# Somatic PIK3CA mutations cause hemifacial myohyperplasia: Alpesilib as a targeted medical treatment

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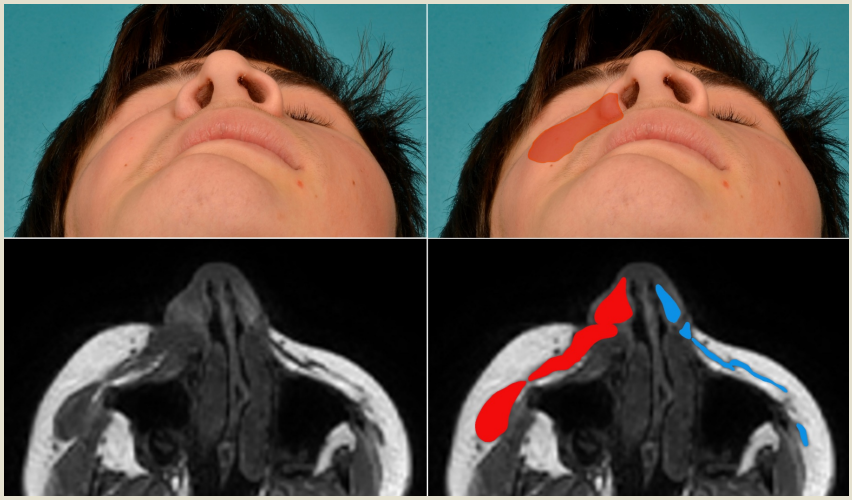
We report 6 patients affected by a particular kind of congenital facial asymmetry characterized by hemifacial muscular hypertrophy. This condition has been previously reported in literature as hemifacial myohyperplasia[1]. There is no currently satisfactory treatment for this disease.

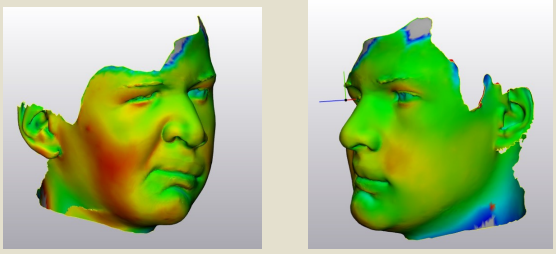
| Characteristics | 1 | 2 | 3 | 4 | 5 | 6 |
|---|---|---|---|---|---|---|
| Sex | F | F | F | M | F | M |
| Side | R | L | L | R | R | L |
| Comorbidities | - | - | + | - | - | - |
| Chin skin dimpling | + | + | + | + | + | + |
| Chin deviation | + | + | + | + | - | + |
| Auricular anomalies | + | + | + | + | - | + |
| Nasal deviation | - | - | + | + | - | + |
| Small nasal vestibule | + | + | + | + | + | + |
| Narrow palpebral fissure | + | + | + | + | - | + |
| Eyebrow dropping | + | + | + | + | - | + |
| Lip commissure canting | - | - | + | + | + | + |



All patients benefited from MRI and electromyography. Four patients had buccinator muscle biopsy, which all showed PIK3CA mutation[2].

One patient is currently under Alpelisib treatment with encouraging results. The superimposition of 3D pictures before and after 45 days of treatment shows volume decrease of the right cheek.

We characterized the origin and the clinical course of a little-known syndrome and provide arguments for a promising medical treatment.



**1** Lee S, Sze R, Murakami C, et al.. Hemifacial myohyperplasia: description of a new syndrome. Am J Med Genet. 2001 Nov 1;103(4):326-33.
**2** Venot Q, Blanc T, Rabia SH, et al.. Targeted therapy in patients with PIK3CA-related overgrowth syndrome. Nature. 2018 Jun;558(7711):540-546