



HAL
open science

A swollen cheek

M. Schlund, R.H. Khonsari, C. Leverd, B. Catteau, J. Ferri, S. Touzet-Roumazeille

► **To cite this version:**

M. Schlund, R.H. Khonsari, C. Leverd, B. Catteau, J. Ferri, et al.. A swollen cheek. *Journal of Stomatology, Oral and Maxillofacial Surgery*, 2020, 121, pp.461 - 462. <10.1016/j.jormas.2019.12.020>. <hal-03491285>

HAL Id: hal-03491285

<https://hal.science/hal-03491285v1>

Submitted on 21 Sep 2022

HAL is a multi-disciplinary open access archive for the deposit and dissemination of scientific research documents, whether they are published or not. The documents may come from teaching and research institutions in France or abroad, or from public or private research centers.

L'archive ouverte pluridisciplinaire **HAL**, est destinée au dépôt et à la diffusion de documents scientifiques de niveau recherche, publiés ou non, émanant des établissements d'enseignement et de recherche français ou étrangers, des laboratoires publics ou privés.



Distributed under a Creative Commons CC BY-NC 4.0 - Attribution - Non-commercial use - International License

A swollen cheek

Matthias Schlund, Roman Hossein Khonsari, Caroline Leverd, Benoît Catteau, Joël Ferri, Sandrine Touzet-Roumzeille

Matthias Schlund, MD, MSc, Chef de Clinique des Universités – Assistant Hospitalier
-Univ. Lille, CHU Lille, INSERM, Service de Chirurgie Maxillo-Faciale et Stomatologie, U1008 - Controlled Drug Delivery Systems and Biomaterials, F-59000 Lille, France

Roman Hossein Khonsari, MD, PhD, Maître de Conférence des Universités – Praticien Hospitalier
-Service de chirurgie maxillofaciale et plastique, Hôpital Universitaire Necker - Enfants Malades, Assistance Publique - Hôpitaux de Paris ; Centre de Référence Maladies Rares des Fentes et Malformations Faciales MAFACE ; Filière Maladies Rares TeteCou ; Université Paris - Descartes, Université Sorbonne Paris Cité ; Paris, France

Caroline Leverd, DDS, Assistante Hospitalière
-Univ. Lille, CHU Lille, Service de pédodontie, F-59000 Lille, France

Benoît Catteau, MD, Praticien Hospitalier
-Univ. Lille, CHU Lille, Service de Dermatologie, F-59000 Lille, France

Joël Ferri, MD, PhD, Professeur des Universités – Praticien Hospitalier
-Univ. Lille, CHU Lille, INSERM, Service de Chirurgie Maxillo-Faciale et Stomatologie, U1008 - Controlled Drug Delivery Systems and Biomaterials, F-59000 Lille, France

Sandrine Touzet-Roumzeille, MD, MSc, Praticien Hospitalier
-Univ. Lille, CHU Lille, Service de Chirurgie Maxillo-Faciale et Stomatologie, F-59000 Lille, France

Corresponding author :
Matthias Schlund
Service de Chirurgie Maxillo-Faciale et Stomatologie
Hôpital Roger Salengro
Rue Emile Laine
59037-Lille-Cedex
France
Tel : +33320443676
Fax : +33320445860
e-mail : matthias.schlund@chru-lille.fr

Case Report

A 9-year-old girl first presented in our department with a non-tender, non-inflammatory lump of the left cheek. The left cheek, the left nasal ala and left upper lip were swollen, whereas the left eyelid and lower lip were normal (Figure 1). This swelling was first discovered during her first year of life and gradually increased with growth. There was no overall growth impairment. Oral examination showed a median maxillary mucosal lesion. The patient had previously presented with similar lesions, which had been surgically removed and diagnosed as neuromas based on histology. There was early teeth eruption of the left maxillary teeth, which was confirmed on the orthopantomogram (Figure 2). There were no other systemic lesions apart from a café-au-lait macule on the right flank. MRI imaging showed isolated fatty tissue enlargement on the affected side without another anomaly (Figure 3). The mucosal lesion was removed and was a neuroma, in line with previous histological results.

What is your diagnosis?

Discussion

The patient presented with facial infiltrating lipomatosis (FIL), which is a mosaic overgrowth disorder. FIL is characterized by soft tissue and/or skeletal overgrowth, early teeth eruption, macrodontia, macroglossia and mucosal neuromas[1]. FIL is a rare disease (prevalence <1/1000000) part of the *PIK3CA*-Related Overgrowth Spectrum (PROS), which encompasses all the clinical syndromes associated with somatic *PIK3CA* mutations, including fibroadipose hyperplasia, hemihyperplasia with multiple lipomatosis, CLOVES syndrome (Congenital Lipomatous Overgrowth, Vascular malformations, Epidermal nevi, Scoliosis/skeletal and spinal syndrome), fibroadipose infiltrating lipomatosis, megalencephaly-capillary malformation, and dysplastic megalencephaly[2]. *PIK3CA* is part of the Phosphoinositide 3-Kinase (PI3K)/AKT/mTOR pathway. Other mutations of genes involved in this pathway can cause different overgrowth syndromes such as Proteus syndrome. Overactivation of this pathway leads to competitive cellular growth, metabolic advantage and angiogenesis[3].

The diagnosis of FIL was confirmed by genomic sequencing (Sanger sequencing) performed on the mucosal lesion, which showed a *PIK3CA* missense mutation (exon 8, c.1258T>C leading to the substitution of the cysteine 420 with arginine). There were no mutations in the genes *PTEN* and *AKT1*. It is important to harvest the overgrown tissue as it is the only tissue harboring the mutation in this mosaic somatic genetic disease.

The diagnosis of PROS is crucial for patient management since new treatments for this spectrum of conditions are now available. Surgery was for a long time the only option to treat these patients but several steps were often required with variable outcomes, and the recurrence rate was high[4]. Targeted therapies such as BYL719, a PI3K α inhibitor, initially developed for cancers with *PIK3CA* mutations, is currently being assessed for PROS.

BYL719 has been successfully used in patients with CLOVES syndrome[5] and is undergoing further investigations.

References

- [1] Maclellan RA, Luks VL, Vivero MP, Mulliken JB, Zurakowski D, Padwa BL, et al. PIK3CA activating mutations in facial infiltrating lipomatosis. *Plast Reconstr Surg* 2014;133:12e – 9e. doi:10.1097/01.prs.0000436822.26709.7c.
- [2] Keppler-Noreuil KM, Rios JJ, Parker VER, Semple RK, Lindhurst MJ, Sapp JC, et al. PIK3CA-related overgrowth spectrum (PROS): diagnostic and testing eligibility criteria, differential diagnosis, and evaluation. *Am J Med Genet A* 2015;167A:287–95. doi:10.1002/ajmg.a.36836.
- [3] Porta C, Paglino C, Mosca A. Targeting PI3K/Akt/mTOR Signaling in Cancer. *Front Oncol* 2014;4:64. doi:10.3389/fonc.2014.00064.
- [4] Serpa MS, Scully C, Molina Vivas AP, de Almeida OP, Costa FD, Alves FA. Infiltrating lipomatosis of the face: case series and literature review. *Oral Surg Oral Med Oral Pathol Oral Radiol* 2017;123:e99–105. doi:10.1016/j.oooo.2016.10.009.
- [5] Venot Q, Blanc T, Rabia SH, Berteloot L, Ladraa S, Duong J-P, et al. Targeted therapy in patients with PIK3CA-related overgrowth syndrome. *Nature* 2018;558:540–6. doi:10.1038/s41586-018-0217-9.

List of figures

Figure 1: Swollen left cheek, nasal ala and upper lip

Figure 2:

- A. Midline mucosal lesion, later confirmed as a neuroma after surgical resection and pathological examination
- B. Orthopantomogram showing early teeth eruption in the left maxilla

Figure 3: Isolated enlargement of the fatty tissues of the left cheek on T1-weighted MRI





