



# Rubinstein-Taybi syndrome, medical and dental care for special needs patients: Clinical case report

## *Síndrome de Rubinstein-Taybi, atención odontoestomatológica a pacientes especiales: reporte de caso clínico*

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### ABSTRACT

Medical and dental care for special needs patients oftentimes require effective and rapid clinical approach so as to improve patients' quality of life. Rubinstein-Taybi syndrome is a rare disorder where anomalies in genes CREBBP and EP300 are observed. This syndrome was first described in 1963 by two North American physicians. Among the main characteristics and manifestations of this syndrome we can count mental retardation (moderate or severe) microcephaly, thickened first finger or toe, growth retardation, characteristic face with hypoplastic lower jaw, downward-sloping arched palate, palpebral fissure, irregularly implanted teeth, crowded teeth, strabismus, low-set, malformed ears, beak-shaped nose, bushy eyebrows and long eyelids. The concept of patient adaptation to the dental office entails achieving and preserving his collaboration during treatment by means of a show-and-tell process. Behavior was modeled with gradual exposition, positive reinforcement and active physical restraint with the mother's help. **Results:** The patient's oral health was satisfactorily reestablished.

**Key words:** Dental clinical approach, Rubinstein-Taybi syndrome.

**Palabras clave:** Abordaje clínico odontológico, síndrome de Rubinstein-Taybi.

### RESUMEN

La atención odontoestomatológica a pacientes especiales, requiere en muchos casos un abordaje clínico rápido y efectivo con el propósito de mejorar su calidad de vida. El síndrome de Rubinstein-Taybi, es un trastorno poco común, donde se observan anomalías en los genes CREBBP y EP300, descrito en 1963 por dos médicos estadounidenses, entre las manifestaciones y características principales tenemos: retraso mental (moderado o severo), microcefalia, primer dedo de la mano o del pie engrosado, retraso del crecimiento, facies característica con maxilar hipoplásico, fisura palpebral inclinada de forma descendente, paladar ojival, dientes de implantación irregular apiñados, estrabismo, orejas mal formadas de implantación baja, nariz en forma de pico, cejas espesas, párpados largos. El concepto de adaptación del paciente a la consulta odontológica representa lograr y mantener su colaboración durante el tratamiento mediante un proceso de enseñanza-aprendizaje. Se realizó el modelamiento de conducta, exposición gradual, refuerzo positivo, restricción física activa con apoyo de la madre. **Resultados:** Se logró restablecer la salud bucal del paciente en forma satisfactoria.

### INTRODUCTION

Medical and dental care for special-needs patients requires, in many cases, rapid and effective clinical approach with the purpose of improving patients' quality of life. In dentistry, when treating special needs patients, support afforded by medical inter-consultation and specialized knowledge in clinical approach will be required so as to achieve satisfactory treatment.

### TAYBI-RUBINSTEIN SYNDROME

Taybi-Rubinstein syndrome is a rare disorder where anomalies of genes CREBBP and EP300<sup>1</sup> are observed. It was first described by two USA doctors, and subsequently named after them: Jack Rubinstein

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a pediatric geneticist and Hooshang Taybi, a pediatric radiologist. This syndrome is a multi-systemic genetic disease with variable clinical expression.<sup>2</sup> Among its main characteristics and manifestations we can count the following:<sup>3</sup> mental retardation (moderate or severe), microcephaly, thickened first finger or toe ( with or without angulation) and growth retardation. Among other characteristics we can find the following: capillary malformations, hypertrichosis , cardiac alterations, genital-urinary anomalies, hypospadias, cryptorchidism, renal malformations. In the face, the following characteristics can be found: hypoplastic maxillary downward sloping palpebral fissure, arched palate, crowded and irregularly implanted teeth, strabismus, low implanted deformed ears, beak shaped nose, thick eyebrows long eyelids (*Figures 1 and 2*).

### CLINICAL DENTAL APPROACH IN PATIENTS WITH DISABILITIES

The concept of patient adaptation to the dental office entails achieving and preserving his collaboration to treatment through a process of show- and- tell through which values, attitudes and knowledge are acquired and which will promote a positive attitude towards dentistry.<sup>4</sup>

There are different techniques and procedures to approach patients with disabilities and special needs, they are not different from those used in pediatric dentistry. Among these techniques we can count the following:

**Distraction: voice distraction technique.** Strategy in this technique targets drawing the patients' attention and stimulating their imagination so as to distract them

from the ongoing dental procedure.<sup>5</sup> **Voice control technique.** With a friendly look, peaceful and soft voice so as to draw the patient's attention, the dentist can increase volume in the voice so as to curb negative behavior in the patient and assess whether communication is being successful. It is necessary to use this technique wisely so as not mistake voice increase with shouts.<sup>5</sup> **Non-verbal communication technique.** Includes a variety of non verbal gestures Among these we can count friendly gestures and facial expressions, soothing caresses, soft physical and visual contact.<sup>5</sup> **Modeling technique.** This technique is used to decrease negative and anxious behavior by stimulating the patient to learn suitable response or behavior to a given dental situation. This can be effected through observation of another patient with suitable behavior during treatment.<sup>5</sup>

**Latent inhibition technique,** this technique is important in patient conditioning since it purports the aim of organizing dental treatment, which will begin with very simple procedures. Implementation of this type of «latent inhibition» is important in order to prevent dental fear; it must be incorporated in the global treatment plan.<sup>5</sup> **Gradual exposition technique.** This technique implies helping the patient to slowly grow accustomed to the treatment, let him gradually become familiar with the dental chair and working equipment in small steps. These small steps or de-sensitization must vary from minimal to maximum exposure, and can be used in all treatment phases: from first exposure to dental situation up to dental procedures such as use of dental burr or anesthesia.<sup>5</sup> **Positive reinforcement technique.** This type of operating conditioning can be included within the frame of reinforcement models, and occurs



**Figures 1 and 2.**

Facial appearance of the patient. Beak shaped nose, strabismus, low-implanted ears.

when reinforcement is a stimulus that increases the probability of emitting the conduct that causes the apparition of the reinforcement, which can be tangible and untangible.<sup>6</sup>

### CLINICAL CASE

A 28 year old female patient with diagnosis of Rubinstein-Taybi syndrome (confirmed genetic clinical diagnosis) attended care facilities of her jurisdiction's Health Center afflicted with dental pain.

#### Medical history

The patient was the outcome of a full-term pregnancy; birth was delayed, patient suffered asphyxia and aspiration of amniotic fluid which caused generalized sepsis and permanence in hospital for 45 days. Following medical advice, the child was subjected to a genetic test which revealed presence of Rubinstein-Taybi syndrome, moreover, the patient was afflicted with infantile encephalopathy. The patient had suffered child diseases such as chicken pox, measles, rubella mumps colds, bronchial problems and skin sensitivity. The patient was afflicted with sporadic convulsions, from her first year of life to the present date. Presently she was only afflicted with seasonal bronchial problems caused by her mouth breathing. The patient exhibited characteristic facial traits, with beak shaped nose, strabismus, low-implanted ears (*Figures 1, 2 and 3*).

#### Dental history

Patient exhibited permanent dentition, with no prior dental intervention; the mother informed she was not



**Figure 3.** The patient's hand exhibit fingers with thickened nodules (joints) which presented self-inflicted lesions.

cooperative and exhibited an instinctual tendency to shut her mouth and bite.

#### Dental clinical diagnosis

Generalized gingivitis, localized periodontitis, dental caries, dental crowding, anterior open bite, posterior cross-bite and impacted tooth (*Figures 4 and 5*).

#### Treatment plan

**Clinical approach and conditioning in a dental environment:** empathy with patient and relatives, establishment of suitable communication with patient and relatives, conduct modeling. Moreover, operatory actions such as securing the patient's head with the operator's arm and forearm to preserve patient's stability require the parent's help in order to ensure patient's trust and feeling of protectiveness about having her mother nearby (Active Restrictive Fixation). Affective positive reinforcement was equally used, and effected with the behavior exhibited on the day of visit (*Figures 6 and 7*).



**Figures 4 and 5.** Opened and closed mouth, dental crowding, Cross-bite.



**Figures 6 and 7.**

Approach technique with active restrictive fixation (family assistance).

**Periodontics.** Physiotherapy, prophylaxis with ultrasound, polishing with bicarbonate and fluoridation with 1.23% fluoride. Control of brushing and use of chlorhexidine to reinforce dental brushing (Figure 8).

**Operative dentistry.** Restoration with liquid resin in teeth 14, 15, 24, 25, 34, 35. Resin restoration in teeth number 12, 25, 37, 46, 47, 48 using self-curing ionomer base (Figures 9 to 12).

Periodic control of dental hygiene and radiographic control of impacted tooth number 38.

**Prognosis.** Favorable prognosis supported by the family's commitment to preserve oral hygiene as well as a periodic controls at the dental office.

## DISCUSSION

Clinical approach for special needs patients in most cases requires specialized training, with knowledge of clinical approach techniques and suitable handling of dental procedure so as to achieve satisfactory results in treatment.

In the present case a patient with Rubinstein-Taybi syndrome was treated. Mixed operative procedures and techniques were used in order to achieve successful dental clinical approach. For the patient's benefit, communication bonds (links) were achieved with the patient, and mainly with the family, who is the entity responsible for this patient's care. Approach mechanisms were represented by active fixation with the family's support, above all to achieve patient cooperation; the patient was very receptive and all that was needed was to make her feel safe in the dental chair. The patient's head was secured (immobilized) with operator's arm and forearm so as to confer free movement to the patient.<sup>4,5</sup>

Dental and medical reports found concerning these patients are scarce, both nationally and internationally; moreover access to these patients is difficult. Tiso<sup>7</sup>



**Figure 8.** Prophylactic treatment, approach technique with restrictive fixation (family assistance) pneumatic ultrasound equipment.

showed that these patients can be treated in an out-patient mode; he informed that gingivitis and periodontitis were related to difficulties encountered when using dental brush. When comparing similar results to those of the present case, it can be said that possibility of gingivitis and bronchial problems will increase since these patients are mouth breathers.

Likewise, Münevveroglu et al<sup>8</sup> informed of intra-oral results in a patient with RTS: he observed mouth opening decrease, narrow palate, malocclusion, gingivitis and caries, all the aforementioned characteristics were similar to those found in the present case. Our results concurred with those reported by Morales<sup>9</sup> who referred that his patient exhibited short physical stature and a lower than normal body mass index. With respect to oral health circumstances, we found similarities in lack of midline alignment among upper and lower incisors, as well as



Figures 9 at 12.

Operative dentistry procedure; restorations with light-curing resin, active restrictive fixation and arm supported over the patient's head in order to restrict movements. Active familial participation.

crowding with high indexes of dental caries. Similarly to the present report, Davidovich et al<sup>10</sup> reported posterior cross-bite as well as similarities in dental caries and gingivitis.

The patient and her family were prepared for the oncoming medical and dental care, which included active restriction approach techniques, with the patient's mother assistance. This differs from techniques of Münevveroglu et al,<sup>8</sup> Davidovich et al,<sup>10</sup> Stalin et al<sup>11</sup> who worked under general anesthesia. Likewise, Freitas et al<sup>12</sup> worked on an ambulatory basis with the patient due to the fact that he was very collaborative.

It is of the utmost importance to bear in mind the fact that patients' characteristics might vary with respect to their collaboration. Their state of mind bears influence, as well as the fact that level of mental retardation will determine effective care. Nevertheless, all actions geared to achieve oral health improvement in these patients are the ultimate objective, so as to establish healthy quality of life.

### CONCLUSIONS

Stomatological handling of special needs patients requires joint use of different approach techniques; in order to achieve success in these patients' oral recovery, we are responsible for improving their quality of life.

Likewise, family participation in the dental environment contributes to patient's stability and

better behavior. Familial involvement also contributes to better participation and care at home.

Procedures to be developed must be performed in the shortest possible time so as to avoid patient's fatigue and annoyance, thus, later care will be administered with equal satisfaction for patient and dentist.

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