the Child Dental Patient with

Rett Syndrome

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European Reference Network
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Definition

- Rett syndrome (RTT) is an **early-onset neurodevelopmental disorder** that primarily affects females, resulting in severe cognitive and physical disabilities, including :
  - loss of speech
  - loss of hand movements/manual dexterity
  - characteristic hand movements such as hang wringing, repetitive mouth and tongue movements
  - intellectual disability/learning problems.
- It is one of the most prevalent causes of intellectual disability in females.

Genetics and epidemiology

- Rett syndrome (RS) is a neurodevelopmental disorder caused by mutations in the Methyl CpG binding protein 2 (*MeCP2*) gene, a gene located on the long arm of the X chromosome .
- *MECP2* encodes the transcriptional repressor methyl-CpG-binding protein 2, a protein involved in synaptic development and function.
- This X chromosome-linked condition **affects females almost exclusively,** with an incidence of 1:10,000–20,000 live births, with rare cases reported in males.

Rett syndrome progression

- Hagberg described **four distinct stages**:
  - *Stage I* is defined by an early onset of developmental stagnation and begins between 6 and 18 months of life when motor regression and autistic behaviors are seen.
  - In *stage II*, between ages 1 to 4 years, infants show regression of psychomotor development. Affected patients start losing the already acquired skills in communication and behavior and show symptoms of mental retardation.
  - *Stage III*, which occurs between ages 4 to 10 years, is marked by the recovery of contact and communication (“pseudo-stationary period »); however motor dysfunction is more prominent.
  - *Stage IV* usually starts after 10 years of age and is characterized by late motor deterioration; patients lose their ability to walk and would become non-ambulatory or completely dependent on a wheelchair for mobility. This stage is characterized by weight loss and skeletal deformities.

Common medical and behavioural aspects

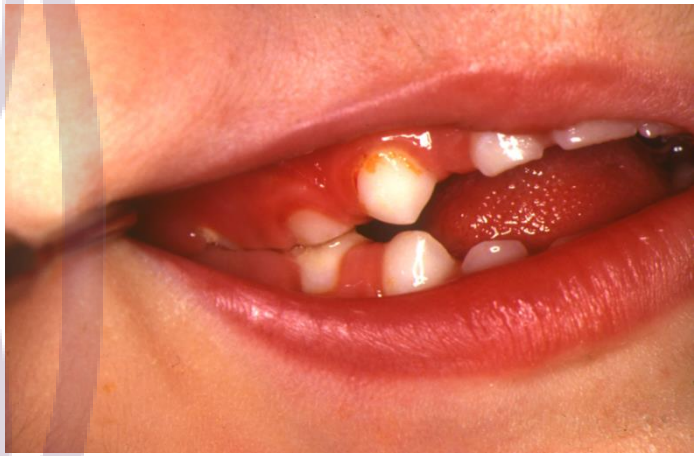
- Hand stereotypies.
- Altered breathing patterns like hyperventilation and breath holding.
- Disturbed sleeping patterns.
- Spasticity.
- Mood fluctuations and signs of fear/anxiety, inconsolable crying and screaming at night.
- Feeding and gastrointestinal problems.
- Epilepsy.
- Scoliosis.
- Small feet and hands.
- Failure to thrive.
- Development of osteoporosis.

Medical management

- To date there is no cure reported for Rett Syndrome.
- However a palliative treatment strategy can alleviate functional, sensory and motor skills. This has led to a survival rate of at least six decades.

Oral characteristics

- Limited reports describe the dental characteristics of RS patients.
- The most common oral finding is **bruxism**:
  - usually appearing at Stage III of RS
  - diurnal bruxism more prevalent than the nocturnal one
  - intermittent and aggressive
  - grinding of the teeth could be accompanied by an increase in stereotyped hand movements
  - causes discomfort for the patient: dental attrition, excessive tooth wear, muscle dysfunction
  - causes anxiety for the caretakers.



Oral characteristics

- Other oral manifestations reported:
  - masseteric hypertrophy
  - erosive tooth wear (maybe associated to gastroesophageal reflux)
  - gingivitis, poor periodontal status
  - dental caries
  - oral soft tissues trauma
  - high arch palate, anterior open bite : this could be due to mouth breathing, tongue thrusting, digit/thumb sucking and mouthing. These parafunctional habits trigger or worsen the anterior open bite as well as palatal shelving
  - also oral findings related to the pharmacological treatment (anxiety, epilepsy…), which include xerostomia, glossitis, erythema multiforme, gingival hyperplasia, dysphagia…

Dental management

1. Behavioral and pharmacological management of RS children

- Dental care in patients with Rett syndrome is often difficult due to communication problems (verbal vocabulary restricted), ambulatory difficulties and patient anxiety during the oral examination.
- For effective intervention, it is recommended that family and clinicians engage closely to determine the communication abilities of children with Rett syndrome.
- Scoliosis is a significant problem in RS patients. This condition may affect the position of the patient in the dental chair.
- It is better to plan brief appointments.
- Use a pain scale.
- Use of a mouth prop is helpful (little or no muscle control).
- Dental management should be carried out under sedation and even under general anesthesia in case of difficulties of delivering dental care under local anesthesia.
- The anesthesiologist should be informed of the possible difficulties of the airway management due to breathing abnormalities and the presence of muscular tonicity.

Dental management

2. Management of bruxism

It must be treated to prevent further dental attrition, discomfort and wear of the occlusion:

- bite planes
- soft splints, margins of the splint extended till the hard palate
- some authors contraindicated the use of splints due to the incontrollable motor function in some patients.
- acupuncture was used in combination with splints and was reported to be effective against temporomandibular problems and facial pain.

Dental management

3. Preventive measures

- The caregiver can have trouble preserving an adequate standard of oral hygiene:
  - difficulties of accessing the child's oral cavity
  - behavioral difficulties.
- Fluoride applications should be used to reduce the likelihood of caries.
- Frequent visits should also be encouraged to dentists for follow up.
- Dentists treating Rett syndrome patients should consider oral health education programs on special care dentistry.

Conclusions

- Rett Syndrome is a **progressive** pediatric **neurodevelopmental** disorder, predominantly affecting **females.** An initial stage of early normal development is followed by regression phase appearing as **severe intellectual disability**.
- Dental practitioners should know the **autistic** behavior and the characteristic **oral manifestations**, **digital-oral habits** and **oro-facial motor functional issues** associated with Rett Syndrome
- More research is needed to understand better the oral implications of Rett syndrome and might help in delivering a more comprehensive dental management.

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