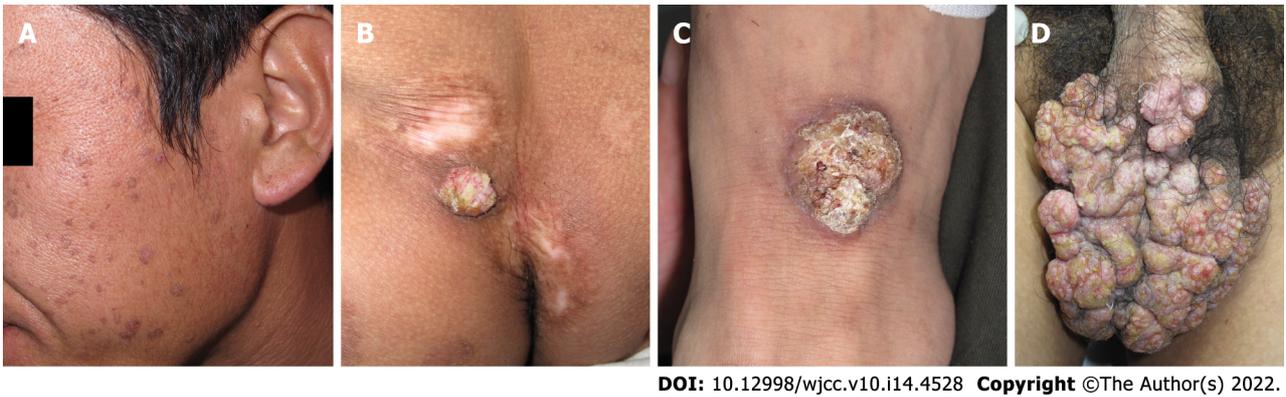


Figure 1 Skin lesions of the patient. A: Small, brown patches on the face. B-D: Hyperkeratotic, verrucous plaques on the trunk and extremities and widespread verrucous plaques with erosions and crust on the scrotum.

Imaging examinations

Skin biopsies taken from the hyperkeratotic plaques of lesions on his scrotum showed irregular acanthosis and papillomatosis as well as a cornoid lamella in the epidermis and the absence of the granular layer beneath it (Figure 2).

Gene testing

After obtaining informed consent, genomic DNA was extracted from the proband and his affected and unaffected family members, and all exons of four mevalonate pathway genes including MVK, mevalonate decarboxylase (MVD), phosphomevalonate kinase (PMVK), and farnesyl diphosphate synthase (FDPS) with intronic flanking sequences were amplified by PCR. In addition, genomic DNA from 100 normal healthy Chinese individuals was extracted as controls. Bidirectional sequencing identified a heterozygous synonymous mutation c.155G>A in exon 18 of the MVK gene, which was not present in the unaffected family members and controls (Figure 3). This c.155 G>A mutation in the MVK gene converted a serine residue to asparagine (p.Ser52Asn) and was the causative mutation for porokeratosis in this family.

FINAL DIAGNOSIS

A clinicopathologic diagnosis of DSAP and Ppt with MVK gene mutation was made.

TREATMENT

The hyperkeratotic plaques on the patient's scrotum were completely removed more than 10 times using a microwave knife (Figure 4).

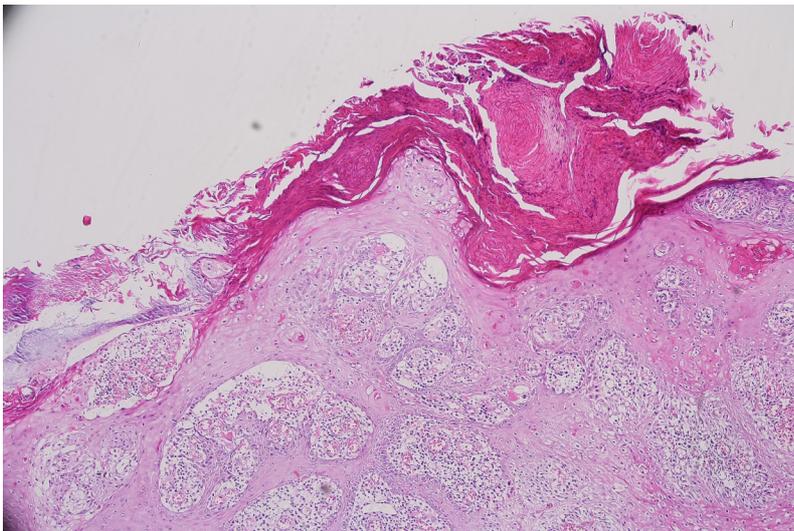
OUTCOME AND FOLLOW-UP

The patient has had no recurrence during 6-years of follow-up.

DISCUSSION

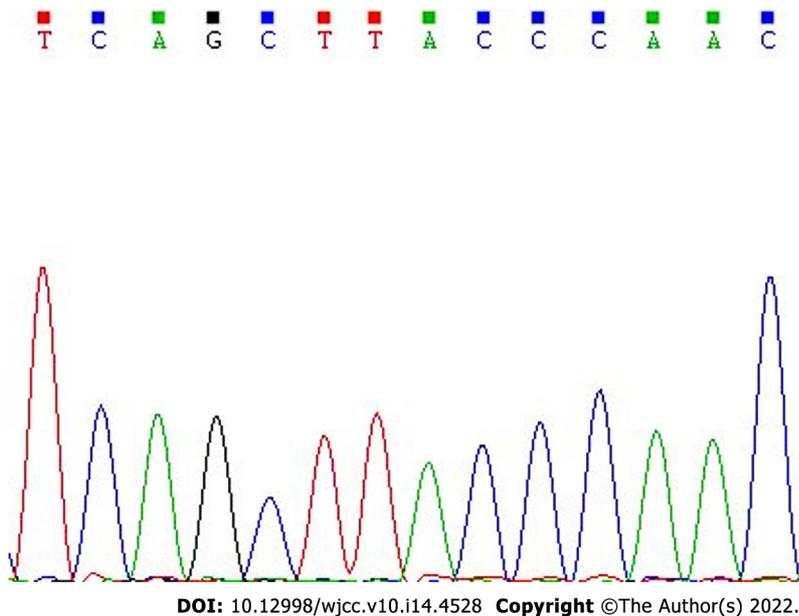
Porokeratosis represents a heterogeneous group of hereditary and acquired disorders of the clonal hyperproliferation of keratinocytes[4]. The characteristic ridge-like, keratotic border termed the "cornoid lamella" confirms the diagnosis of porokeratosis. Several clinical variants of porokeratosis have been described, all of which share this distinctive feature.

DSAP is the most common type of porokeratosis. It presents as keratotic papules with a well-demarcated elevated border, usually ranging from 3 mm to 10 mm in diameter. They are typically skin-colored to tan-brown to pink-red in color. DSAP usually occurs in the third or fourth decade of life, and patients frequently report a history of extensive exposure to ultraviolet radiation. For this reason, it usually occurs in body areas exposed to the sun, especially the shins and extensor forearms. Lesions on



DOI: 10.12998/wjcc.v10.i14.4528 Copyright ©The Author(s) 2022.

Figure 2 Histopathological examination shows a cornoid lamella in the epidermis (hematoxylin-eosin staining, original magnification: x100).



DOI: 10.12998/wjcc.v10.i14.4528 Copyright ©The Author(s) 2022.

Figure 3 Analysis of the mutation in the mevalonate kinase gene. A novel mevalonate kinase missense mutation (c.155G>A) was present in the patient's family.

our patient's face were typical of DSAP. Although 15% of DSAP patients have facial lesions, exclusively facial DSAP is an unusual clinical presentation[5]. Ppt is an unusual psoriasiform variant of porokeratosis[6-8], which typically presents with pruritic, red to brown, keratotic, or verrucous papules and plaques on the buttocks or genital skin[8]. Sometimes, these lesions can coalesce. Due to their clinical similarities, Ppt is often mistaken for psoriasis[9] and chronic eczema. Therefore, a skin biopsy is a useful method for differential diagnosis. The verrucous plaques on our patient's scrotum, buttocks, and limbs were diagnosed as Ppt according to the pathologic findings.

Previous studies have reported the coexistence of multiple types of porokeratosis in the same individual[10-20]; however, the co-occurrence of DSAP with Ppt is rare. To the best of our knowledge, this is the fifth case reported in the English literature to date[7,21-23] (Table 1).

The pathogenesis of porokeratosis is poorly understood. Risk factors include genetic susceptibility, exposure to ultraviolet radiation, and immunosuppression. Previous studies reported mutations in the phosphomevalonate kinase pathway genes, including MVD, MVK, PMVK, and FDPS in patients with porokeratosis[24,25]. At least one mutation in a mevalonate pathway gene was found in 98% of familial cases and in > 70% of sporadic porokeratosis cases[3]. Among the above five patients previously

Table 1 Clinical features of patients with coexisting disseminated superficial actinic porokeratosis and porokeratosis ptychotropica

Ref.	Gender	Age	Ethnicity	Nationality	Clinical variants	Genetics	Risk factors	Treatment
Thomas <i>et al</i> [21]	Female	78 yr	NA	United Kingdom	DSAP, Ppt	NA	None	NA
McGuigan <i>et al</i> [7]	Male	84 yr	NA	United States	DSAP, Ppt	NA	Sun exposure, non-melanoma skin cancer	0.025% tretinoin cream, 0.05% tazarotene cream.
Murase <i>et al</i> [22]	Female	73 yr	Korean	Korea	DSAP, Ppt linear porokeratosis	NA	Family history	NA
Peng <i>et al</i> [23]	Male	70 yr	Chinese	China	DSAP, Ppt	MVK missense mutation (c.1039G>C, p.Gly347Arg)	Family history	Oral acitretin (20 mg daily)
Our patient	Male	45 yr	Chinese	China	DSAP, Ppt	MVK missense mutation (c.155G>A, p.Ser52Asn)	Family history	Microwave knife

DSAP: Disseminated superficial actinic porokeratosis; Ppt: Porokeratosis ptychotropica; MVK: mevalonate kinase; NA: Not available.



DOI: 10.12998/wjcc.v10.i14.4528 Copyright ©The Author(s) 2022.

Figure 4 The lesions on the patient’s scrotum were completely removed after microwave knife treatment.

reported, two had genetic results. Peng *et al* [23] identified a novel MVK missense mutation (c.1039G>C, p.Gly347Arg) in a family with coexisting DSAP and Ppt. In our patient’s family, another MVK missense mutation (c.155G>A, p.Ser52Asn) was found. Although this mutation was previously reported as benign, we suggest it is a novel mutation associated with porokeratosis. Functional analysis of this mutation *in vitro* and *in vivo* should be performed in future studies. The findings in these two cases suggest that the mutation of phosphomevalonate kinase pathway genes, especially the MVK gene, might have an important role in the pathogenesis of DSAP coexisting with Ppt. Genetic analysis is useful in affected families to predict the occurrence of these lesions in other individuals of the same family.

Patients with porokeratosis have a 7.5%-10% risk of malignant transformation to squamous cell carcinoma or basal cell carcinoma [26]. Thus, clinical surveillance with regular skin examinations and patient education regarding the warning signs of skin cancer and sun protection are necessary aspects of the management in all patients with porokeratosis. However, some patients with functional impairments or appearance requirements want to have their porokeratosis lesions removed. There are five main types of treatment for porokeratosis: Topical or systemic drug therapy, surgical excision, cryotherapy, laser ablation, and photodynamic therapy. However, the disease commonly reoccurs. We used a microwave knife to remove the hyperkeratotic plaques on the patient’s scrotum. The patient did not show any functional impairment related to this treatment and did not develop disease aggravation or recurrence during 6-years of follow-up. This indicates that the microwave knife is an effective and

safe therapy for porokeratosis.

CONCLUSION

In conclusion, we report an unusual case of DSAP coexisting with Ppt and identified a novel MVK gene mutation in this patient's family. The microwave knife is an effective and safe therapy for porokeratosis and clinical surveillance for malignant transformation is necessary for all patients with porokeratosis.

FOOTNOTES

Author contributions: Xu HJ collected all the clinical data and wrote the draft manuscript. Wen GD completed the gene analysis; all authors read and approved the final manuscript.

Informed consent statement: Written informed consent was obtained from the patient for publication of this case report and any accompanying images. A copy of the written consent is available for review from the Editor of this journal.

Conflict-of-interest statement: The authors declare that they have no conflict of interest to report.

CARE Checklist (2016) statement: The authors have read the CARE Checklist (2016), and the manuscript was prepared and revised according to the CARE Checklist (2016).

Open-Access: This article is an open-access article that was selected by an in-house editor and fully peer-reviewed by external reviewers. It is distributed in accordance with the Creative Commons Attribution NonCommercial (CC BY-NC 4.0) license, which permits others to distribute, remix, adapt, build upon this work non-commercially, and license their derivative works on different terms, provided the original work is properly cited and the use is non-commercial. See: <https://creativecommons.org/licenses/by-nc/4.0/>

Country/Territory of origin: China

ORCID number: Hong-Jun Xu 0000-0002-4002-1549; Guang-Dong Wen 0000-0002-9221-7033.

S-Editor: Liu JH

L-Editor: Webster JR

P-Editor: Liu JH

REFERENCES

- 1 **Kanitakis J.** Porokeratoses: an update of clinical, aetiopathogenic and therapeutic features. *Eur J Dermatol* 2014; **24**: 533-544 [PMID: 25115203 DOI: 10.1684/ejd.2014.2402]
- 2 **Lucker GP, Steijlen PM.** The coexistence of linear and giant porokeratosis associated with Bowen's disease. *Dermatology* 1994; **189**: 78-80 [PMID: 8003795 DOI: 10.1159/000246791]
- 3 **Zhang Z, Li C, Wu F, Ma R, Luan J, Yang F, Liu W, Wang L, Zhang S, Liu Y, Gu J, Hua W, Fan M, Peng H, Meng X, Song N, Bi X, Gu C, Zhang Z, Huang Q, Chen L, Xiang L, Xu J, Zheng Z, Jiang Z.** Genomic variations of the mevalonate pathway in porokeratosis. *Elife* 2015; **4**: e06322 [PMID: 26202976 DOI: 10.7554/eLife.06322]
- 4 **Sertznig P, von Felbert V, Megahed M.** Porokeratosis: present concepts. *J Eur Acad Dermatol Venereol* 2012; **26**: 404-412 [PMID: 21929548 DOI: 10.1111/j.1468-3083.2011.04275.x]
- 5 **Sawyer R, Picou KA.** Facial presentation of disseminated superficial actinic porokeratosis. *Ear Nose Throat J* 1989; **68**: 57-59 [PMID: 2721409]
- 6 **Tallon B, Blumental G, Bhawan J.** Porokeratosis ptychotropica: a lesser-known variant. *Clin Exp Dermatol* 2009; **34**: e895-e897 [PMID: 20055860 DOI: 10.1111/j.1365-2230.2009.03664.x]
- 7 **McGuigan K, Shurman D, Campanelli C, Lee JB.** Porokeratosis ptychotropica: a clinically distinct variant of porokeratosis. *J Am Acad Dermatol* 2009; **60**: 501-503 [PMID: 19022531 DOI: 10.1016/j.jaad.2008.06.038]
- 8 **Yeo J, Winhoven S, Tallon B.** Porokeratosis ptychotropica: a rare and evolving variant of porokeratosis. *J Cutan Pathol* 2013; **40**: 1042-1047 [PMID: 24274427 DOI: 10.1111/cup.12233]
- 9 **De Simone C, Paradisi A, Massi G, Proietti I, Capponi A, Amerio PL, Capizzi R.** Giant verrucous porokeratosis of Mibelli mimicking psoriasis in a patient with psoriasis. *J Am Acad Dermatol* 2007; **57**: 665-668 [PMID: 17870431 DOI: 10.1016/j.jaad.2007.03.020]
- 10 **Moreland ME, Wyre HW Jr.** Porokeratosis. Two morphologic forms within a family. *Arch Dermatol* 1981; **117**: 245-246 [PMID: 7212751 DOI: 10.1001/archderm.117.4.245]
- 11 **Dover JS, Phillips TJ, Burns DA, Krafchik BR.** Disseminated superficial actinic porokeratosis. Coexistence with other porokeratotic variants. *Arch Dermatol* 1986; **122**: 887-889 [PMID: 3740871]

- 12 **Commens CA**, Shumack SP. Linear porokeratosis in two families with disseminated superficial actinic porokeratosis. *Pediatr Dermatol* 1987; **4**: 209-214 [PMID: 3321004 DOI: 10.1111/j.1525-1470.1987.tb00780.x]
- 13 **Gautam RK**, Bedi GK, Schgal VN, Singh N. Simultaneous occurrence of disseminated superficial actinic porokeratosis (DSAP), linear, and punctate porokeratosis. *Int J Dermatol* 1995; **34**: 71-72 [PMID: 7896495 DOI: 10.1111/j.1365-4362.1995.tb04387.x]
- 14 **Freyschmidt-Paul P**, Hoffmann R, König A, Happel R. Linear porokeratosis superimposed on disseminated superficial actinic porokeratosis: report of two cases exemplifying the concept of type 2 segmental manifestation of autosomal dominant skin disorders. *J Am Acad Dermatol* 1999; **41**: 644-647 [PMID: 10495390]
- 15 **Suh DH**, Lee HS, Kim SD, Cho KH, Kim KH, Park KC. Coexistence of disseminated superficial porokeratosis in childhood with congenital linear porokeratosis. *Pediatr Dermatol* 2000; **17**: 466-468 [PMID: 11123781 DOI: 10.1046/j.1525-1470.2000.01823.x]
- 16 **Kaur S**, Thami GP, Mohan H, Kanwar AJ. Co-existence of variants of porokeratosis: a case report and a review of the literature. *J Dermatol* 2002; **29**: 305-309 [PMID: 12081163 DOI: 10.1111/j.1346-8138.2002.tb00268.x]
- 17 **Boente Mdel C**, López-Baró AM, Frontini Mdel V, Asial RA. Linear porokeratosis associated with disseminated superficial actinic porokeratosis: a new example of type II segmental involvement. *Pediatr Dermatol* 2003; **20**: 514-518 [PMID: 14651573 DOI: 10.1111/j.1525-1470.2003.20613.x]
- 18 **Pearson IC**, Cliff S. Case 6: plaques extending in a linear pattern from left ankle to hip forming over a 2-year period. Diagnosis: linear porokeratosis with disseminated superficial porokeratosis erupting in pregnancy. *Clin Exp Dermatol* 2003; **28**: 345-346 [PMID: 12780739 DOI: 10.1046/j.1365-2230.2003.01278.x]
- 19 **Suárez-Amor O**, Pereiro-Ferreirós M, Ginarte M, Peteiro C, Toribio J. Coexistence of linear porokeratosis and disseminated superficial actinic porokeratosis: a type 2 segmental manifestation. *Acta Derm Venereol* 2007; **87**: 363-364 [PMID: 17598043 DOI: 10.2340/00015555-0215]
- 20 **Palleschi GM**, Torchia D. Porokeratosis of Mibelli and superficial disseminated porokeratosis. *J Cutan Pathol* 2008; **35**: 253-255 [PMID: 18190456 DOI: 10.1111/j.1600-0560.2007.00787.x]
- 21 **Thomas C**, Ogboli MI, Carr RA, Charles-Holmes R. Hypertrophic perianal porokeratosis in association with superficial actinic porokeratosis of the leg. *Clin Exp Dermatol* 2003; **28**: 676-677 [PMID: 14616847 DOI: 10.1046/j.1365-2230.2003.01410.x]
- 22 **Murase J**, Gilliam AC. Disseminated superficial actinic porokeratosis co-existing with linear and verrucous porokeratosis in an elderly woman: Update on the genetics and clinical expression of porokeratosis. *J Am Acad Dermatol* 2010; **63**: 886-891 [PMID: 20451293 DOI: 10.1016/j.jaad.2009.07.038]
- 23 **Peng JM**, Xiao XM, Chen JW, Chen LF, Cheng B, Ji MK, Zhang ZH. Novel mutation in MVK gene for co-occurrence of disseminated superficial actinic porokeratosis with porokeratosis ptychotropica. *J Dermatol* 2021; **48**: e137-e139 [PMID: 33458876 DOI: 10.1111/1346-8138.15748]
- 24 **Cui H**, Li L, Wang W, Shen J, Yue Z, Zheng X, Zuo X, Liang B, Gao M, Fan X, Yin X, Shen C, Yang C, Zhang C, Zhang X, Sheng Y, Gao J, Zhu Z, Lin D, Zhang A, Wang Z, Liu S, Sun L, Yang S, Cui Y. Exome sequencing identifies SLC17A9 pathogenic gene in two Chinese pedigrees with disseminated superficial actinic porokeratosis. *J Med Genet* 2014; **51**: 699-704 [PMID: 25180256 DOI: 10.1136/jmedgenet-2014-102486]
- 25 **Leng Y**, Yan L, Feng H, Chen C, Wang S, Luo Y, Cao L. Mutations in mevalonate pathway genes in patients with familial or sporadic porokeratosis. *J Dermatol* 2018; **45**: 862-866 [PMID: 29722423 DOI: 10.1111/1346-8138.14343]
- 26 **Le C**, Bedocs PM. Disseminated Superficial Actinic Porokeratosis. StatPearls. Treasure Island (FL): StatPearls Publishing Copyright © 2021, StatPearls Publishing LLC. 2021



Published by **Baishideng Publishing Group Inc**
7041 Koll Center Parkway, Suite 160, Pleasanton, CA 94566, USA
Telephone: +1-925-3991568
E-mail: bpgoffice@wjgnet.com
Help Desk: <https://www.f6publishing.com/helpdesk>
<https://www.wjgnet.com>

