Oral Phenotype of Singleton-Merten Syndrome: A Case Report

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AIMS : Singleton-Merten syndrome is a rare autosomal dominant disorder with blood vessels calcifications, teeth anomalies and bone defects. We aim to describe the oral features of a Singleton-Merten patient, the diagnosis and the treatment he received.

METHOD : A 10-years-old patient presented at the pediatric dentistry department for an emergency. After a clinical and radiological examination, he was referred to the Reference Center of Rare Oral and Dental Diseases. In order to investigate the diagnosis, he received a genetic consultation.

RESULTS : Whole genome sequencing analysis revealed a heterozygous *de novo* variant in *IFIH1* (NM_022168.4) c.2465G>A (p.Arg822Gln) and confirmed a Singleton-Merten syndrome. We carried out the necessary health education and care, a partial denture replaced the hopeless teeth and maintained the vertical dimension.

Figure 1: Extra-oral photographies: dysmorphic facial features, fine and space hair, trigonocephaly with triangular face, discrete hypertelorism, long arched eyebrow, low set-ears, muscle weakness, cutaneous xerosis and ophtalmologic glaucoma, without intellectual disability

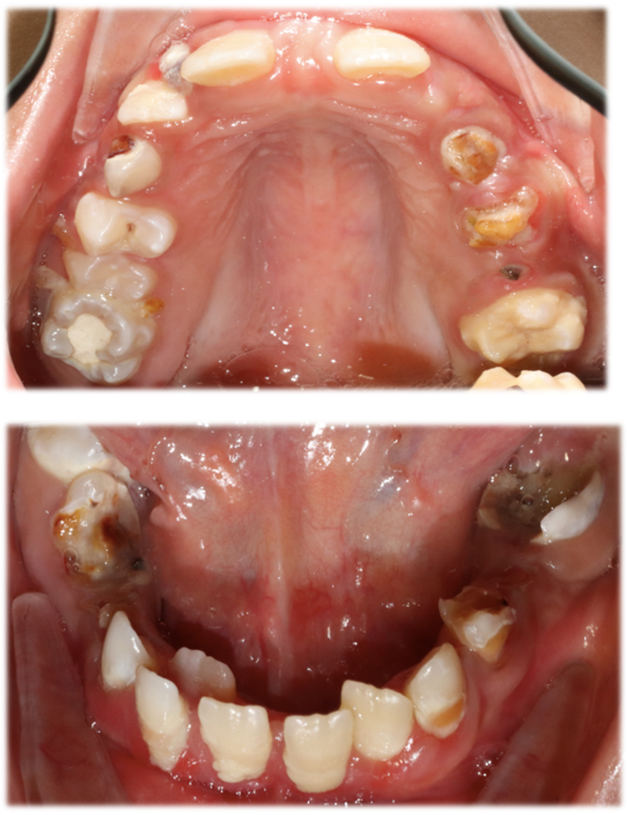

Figure 3: Intra-oral initial photographies: numerous carious lesions, severe teeth malposition espeacially in the anterior arch, hygiene defiency with a 100% plaque index, dental and jawbones anomalies

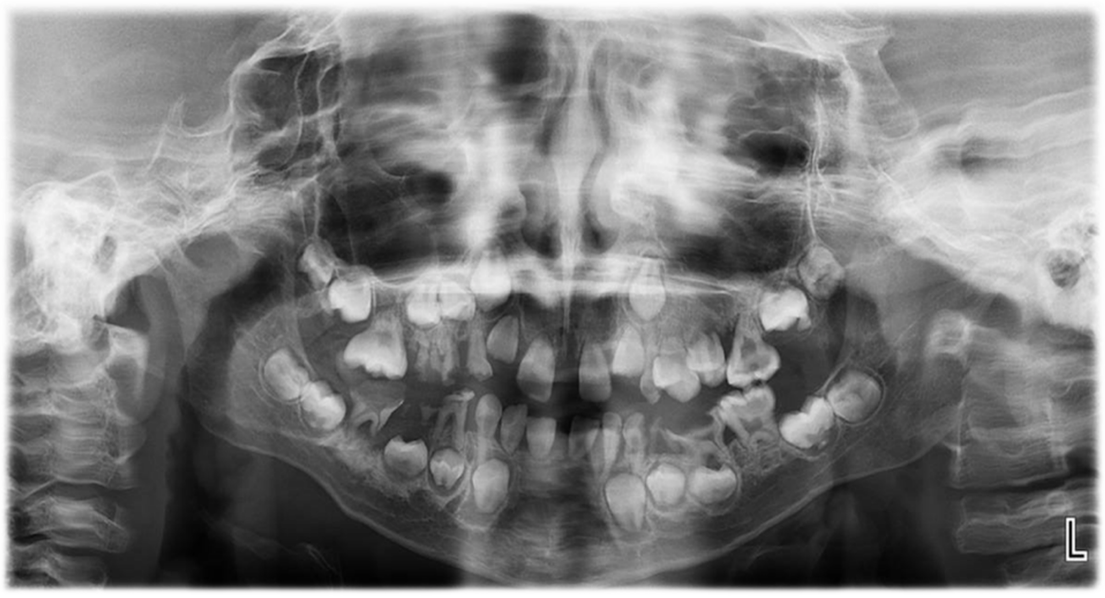

Figure 2: Initial X-ray panoramic at 10 years old : no dental agenesis, short roots, decrease of the alveolar bone height

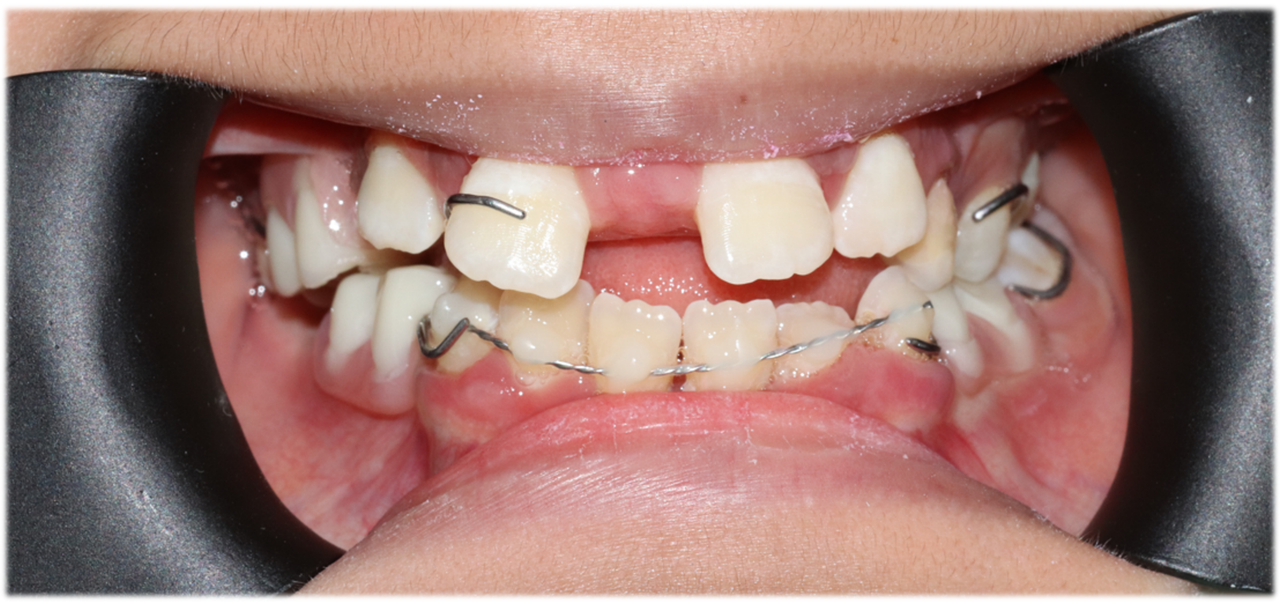

Figure 4: Final intra-oral photographie; We carried out the necessary health education, care and a periodontal splinting, while a partial denture replaced the hopeless teeth and maintained the vertical dimension.

Location	Phenotype	Phenotype MIM number	Inheritance	Gene/Locus	Gene/Locus MIM number
2q24.2	Singleton-Merten type 1	182250	AD	IFIH1	606951

CONCLUSION

Few descriptions of oral features occurred in literature between congenital findings and “acquired” pathologies, such as missing teeth. The oral phenotype of these patients remains insufficiently described, while it may contribute to diagnosis. Early diagnosis may prevent delayed teeth loss and improve quality of life.