Scientific Article

Oral manifestations in Rett syndrome: a study of 17 cases

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Abstract

Seventeen patients with a mean age of 7.33 (range 2.7–12.7) years with Rett syndrome (a progressive neurological disorder that occurs mainly in females) were evaluated for oral manifestations and habits. The most frequent habits were digit/hand sucking and/or biting (17/17), bruxism (14/17), mouth breathing (7/17), drooling (5/17), and tongue thrusting (5/17). Gingivitis (13/17) was the most common alteration of soft tissues. Only 2.7% of tooth surfaces were decayed. Nonphysiological dental attrition was present in 71% (12/17) of the children. Palatal shelving could be observed in 53% (9/17) of the children, probably related to the digit/hand sucking and/or biting habits. A high prevalence of anterior open bite (9/17) was observed. No patients exhibited anomalies of tooth number, size, form, structure, or eruption. (Pediatr Dent 19:349–52, 1997)

Rett syndrome (RS) is a progressive neurological disorder estimated to affect 1:10,000–1:15,000 live female births.[1] It remained almost unknown until 1983, when Hagberg et al.[2] published a pooled French–Portuguese–Swedish series of 35 patients, increasing the interest and awareness of this condition worldwide.

RS presents clear signs and symptoms. The pregnancy of mother, delivery, physical growth parameters at birth, neonatal period, and the first few months of life are fairly normal and uneventful for patients with RS.[1,3,4] Usually from 6 to 18 months of the child's life, changes like developmental stagnation, altered communicative ability, loss of active play interest and partial or complete loss of acquired purposeful manual skills occur quickly. The progressive evolution of changes leads to a social withdrawal. The child assumes autistic behavior. The stereotypic hand movements such as hand wringing/squeezing, clapping/tapping, mouthing, and "washing"/rubbing automatisms appear after purposeful hand skills are lost. Gait apraxia and truncal ataxia appear between ages 1 and 4 years.[1,3–5]

Other common symptoms are: periodic apnea during wakefulness, intermittent hyperventilation, breath-holding spells, forced expulsion of air or saliva, electroencephalogram abnormalities, seizures, peripheral vasomotor disturbances, constipation, abdominal bloating, night laughing, hypotrophic small feet and hands, weight loss, growth retardation, and apparent insensitivity to pain.[1,3–7] In 1989, Rice[8] noted that the Rett patient is extremely thin in spite of having a good appetite, and that difficulty in gaining weight is a threat to the child's life.

Oral findings

Although mentioned in some previous reports, oral findings in Rett syndrome have received scant attention in the literature. Bruxism is mentioned frequently. The RS bruxism is an episodic creaking sound similar to that of a slowly uncorked wine bottle. It appears to be produced far back in the jaw. It is a helpful supporting sign, even if it is not absolutely pathognomonic.[1,3,9] Coleman et al.[6] reported in 1988 that bruxism was present in 95% of 63 children with RS studied, according to their parents. They also noted protrusion of the tongue in 65%, accompanied by hypersalivation in 84%.

Buccino and Weddell[10] (1989) were the first to comment on the stomatological aspects in Rett syndrome in the dental literature. They included bruxism, drooling, biting the hands, digit/hand sucking, and tongue protrusion as habits and hypersalivation, micrognathia, abnormal chewing, narrow maxillary arch, and high arched palate as oral signs. They reported a case of a 4-year, 3-month-old white female with RS with all 20 primary teeth caries-free and sound enamel, and who was asymptomatic. Generalized attrition of the primary teeth, especially the maxillary and mandibular anterior teeth, was present and associated with the neurological and psychological disturbances that led to bruxism.

A 5-year-old girl with Rett syndrome was reported by Peak et al.[9] in 1992. The extraoral examination revealed hypertelorism and obvious bilateral masseteric hypertrophy with constant bruxism producing a grating sound. The intraoral examination showed a complete primary dentition with no caries. The incisors and canines presented gross attrition, but the molars were

normal except for wear consistent with bruxism.

To date, the dental literature on RS has been confined to case reports.[9,10] A comprehensive view of oral health of children with RS has not been determined.

These children are cared for by dental professionals, so it is important to report the most frequent oral manifestations and habits. This report describes the oral manifestations and associated habits in 17 RS children examined between 1993 and 1994 at the School of Dentistry of São Paulo University in Brazil.

Methods and materials

Seventeen girls with RS, previously diagnosed by a pediatric neurologist service, associated with the ABRE-TE—São Paulo (The Brazilian Rett Syndrome Association of São Paulo), were referred to the School of Dentistry of the São Paulo University for dental treatment and examined clinically by two dentists with parental consent. The examiners were trained by a calibrated examiner. In addition, the standard diagnostic criteria were reviewed.

Using a specially written clinical evaluation form, personal data (including pregnancy, delivery, and medical history, diet history, dental history, and use of fluoride) and data of the general and clinical examinations were recorded. In order to avoid variability among examiners, one performed the examination of the hard tissues and another the examination of the soft tissues and occlusion.

Caries data were obtained under artificial light after professional mechanical tooth cleaning by using mouth mirror and probe. Modified WHO Criteria[11] (1990) were used for recording dental caries.

The criteria for diagnosing bruxism included either a clinical exam or a parental report or both. The clinical indicators of diagnosing this parafunction were the presence of dental wear/attrition and bruxofacets.[12]

The plaque index of O'Leary et al.[13] (1972) was used to assess the oral hygiene satus. After disclosing with "fucsin solution", the percentage of surfaces with plaque was determined.

The evaluation of the occlusion included the anteroposterior relationship, the vertical relationship, and the lateral relationship. In the anteroposterior relationship, primary molar and canine relationships or permanent molar and canine relationships were determined during the intraoral examination. The vertical relationship was examined for open bite or deep bite. The lateral relationship of the arches was examined for midline discrepancies and posterior crossbites.[14] In the primary dentition, the anterior segment was examined for spacing between teeth, to be classified as Baume type I arch or Baume type II arch.[15]

The palatal depth was observed during the examination and compared subjectively without using Korkhaus's methods.

Results

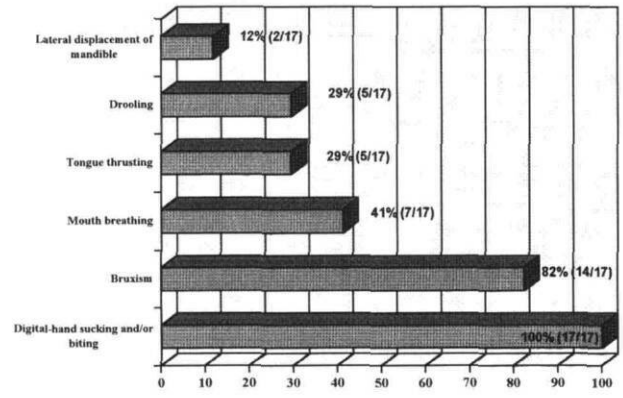

Fig 1. Oral habits presented in 17 children with Rett syndrome.

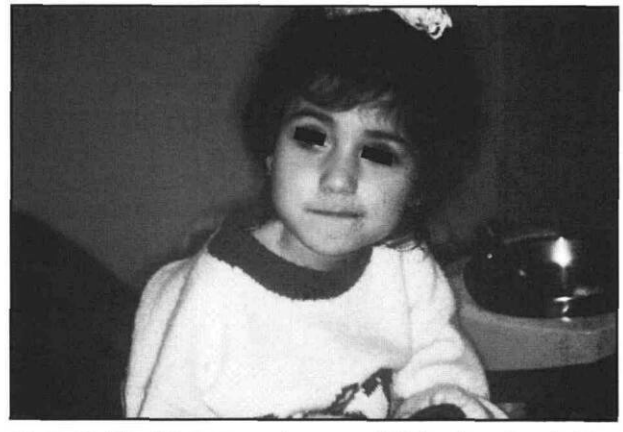

Fig 2. Child with Rett syndrome exhibiting bruxism habit

The 17 girls with confirmed Rett syndrome diagnosis were aged from 2.7 to 12.7 years old (mean age 7.33 years). At the time of the examination 6 of the 17 RS patients were in the primary dentition, 10 of 17 in the mixed dentition, and one in the permanent dentition.

Oral habits

The percentage of the oral habits exhibited by the patients as reported by their parents and/or observed in the examination is shown in Fig 1.

All of the children (17 of 17) exhibited hand mouthing habits, sucking, and/or biting fingers and hands. A high percentage of children (14 of 17) exhibited bruxism (Fig 2). Mouth breathing habit was observed in 41% (7 of 17) of the examined children. Tongue thrusting habit (5 of 17) and lateral displacement of the mandible (2 of 17) were noted during the examination of the children since it would be difficult for parents to notice these habits. Only 5 of 17 RS children showed a drooling habit.

Oral manifestations

The oral manifestations of the hard and soft tissues and the occlusion of the 17 RS children at the time of the examination are shown in the Table.

A large bacterial plaque accumulation (plaque index

= 100%) and a high frequency of gingivitis were observed (Fig 3).

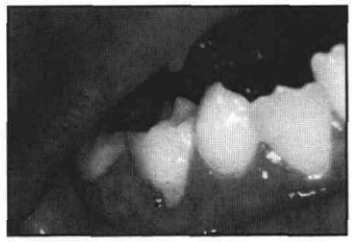

Fig 3. Gingivitis aspect and high plaque accumulation (➤) observed in Rett syndrome patients examined.

An occasional finding in the soft tissues was benign migratory glossitis, observed in only one child, while two children exhibited an ischemic area in the palatal mucosa. Nonphysiological dental attrition was noticed in 71% (12 of 17) of the children involving most often the anterior primary teeth. Four of 17 children showed trauma in the anterior teeth.

Only 2.7% of all dental surfaces examined were decayed despite the ineffective or nonexistent oral hygiene habits and the softened food within the characteristic standards of the RS child reported by Rice[8] (1989).

A relevant finding is that 7 of the 17 children were caries-free (Fig 4) despite the high caries prevalence in children in Brazil.

Palatal shelving probably associated to sucking habits and mouth breathing was observed in 53% (9 of 17) of the children examined.

The anteroposterior relationship, the vertical relationship, and the lateral relationship between the maxillary and mandibular arches were evaluated during the occlusion examination. In seven children who were in the primary dentition, the relationship of the maxillary and mandibular second molars was evaluated. A flush terminal plane was present in 57% (4 of 7) of the children and a mesial step was present in the other 3. In the anterior segment, 57% (4 of 7) had a Baume type I arch, 29% (2 of 7) had a Baume type II arch, and 14% (1 of 7) had a mixed arch. In only one child open bite was observed while another child presented a deep bite.

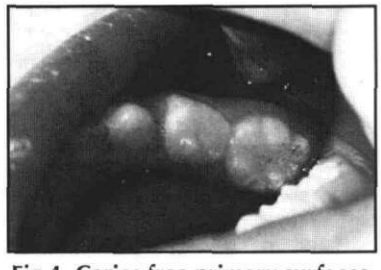

Fig 4. Caries free primary surfaces in a child with Rett syndrome.

In 10 children it was possible to establish the first permanent molar relationship. Ninety percent (9 of 10) had a class I molar relationship and 10% a class II according to Angle's classification. Eighty percent of the children (8 of 10) had open bite probably related to sucking/biting habits, mouth breathing, and tongue thrusting. None of the 17 children had abnormal lateral relationship. A reduction of the vertical dimension was observed in 23% (4 of 17) of the children.

None of the children had any anomaly of tooth size, number, form, structure, and eruption.

TABLE. PERCENTAGE OF ORAL MANIFESTATIONS OF THE HARD AND SOFT TISSUES OBSERVED IN 17 CHILDREN WITH RETT SYNDROME EXAMINED

	Oral manifestations	*Percentage*	
Soft tissues	**Gingivitis**	**76%**	**(13/17)**
	"Benign migratory glossitis"	**6%**	**(1/17)**
Hard tissues	Slight dental attrition	47.5%	(8/17)
	Severe dental attrition	23.5%	(4/17)
	Decayed dental surfaces*	2.7%	
	Palatal shelving	53%	(9/17)

* 97.3% caries free surfaces

Discussion

This study examined the oral findings of a sample of children with a confirmed diagnosis of Rett syndrome. The most common oral findings included digit/hand sucking and/or biting, bruxism, mouth breathing, drooling, and tongue thrusting as habits and a high gingivitis prevalence, a low caries prevalence, nonphysiological dental attrition, palatal shelving, and a high prevalence of anterior open bite as oral signs.

Rett syndrome children present frequent habits of mouthing hands and sucking and/or biting hands and fingers, which are stereotypic hand movements and pathognomonic signs of the condition. These habits were observed in all children examined and could have caused changes in the occlusion.

In agreement with Coleman et al.[6] (1988), Hagberg[1] (1989), Peak et al.[9] (1992), and Pereira[3] (1993), who stated that bruxism is a helpful supporting sign in identifying the syndrome because of its high frequency, it was observed in most of the 17 children.

It is well known that several etiologic factors are responsible for mouth breathing such as swollen tonsils and adenoids, nasopharyngeal obstructions and deformities, and upper airway infections. It seemed that upper airway infections were the principal factor for this habit among the examined children.

Buccino and Weddell[10] (1989) included drooling as one of the most often reported habits in the specialized medical literature. However only 5 of 17 RS children showed it, which could be associated more with their difficulty in swallowing saliva then with hypersalivation itself.

The high frequency of gingivitis observed was probably related to the large bacterial plaque accumulation, which was examined with the use of disclosing agents. This large bacterial plaque accumulation was due to poor oral hygiene habits, because children with Rett syndrome are incapable of taking care of their own teeth as they no longer have purposeful hand skills. In addition, their parents'

instructions and attitudes toward an efficient oral hygiene practice were not effective.

As Buccino and Weddell[10] (1989) and Peak et al.[9] (1992) also reported that their samples were caries-free, more studies in this area should be conducted.

Dental attrition was probably due to the excessive habit of bruxism. This finding is in agreement with Peak et al.[9]

Rett syndrome children with gait apraxia are more susceptible to falls and therefore to injuries in the anterior teeth.

Except for the digit/hand sucking and/or biting habits exhibited by all patients with this condition, no single oral finding noticed in this survey was pathognomonic of RS. However, patterns of some of these findings may be characteristic of the syndrome, such as bruxism, dental attrition, open bite, palatal shelving, and gingivitis. Routine oral examination and care are a necessary part of the overall medical supervision for patients with RS. It is hoped that this paper will add information to this rare syndrome as well as help physicians and dentists who care for RS patients. In addition, it is hoped that other studies on this condition will be conducted in order to determine the real state of oral health in RS population.

Conclusions

1. The most frequent habits observed were digit/hand sucking and/or biting (17 of 17), bruxism (14 of 17), mouth breathing (7 of 17), drooling (5 of 17), and tongue thrusting (5 of 17).
2. Gingivitis was the most common alteration of soft tissues.
3. A low prevalence of decayed tooth surfaces (2.7%) was present.
4. More than half (12 of 17) of the children presented nonphysiological dental attrition.
5. Palatal shelving (high palate) was observed in 53% (9 of 17) of the children.
6. A high prevalence of open bite (9 of 17) was observed.
7. None of the children had any anomaly of tooth number, size, form, structure, and eruption.

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