**Supplementary Table 1 -** Phenotypic and genotypic features of patients with clinical segmental odontomaxillary dysplasia and *ACTB* mosaicism.

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| **Patient** | **Clinical Features** | **Histology** | **Tissue genotyped** | **Variant coordinates** |
| 1 | * Slightly thickened right upper lip
* Subtly increased

pigmentation with a strict midline cutoff* Dry area on the right lower cheek with increased follicularity
* Overgrowth of right upper gums and increased spacing between the teeth
 | Not performed | Skin DNA extracted directly | chr7:5568275 G>A NM\_001101.3(ACTB\_v001):c.439C>T p.(Arg147Cys) |
| 2  | * Subtle facial asymmetry
* Hypertrichotic area, right cheek
* Subtle right lip thickening and distortion with discontinuity of vermilion border
* Anteriorly displaced right upper tooth with gum hyperplasia
 | Mild patchy hyperkeratosis and widespread superficial perivascularchronic inflammation, accompanied by pigment incontinence and a few basal epidermal apoptotic bodies. | Skin DNA extracted directly | chr7:5568275 G>A NM\_001101.3(ACTB\_v001):c.439C>T p.(Arg147Cys) |
| 3 | * Left facial undergrowth
* Left commissural lip fissure
* Hypertrichosis of the left upper lip and under the left eye
 | Mild epidermal atrophy, mild hyalinization of the superficial dermis and mild perivascular lymphocytic infiltrate.” | DNA extracted from FFPE skin biopsy. Lymphocyte DNA extracted directly from whole blood. | **Skin:**chr7:5568275 G>A NM\_001101.3(*ACTB*\_v001):c.439C>T p.(Arg147Cys)**Lymphocyte DNA:**Mutation negative |
| 4 | * Hypertrichotic area right cheek
* Subtle thickening right lower cheek with mild increase in pigmentation
 | Increased collagen fibres (van-Gieson elastica-staining),and multiple musculi arrectores pilorum (Alpha-SMA staining) | DNA extracted FFPE skin | chr7:5568275 G>A NM\_001101.3(ACTB\_v001):c.439C>T p.(Arg147Cys) |