

# Orthodontic management of patient with Silver–Russell Syndrome (SRS). A case report



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## Abstract

**Background** SRS is classified as a rare syndrome with an estimated incidence of 1 in 30.000/100.000 [Christoforidis A. et al., 2005]. It's a clinically and genetically heterogeneous disorder that presents a very wide phenotypic range. Due to its heterogeneity, SRS diagnosis is difficult, and the disease is probably underdiagnosed [Eggermann T. et al., 2009].

**Case report** M., a 7-year-old patient affected by SRS syndrome, comes to the first visit with a history of pain in the upper retroincisive gum due to the deep bite (gingival impingement). The pain prevents the correct chewing during meals and makes the orthodontic treatment necessary.

**Conclusions** The elaboration of a personal orthodontic treatment plan allows the patient to recover the correct masticatory function and improve her facial aesthetic.

## Introduction

The term Silver Russel Syndrome (SRS) describes a primary clinical condition characterized by facial features, intrauterine growth restriction (IUGR), poor postnatal growth, relative macrocephaly and body asymmetry [Perkins RM et al, 2002]. Two main molecular abnormalities are diagnosed at the origin of disease: the maternal uniparental disomy for chromosome 7 (5-10% of cases) and methylation abnormalities of chromosome 11p15 (in particular, ICR1 hypomethylation can be detected in up to 60% of SRS patients) [Wakeling EL et al, 2011]. However, in a significant portion of these patients the molecular cause is currently unknown. The clinical diagnosis is made when at least four of the six criteria of the Netchine-Harrison clinical score (NCHSS, 2015) [Azzi S et al, 2015] are present. If this is the case, both genetic and molecular tests should be conducted to confirm the result. SRS should be differentially diagnosed with prenatal growth restriction caused by placental dysfunction such as Wiedemann Rautenstrauch syndrome, 3M syndrome and Mulibrey dwarfism. Birth, height and weight of these patients are lower than normal, while the head circumference is typically preserved (pseudohydrocephalic appearance) [Perkins RM et al, 2002]. From oral-facial point of view, the primary features of disease include broad forehead, triangular face appearance with big eyes, small

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and pointed chin, wide mouth with thin lips and downturned corners. All these clinical signs tend to be less identifiable in adulthood. Regarding orthopedic-orthodontic pathologies, the following are the main ones described in literature: micrognathia with crowded teeth, II skeletal class, increased overjet and overbite values, convergent facial type, ogival and narrow palate and reduced facial height. Some dental anomalies such as early eruption, the presence of supernumerary elements or agenesis and some structural alterations like enamel hypoplasia are also reported [Bergman A et al, 2003]. Children with SRS are frequently treated with Growth Hormone (GH) by daily injection from the age of 2 until adolescence. The therapy seems to give positive results in terms of increased stature growth. Reaching a correct height before school starts can positively affect some psychosocial factors and promote interpersonal relationships [A. Dimitrijevic Carlsson et al, 2023].

## Case report

In September 2021, M., a 7-year-old female patient, came for her first orthodontic visit. The patient's general state of health was good, and the anamnesis was negative for any food or drug allergy. M. was not on GH therapy. She reported pain during chewing which resulted in insufficient nutrition. An appropriate nutrition is very important for these patients, who already present a poor postnatal growth. The functional analysis highlighted the presence of atypical swallowing and the interposition of the lower lip [R. Cond et al, 2012]. Parents also reported a rare snoring without apnoeic events [L. Agostini et al, 2023]. In M. some of the patient's primary facial features of disease like triangular face shape, convex profile, broad forehead, wide mouth with thin lips were observed (Fig. 1 and 2). The weight and height were lower than normal for her age. Intraoral examination showed a Class II molar relationship (right and left), increased overjet and overbite, gingival impingement at the upper retroincisive gum and upper incisors eso-inclination (Fig. 3-4). Even if there was a little contraction of the transverse diameters (39.1/38.8), that resulted in an important upper and lower anterior dental crowding (Fig. 5 and 6).



FIG. 1-2 Extraoral frontal and profile view.

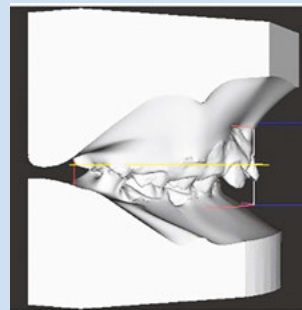


FIG. 3 Intraoral frontal view.

FIG. 4 Initial digital model (right profile view).

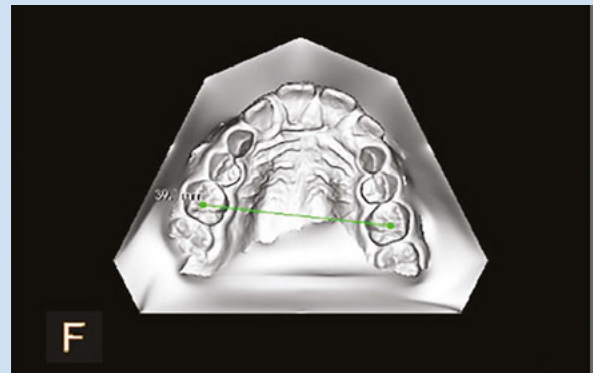
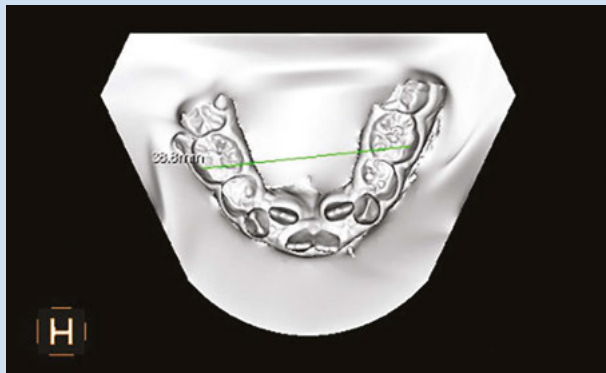


FIG. 5-6 Initial digital model (upper and lower view).



FIG. 7 Orthopantomogram x-ray.

FIG. 8 Lateral teleroadiography for cephalometric analysis.

**FIG. 9** Frontal occlusion after treatment.



The dental crowding was also observed by the orthopantomogram x-ray, which confirmed the presence of all the elements of the permanent series (Fig. 7). Cephalometric analysis diagnosed a skeletal class II in a brachycephalic patient with a convergent facial type. The values also identified a serious hypo-maxilla (Fig. 8).

### Discussion

The orthodontic therapy goals include palatal expansion, dental alignment, dental levelling and the creation of the correct arch shapes. Treatment is done with Leaf Self Expander (LSE) appliance (6 mm, 450 mg) on deciduous second molars for 12 months and partial-banded orthodontic treatment of the maxillary and mandibular arches for dental alignment (not less than 6 months). In order to limit the interposition of the lower lip, a myofunctional removable device is also included until the beginning of the second phase of permanent teeth development. The orthodontic treatment is complicated since the diagnostic moment. The small size of the oral cavity hinders the impression stage and the placement of the orthodontic appliance. After about 6 months the palatal expansion appliance is removed due to the presence of a lesion on the lateral surface of the tongue. M's compliance is very difficult. The bleeding and the pain frequently reported by the patient prevent the conclusion of the therapy. The reached palatal expansion (4 mm) is preserved by another device (Nance appliance) for 3 months. After this period all the fixed appliances are removed and replaced by superior and inferior customized Essix with a specific addition of resinous material for the mandibular advancement. No other type of myofunctional device would be accepted by the little patient. At the moment she has no feeding problems and she is not undergoing treatment. The refinement of occlusion will be evaluated at completed permanent dentition.

### Conclusion

The adopted orthodontic solution results in the resolution of the functional problem but also in the improvement of the poor facial esthetic. Gingival impingement is solved, giving the patient pain-free chewing (Fig. 9). Post-treatment records reveal an acceptable occlusion even if overjet and overbite levels remain above reference values. In order to promote a functionally and aesthetically harmonious craniofacial development, it is necessary to resolve the cranioskeletal discrepancy in developmental age (6 – 12 years). In this clinical case, the early approach involves the use of an orthopaedic appliance (that takes advantage of sutural immaturity to stimulate transverse bone growth) and a functional activator (to stimulate mandibular advancement) to be used preferably in the period immediately before the pubertal growth peak. Given the high prevalence of orthopedic-orthodontic

pathologies in these patients, early diagnosis is important to establish the most effective treatment plan [S. Saccomanno et al, 2022]. It should also be remembered that children with SRS are at greater risk of developing the carious disease due to frequent sugar intake (to counteract hypoglycemic episodes), enamel hypoplasia and the presence of gastroesophageal reflux. Therefore, a multidisciplinary approach, that require the collaboration of several figures such as the pediatric dentist, orthodontist and maxillofacial surgeon to the care of these patients, is necessary [F. Braiotta et al, 2023].

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