Sindrome CLAPO

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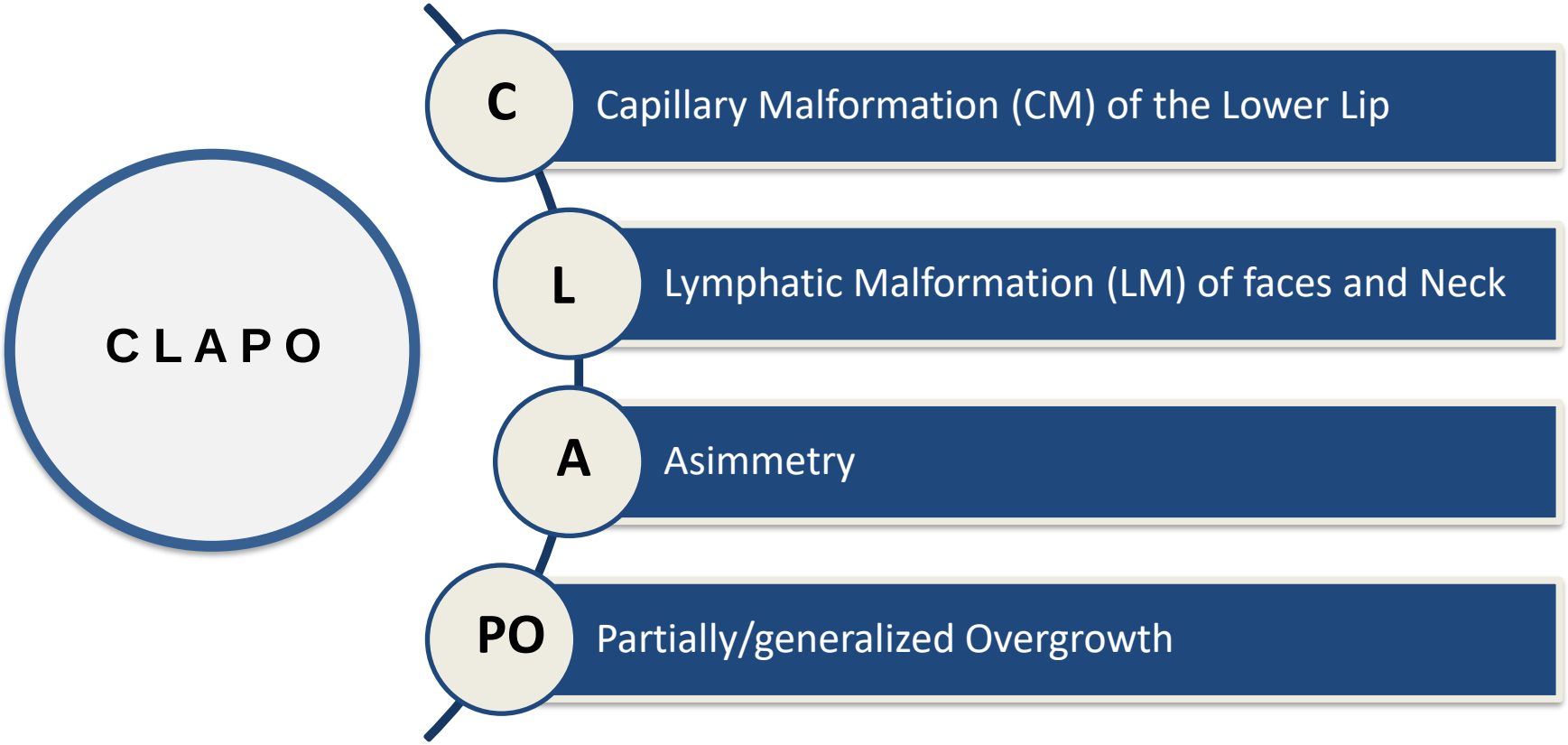
SISAV
SOCIETÀ ITALIANA PER LO STUDIO DELLE ANOMALIE VASCOLARI

1869 2019
Bambino Gesù
OSPEDALE PEDIATRICO


C L A P O
C
Capillary Malformation (CM) of the Lower Lip
L
Lymphatic Malformation (LM) of faces and Neck
A
Asimmetry
PO
Partially/generalized Overgrowth

ISSVA classification for vascular anomalies ©

(Approved at the 20th ISSVA Workshop, Melbourne, April 2014, last revision May 2018)

This classification is intended to evolve as our understanding of the biology and genetics of vascular malformations and tumors continues to grow

Vascular malformations associated with other anomalies		
Klippel-Trenaunay syndrome: *	CM + VM +/- LM + limb overgrowth	PIK3CA
Parkes Weber syndrome:	CM + AVF + limb overgrowth	RASA1
Servelle-Martorell syndrome:	limb VM + bone undergrowth	
Sturge-Weber syndrome:	facial + leptomeningeal CM + eye anomalies +/- bone and/or soft tissue overgrowth	GNAQ
Limb CM + congenital non-progressive limb overgrowth		GNA11
Maffucci syndrome:	VM +/- spindle-cell hemangioma + enchondroma	IDH1 / IDH2
Macrocephaly - CM (M-CM / MCAP) *		PIK3CA
Microcephaly - CM (MICCAP)		STAMBP
CLOVES syndrome: *	LM + VM + CM +/- AVM + lipomatous overgrowth	PICK3CA
Proteus syndrome:	CM, VM and/or LM + asymmetrical somatic overgrowth	AKT1
Bannayan-Riley-Ruvalcaba sd:	AVM + VM +macrocephaly, lipomatous overgrowth	PTEN
CLAPO syndrome: *	lower lip CM + face and neck LM + asymmetry and partial/generalized overgrowth	PIK3CA

Unknown Eziology

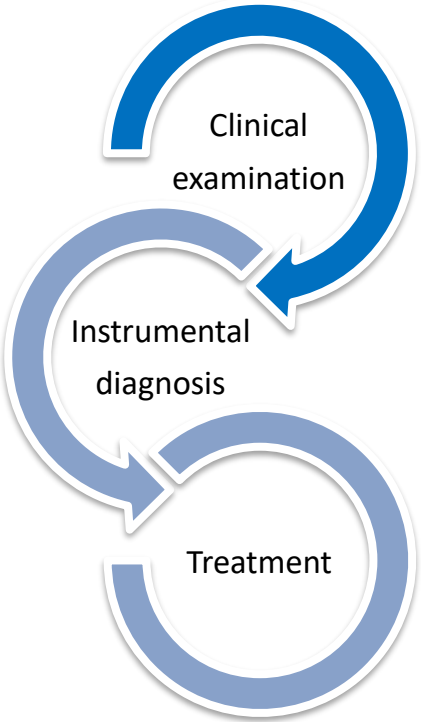
- OSCVA - *overgrowth syndromes with complex vascular anomalies.*
- 2018 Rodriguez – Laguna: Somatic activating PIK3CA mutations → PROS - *PIK3CA overgrowth spectrum*

8 cases reported in scientific literature

- 2008 Lopez- Gutierrez et al.: first 6 cases
- 2016 Kramer: 1 case
- 2018 Flores-Terry: 1 case

Spectrum of abnormalities in CLAPO SYndr.

- 2018 Roè-Crespo: 1 case
- 2018 Gonzalez-Hermosa: 1 case

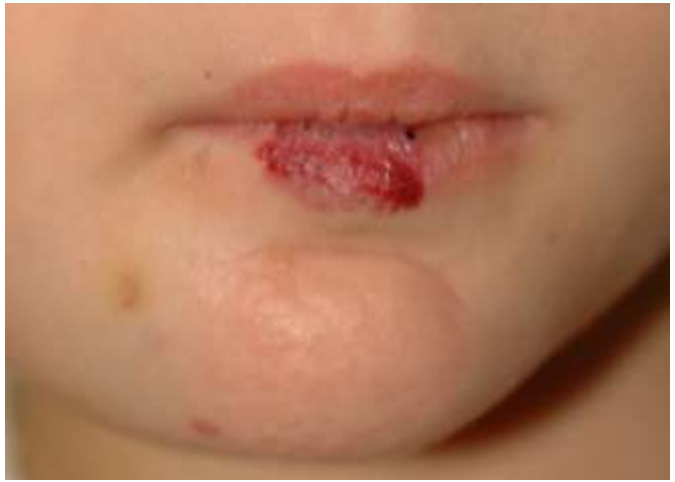


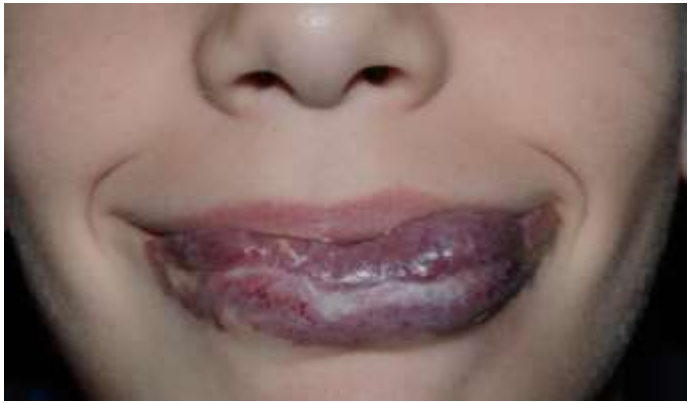
1°: CM of the lower lip

2°: presence of LM often in association with VM

Asimmetry

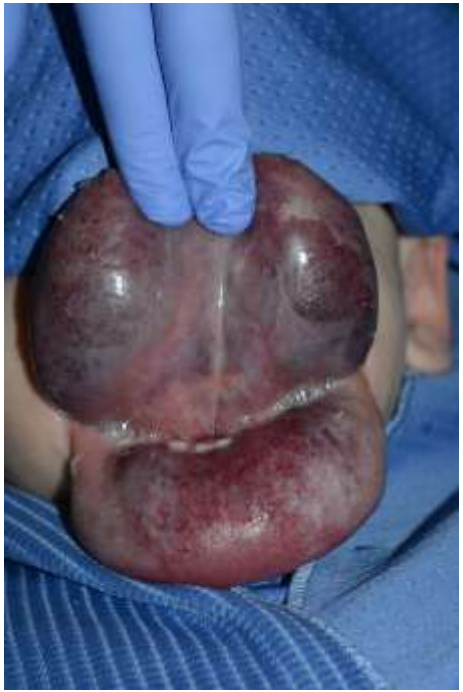
Overgrowth

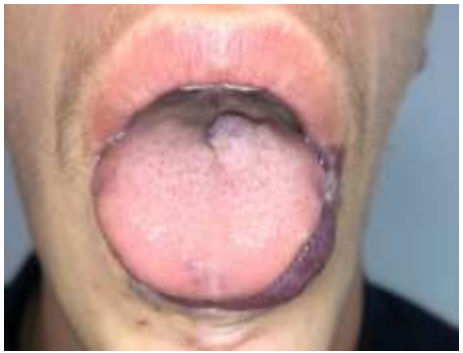




CM of the lower lip:

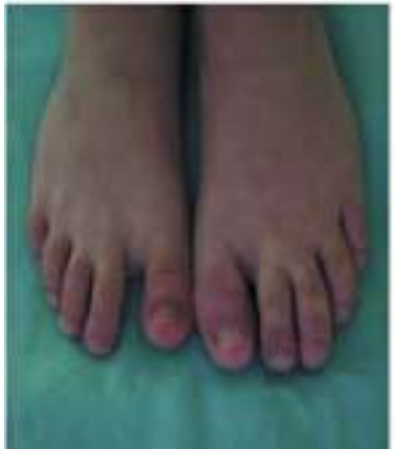
- always present at birth
- present in the midline
- with a predominant brown/purple color
- with a symmetrical distribution
- ranging from 2 to 11 cm
- with well-defined borders in several patients
- often affecting the portion of the skin under the lip or the intra-oral mucosa

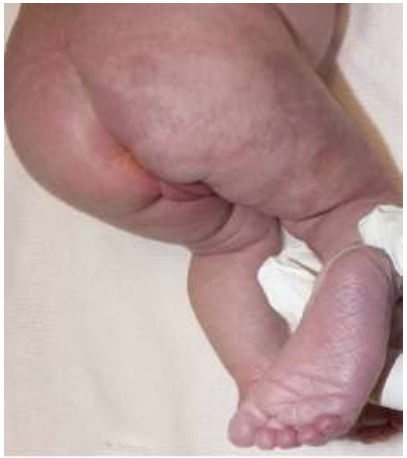




LM involving lip, oral mucosa, neck, and tongue:

- Often associated with VM
- Not always evident at birth
- Complaints and morbidity included bleeding, infection, swelling, vesicle formation and malocclusion.





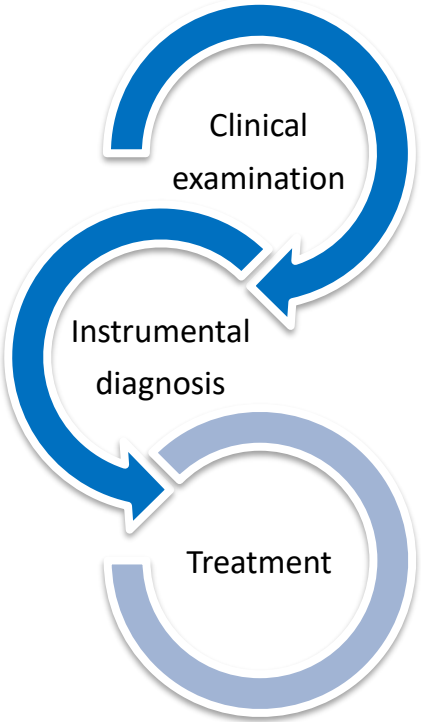
Asimmetry:

- Face or limbs

Overgrowth:

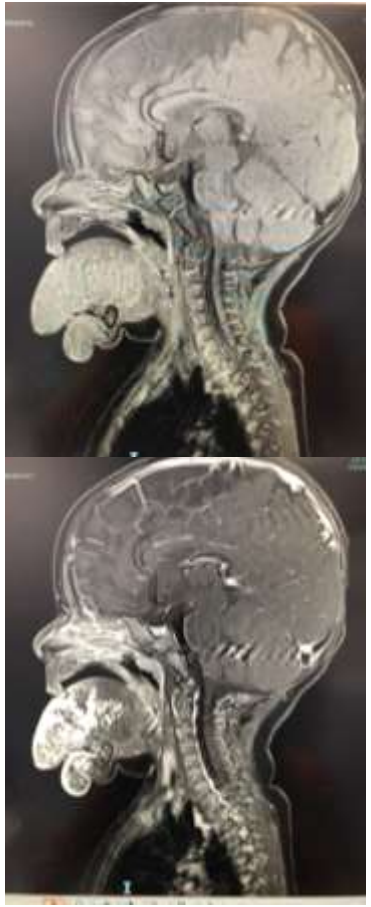
- not always evident from the beginning
- Partially /generalized

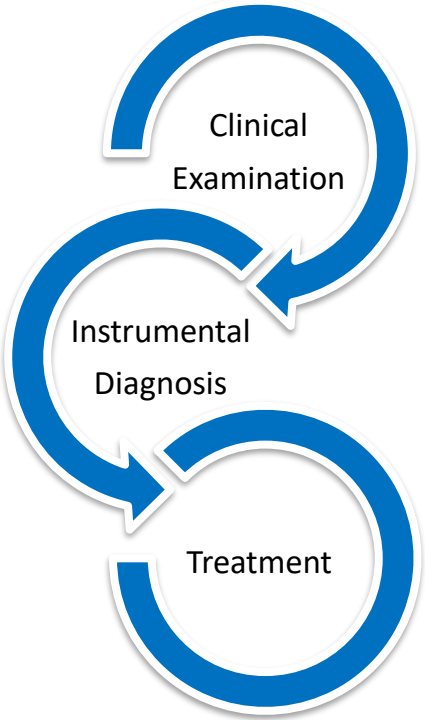
*Flores-Terry MÁ, Zamberk-Majlis P, Cortina-de la Calle MP, García-Arpa M. CLAPO Syndrome. Actas Dermosifiliogr. 2018;109:180.
* González-Hermosa MR, Guerra E, Tuduri I, Vicente I, López-Almaraz R. CLAPO syndrome: Effective response to treatment with oral rapamycin. Dermatologic Therapy. 2019;32:e12991



→ MRI of overgrowth segments and/or CM, LM, VM

→ Doppler Ultrasound

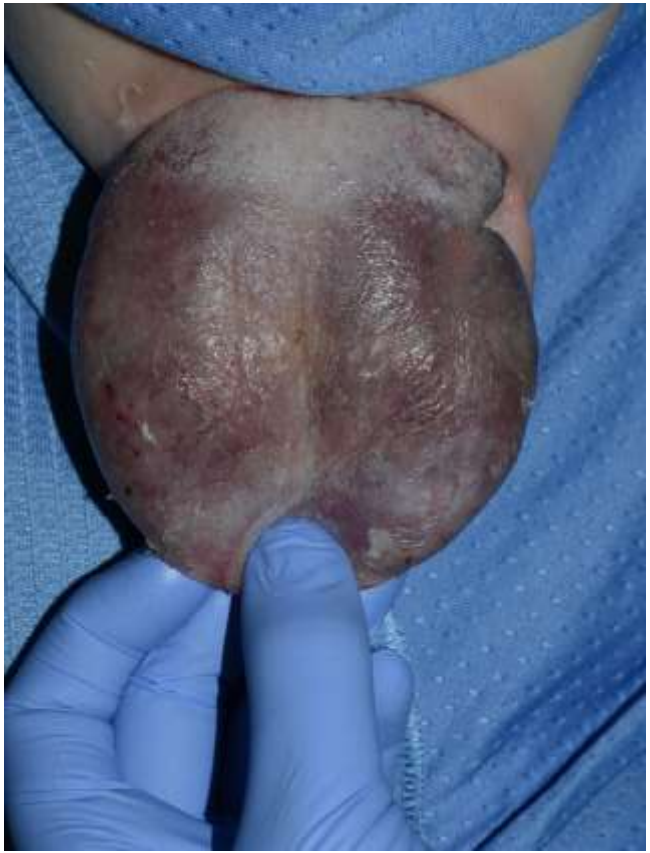


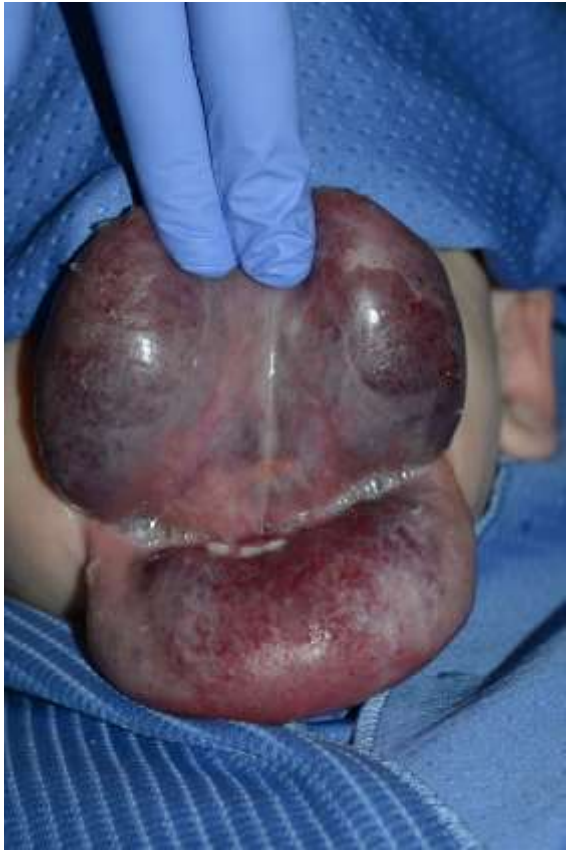


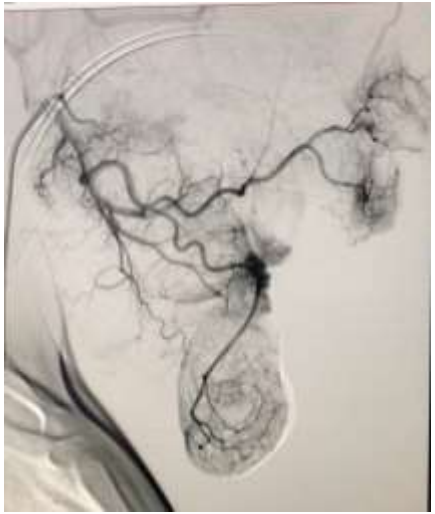
Oral Rapamycin* → Sclerotherapy, CO_2 laser photocoagulation → Surgical treatment

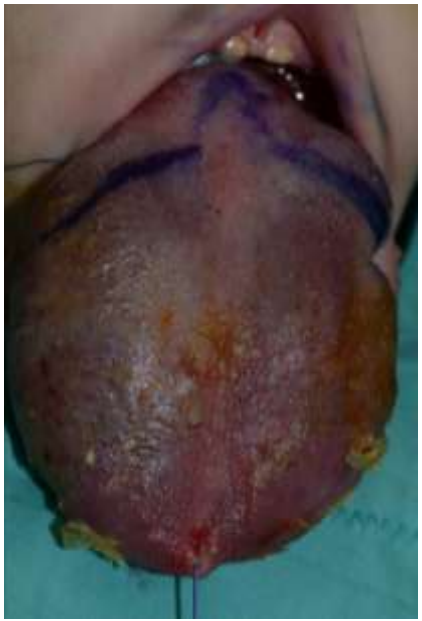
* González-Hermosa MR, Guerra E, Tuduri I, Vicente I, López-Almaraz R. CLAPO syndrome: Effective response to treatment with oral rapamycin. Dermatologic Therapy. 2019;32:e12991

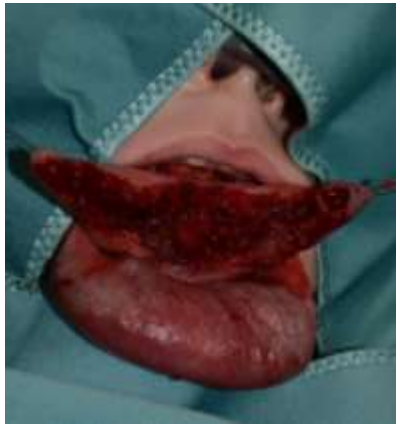
Clinical Case

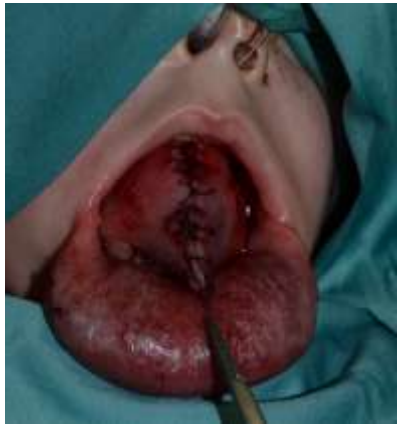


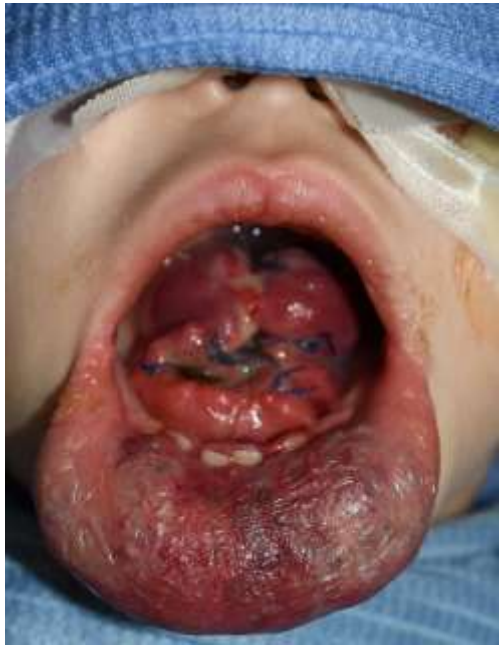


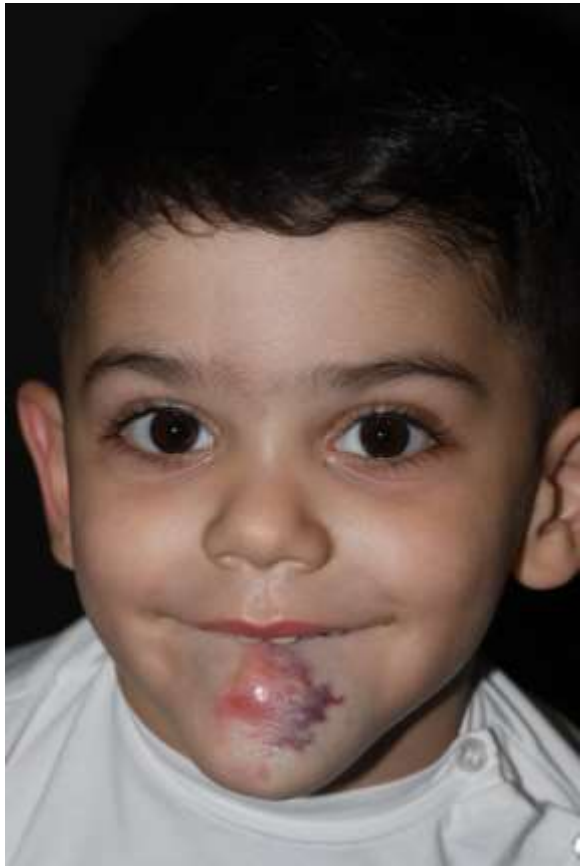












Grazie

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