the Child Dental Patient with

Down Syndrome

Down Syndrome – generalities

- The most common chromosomal disorder and cause of intellectual disability (Mubayrik 2016);
- Accounts for~8% of all congenital anomalies
- Congenital autosomal disorder characterized by generalized growth and mental deficiency.
- Approximately 95% of DS cases have the extra chromosome 21; the other 5% of chromosomal abnormalities include translocation (3%) and mosaicism or partial trisomy (2%).

Incidence / Prevalence

Overall **incidence**: 1 in 600-800 live births, increasing trend

Prevalence is increasing due to the extension of life expectancy of people with DS

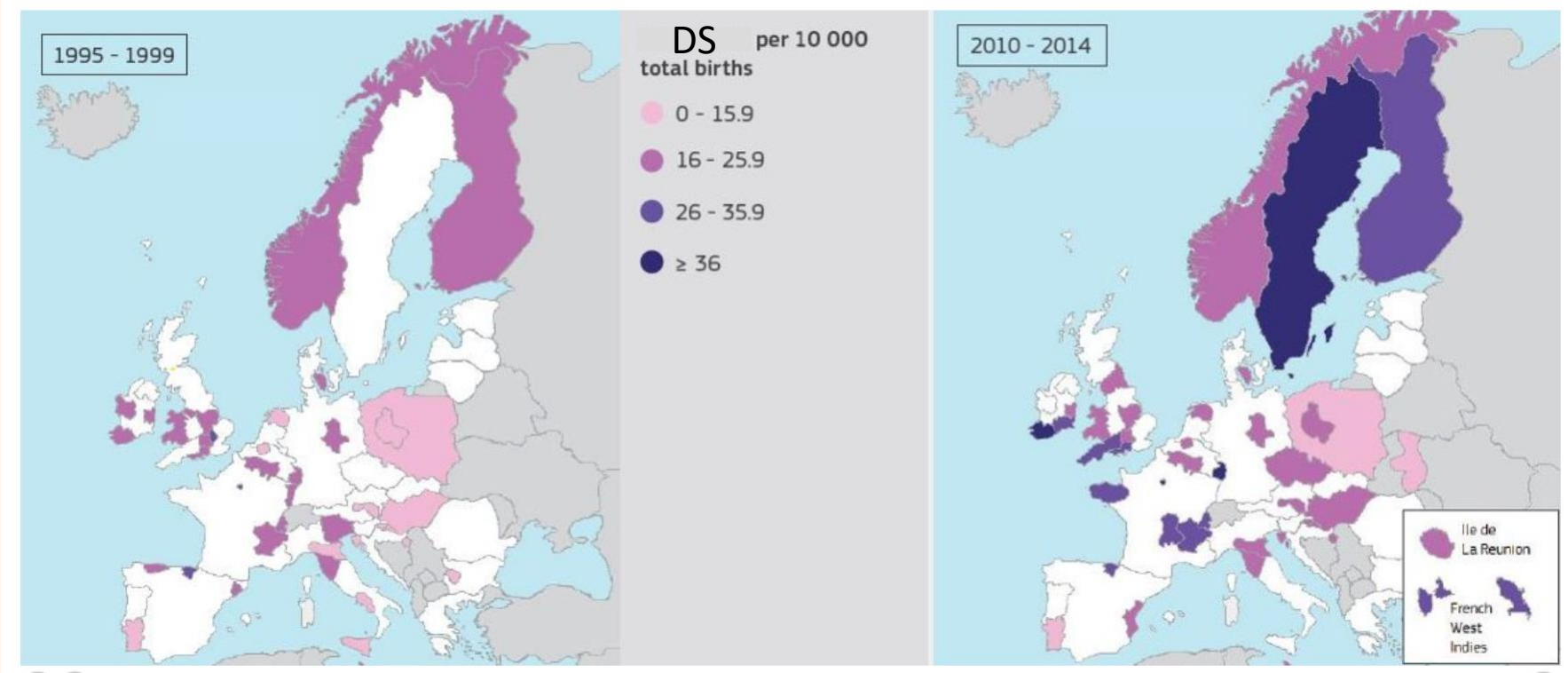
Differences between countries/regions:

- Romania (2015) – 4420 persons with DS(in a population of 19.82 million people): 2611 children and 1809 adults
  - most of Romanian SD persons are from Iași county (www.downinfoplus.ro)
- France (2021*) - 70000 persons with DS (in a population of 65.47 million people);
  - 0.79/1000 (1980-87)→ 1.81/1000 (1988-1992)(Stoll et al, 1994)
  https://pubmed.ncbi.nlm.nih.gov/7710252/
- Italy (2010**) – 38.000 persons with DS (1 / 700 live births)
- Turkey (2012***) – 1/1000 live births
- Dubai, UAE (1999-2003****): overall 1/449 live births (2.2 per 1,000);
  - 1/319 live births (3.13 per 1,000) among UAE nationals
  - 1/602 live births (1.66 per 1,000) among non-UAE nationals.

* https://www.academie-medecine.fr/down-syndrome-world-day/?lang=en#:~:text=The%20incidence%20of%20Down's%20syndrome,disease%20(ORPHA%3A%20870)
** https://www.reuters.com/article/us-italy-downs-idUSTRE61M33920100223
*** DOI: 10.4238/2012.September.10.1
**** https://doi.org/10.1159/000096136

Many couples tend to postpone parenting until later in life → increase of incidence of DS

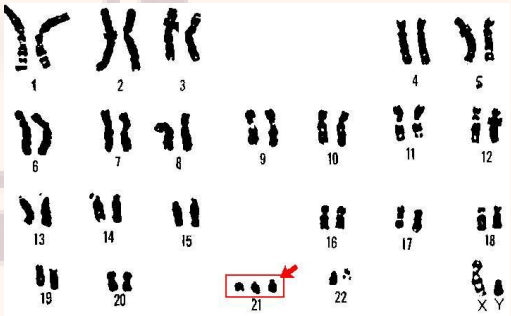
Down syndrome in EU(*):
1995-99: average 1 in 625 births; 2010-14 average 1 in 435 births



(*) Source: https://ec.europa.eu/jrc/en/news/down-syndrome-europe-has-disorder-epidemiology-changed-over-last-quarter-century

Etiology of trisomy 21

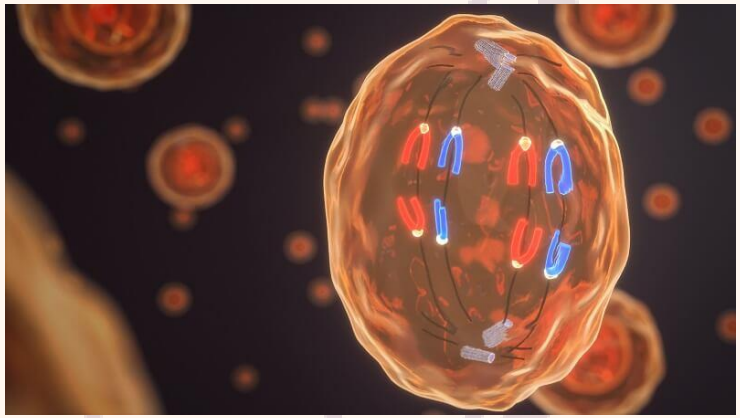
- Genetic epidemiology : most prevalent chromosomal disorder (prevalence of 1/700)
- Down syndrome corresponds to a genetic disorder when abnormal cell division during gametogenesis results in **extra full or partial copy of chromosome 21 : chromosomal nondisjunction**
- → Supplementary genetic material responsible for the developmental anomalies and clinical features of Down syndrome



Risk factors for trisomy 21

- Increased risk of chromosomal nondisjunction linked to **increase of maternal age** :

  - Risk of 1/500 at age 20 y
  - 1/400 at age 35 y
  - 1/100 at age 40 y
  - 1/28 at age 50 y



Other risk factors :

- Presence of a chromosomal translocation in the mother or father that can be transmitted to the child
- Already having a child with Down syndrome

Genetics of trisomy 21

Different types of chromosomal changes causing Down syndrome

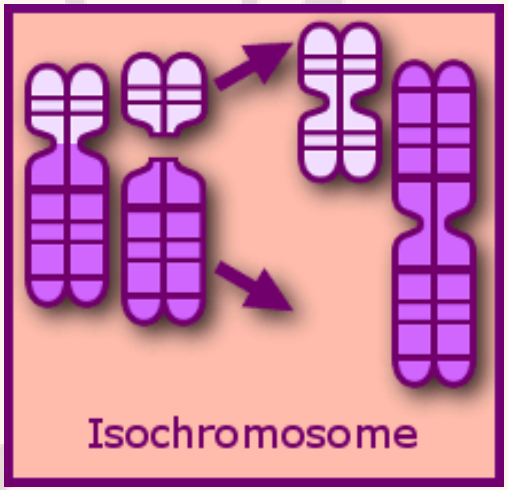
- **Complete trisomy 21 :** majority of cases (95%) with the chromosome 21 extra-copy coming from the mother (maternal non disjunction during meiosis).
- **Mosaic trisomy :** corresponds to less than 5% of the cases. Mosaicism with most of the cells having the extra-copy but some cells with a normal number of chromosomes. Results from an error in cell division early during embryonic development but gametes without chromosomal anomalies.
- **Translocation trisomy 21:** only part of the extra-copy of chromosome 21 is in the cells, the extra-part is translocated to another chromosome (translocation 21-14 or 21-22) (about 3% of cases).

Patients with a mosaic trisomy or translocation trisomy can have the same clinical features and phenotype than patients with complete trisomy 21 → **clinical variability in all types of trisomy**

Genetics of trisomy 21

- **Partial trisomy 21**: rare form characterized by the presence of a partial extra-copy of chromosome 21. Patient presents a moderate phenotype.

- **Trisomy 21 with isochromosome:** isochromosome formed of 2 short arms or 2 long arms with loss of the other arm**.** This anomaly can result in a true false negative chorionic villi sampling with foeto-placental discordance.



Genetics of trisomy 21

- **Reciprocal chromosomal translocations**

Possible reciprocal translocation between each autosome and gonosome and chromosome 21.

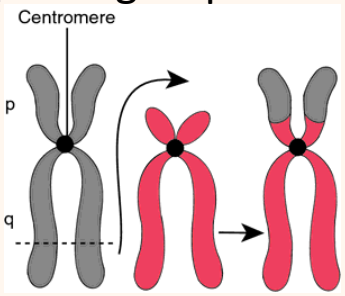
Corresponds to **3-4 % of cases**

The translocation can be inherited from parents having a rearranged genetic material.

- **Robertsonian translocation**

Fusion between the chromosome 21 and another chromosome : most common t (14; 21) or t (22; 21)

Only acrocentric chromosomes can lead to robertsonian translocation : group D (13-14-15) and group G (21-22)



Genetics of trisomy 21

- **Chromosome 21 ring**

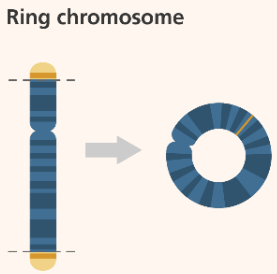
Rare chromosomal disorder with a breakage of chromosome and loss of genetic material at both ends, the ends joining together to form a ring

Amount of material genetic lost at both ends vary

Phenotypic variability linked to the amount and location of genetic material lost

Chromosome 21 ring linked to a *de novo* anomaly during early embryonic development

In most cases parents are unaffected



Source: https://rarediseases.org/rare-diseases/chromosome-21-ring/

Physical Features- General

- Short stature
- Generalized hypotonia
- Polydactily/syndactily/ clinodactily
- Hyperextensibility of joints
- Dermatoglyphic abnormatilies
- Skeletal abnormalities
- Protuberant abdomen
- Hypogonadism and delayed puberty

Physical Features- Craniofacial

- upslanting palpebral fissures (mongoloid), ocular defects
- short ears
- flat face with a broad nose bridge
- follicular hyperkeratosis
- persistent facial flushing
- alopecia areata (*)
- dry skin
- brachycephaly
- flat occiput
- broad and short neck
- hypoplasia of the maxilla
- small chin

(*) unpredictable, patchy hair loss

Clinical Features 1

- Mild to moderate intellectual disability:
  - Limited ability to learn, communicate and adapt
  - Delayed or impaired language development (they understand more than they can verbalize).
  - Ordinary activities of daily living and understanding the behaviour of others as well as their own can represent challenges.

Clinical Features 2

- Cardiac disorders
  - mitral valve prolapse occurs in more than half of all adults with this developmental disability.
  - Many others are at risk of developing valve dysfunction that leads to congestive heart failure, even if they have no known cardiac disease

- Compromised immune system
  - lead to more frequent oral and systemic infections and a high incidence of periodontal.
  - Aphthous ulcers, oral *Candida* infections, and acute necrotizing ulcerative gingivitis are common.
  - Chronic respiratory infections contribute to mouth breathing, xerostomia, and fissured lips and tongue.

Clinical Features 3

- Hypotonia:
  - muscles in the mouth leading to an open bite
  - large skeletal muscles
  - muscles controlling facial expression and mastication result in problems with chewing, swallowing, drooling, and speaking
  - atlantoaxial instability, a spinal defect that increases the mobility of the cervical vertebrae and often leads to an unsteady gait and neck pain.
- Seizures
  - Especially among infants, but can usually be controlled with anticonvulsant medications.
  - The mouth is always at risk during a seizure
  - People with controlled seizure disorders can easily be treated in the general dental office.

Clinical Features 4

- Hormonal disturbances (diabetes, hypothyroidism)
- Higher risk of leukemia
- Hearing loss and deafness
  - may further complicate poor communication skills, but these, too, can be accommodated with planning.
  - Patients with a hearing problem may appear to be stubborn because of their seeming lack of response to a request.
- Visual impairments
  - strabismus (crossed or misaligned eyes)
  - glaucoma
  - cataracts

Orofacial features- The maxilla

The midfacial region may be underdeveloped, affecting the appearance of the lips, tongue, and palate.

- **The maxilla,** the bridge of the nose, and the bones of the midface region are smaller than in the general population, creating a prognathic occlusal relationship.
- Mouth breathing may occur because of smaller nasal passages.
- Often strong gag reflex due to placement of the tongue, as well as anxiety associated with any oral stimulation.

Orofacial features- The palate

- Although normal sized, may appear highly vaulted and narrow.
- This deceiving appearance is due to the unusual thickness of the sides of the hard palate.
- This thickness restricts the amount of space the tongue can occupy in the mouth and affects the ability to speak and chew.
- Cleft palate

Orofacial features-The lips

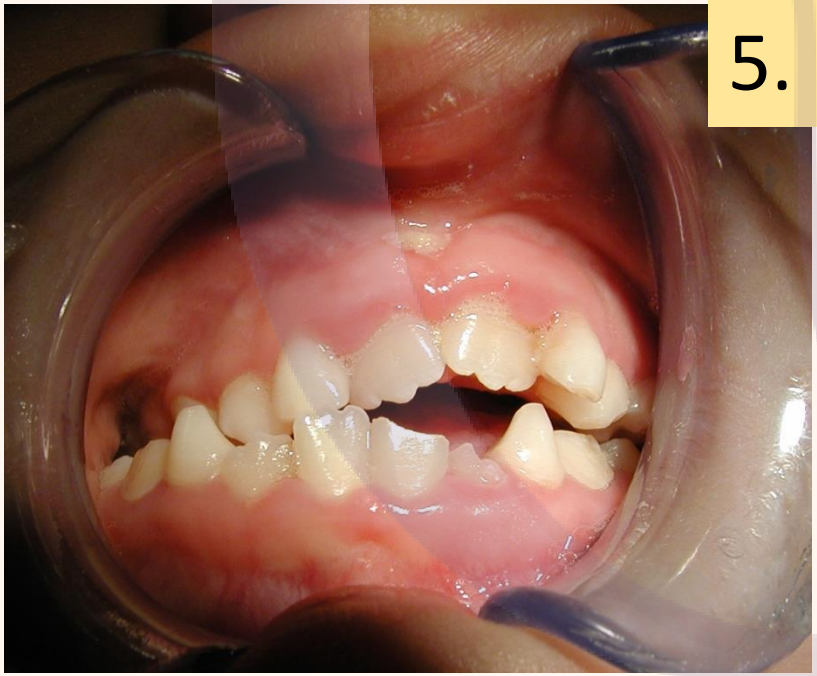
- Lips may grow large and thick.
- Fissured lips may result from chronic mouth breathing.
- Hypotonia may cause the mouth to droop and the lower lip to protrude.
- Increased drooling, compounded by a chronically open mouth → angular cheilitis.

Orofacial features- The tongue

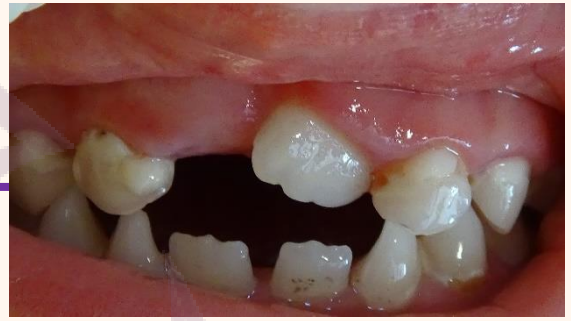
- Develops cracks and fissures with age; this condition can contribute to halitosis.
- Tongue may protrude because of a smaller midface region and poor muscular control.
- Persistent mouth opening due to the large tongue:
  - Mouth breathing
  - Drooling

Dental features

1. Malocclusion
2. Dental abnormalities
3. Periodontal disease
4. Dental caries
5. Dental trauma



Dental features - Malocclusion



- **Malocclusion** is caused by the delayed eruption of permanent teeth and the underdevelopment of the maxilla.
- A smaller maxilla contributes to an **open bite**, leading to **poor positioning of teeth** and increasing the likelihood of periodontal disease and dental caries.
- Malalignments in both primary and permanent dentition
- Angle III malocclusion – in 2/3 of DS individuals (due to underdevelopment of the midface)
- Anterior and posterior crowding, especially in maxilla

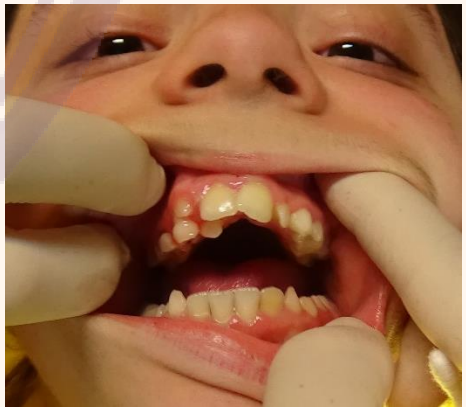
Dental features - Dental abnormalities

- **Congenitally missing teeth** (Third molars, laterals, and mandibular second bicuspids)

- **Delayed eruption of teeth,** often following an abnormal sequence. Primary teeth may not appear until age 2, with complete dentition delayed until age 4 or 5. Primary teeth are then retained in some children until they are 14 or 15.

- **Microdontia** and **malformed teeth-** crowns tend to be smaller, and roots are often small and conical, which can lead to tooth loss from periodontal disease.

- Severe illness or prolonged fevers can lead to **hypoplasia** and **hypocalcification**.



Dental features - Periodontal disease

- **Periodontal disease** is the most significant oral health problem in people with Down syndrome.
- Children experience rapid, **destructive periodontal disease**.
- **Loss of permanent anterior teeth** in early teens.
- Contributing factors:
  - Poor oral hygiene
  - Malocclusion
  - Bruxism
  - Conical-shaped tooth roots
  - Abnormal host response because of a compromised immune system.
- **Persistent gingival lesions**, prolonged wound healing, or **spontaneous gingival haemorrhage** may suggest an underlying medical condition and warrant consultation with the patient's physician.

Dental features - Dental caries

- **Children and young adults have fewer caries due to**:
  - delayed eruption of primary and permanent teeth;
  - missing permanent teeth;
  - small-sized teeth with wider spaces between them, which make it easier to remove plaque.
  - the diets are closely supervised to prevent obesity (reduced consumption of cariogenic foods and beverages).

- **Adults are at an increased risk of caries due to**:
  - xerostomia;
  - cariogenic food choices
  - hypotonia contributes to chewing problems and inefficient natural cleansing action (food remains on the teeth after eating).

Reported caries prevalence and caries experience indexes in children/young adults with Down Syndrome

Authors/ year	Country	Sample	Age (years)	Ip (%)	dmft	DMFT
Savin et al (2017)	Romania	25	6-12	92	8.56	
Al-Maweri & Al-Sufyami (2014)	Yemen	96	6-15	93.8	4.44±3.38	2.45±3.04
Ghaith et al. (2019)	UAE	106	4-18	57.6	3.42±4.15	3.32±4.62
Normastura et al. (2013)	Malaysia	53	11.7	57 (TT) 74(PT)	4.2±5.66	4.7±4.97
Morinushi et al. (1995)	Japan	74	2-18	53.9	-	-
Macho et al. (2008)	Portugal	138	2-26	28	-	-
Stabholz et al. (1991)	Israel	32	3-13	16	-	-

Reported met need (MNI) and restorative indexes (RI) in children with Down Syndrome

Authors/year	Country	n	Age (yr.)	Ip (%)	Met need index (MNI)*	Restorative index (RI)**
Savin et al (2017)	Romania	25	6-12	92	0.26	0.19
Al-Maweri & Al-Sufyami (2014)	Yemen	96	6-15	93.8	4.44	10.35
Ghaith et al. (2019)	UAE	106	4-18	57.6	40 (TT) 35.6 (PT)	27 (TT) 26.81 (PT)

(*) MNI=M+F/DMF

(**) RI=F/F+D

Dental features - Dental trauma

- Trauma and injury to the mouth may occur from falls or accidents.
- Suggest a **tooth-saving kit** for group homes.
- Emphasize to caregivers that trauma require immediate professional attention and explain the procedures to follow if a permanent tooth is knocked out.

- Instruct caregivers to locate any missing pieces of a fractured tooth, and explain that radiographs of the patient's chest may be necessary to determine whether any fragments have been aspirated.
- **Physical abuse** often results in oral trauma. Abuse is reported more frequently in people with developmental disabilities than in the general population.

People with Down Syndrome are prone to:

- **Bacterial endocarditis**:
  - Treat acute necrotizing ulcerative gingivitis and other infections aggressively.
  - Consult the patient's physician when in doubt about the medical history and the need for antibiotic prophylaxis (http://www.heart.org).

- **Seizures**:
  - Consult patient's physician. Record information in the chart about the frequency of seizures and the medications used to control them.
  - Determine before the appointment whether medications have been taken as directed.
  - Know and avoid any factors that trigger your patient's seizures.
  - Be prepared to manage a seizure.

Oral care for children with Down Syndrome. Focus on:

- Schedule patients with Down syndrome **early in the day** if possible. Early appointments can help ensure that everyone is alert and attentive and that waiting time is reduced.

- Before the appointment, obtain and review the patient's **medical history.**

    - Consultation with physicians, family, and caregivers is essential to assembling an accurate medical history.

- Determine who can legally provide **informed consent** for treatment.

Oral care for children with Down Syndrome. Focus on:

- Set the stage for a successful visit by involving the **entire dental team**—from the receptionist's friendly greeting to the caring attitude of the dental assistant in the operatory.

- Provide oral care in an environment with **few distractions**. Try to reduce unnecessary sights, sounds, or other stimuli that might make it difficult for your patient to cooperate.

- **Listen actively**, since speaking may be difficult for people with Down syndrome. Show your patient whether you understand.

- Talk with the parent or caregiver to determine your patient's intellectual and functional abilities, then **explain** each procedure at a level the patient can understand.

- Use simple, clear instructions, and repeat them often to compensate for any short-term memory problems.

Oral care for children with Down Syndrome. Focus on:

Behaviour management

- is not necessarily a problem; people with Down syndrome tend to be warm and well behaved.
- Some can be stubborn or uncooperative, but most just need a little extra time and attention to feel comfortable. Talk to the caregiver or physician about techniques they have found to be effective in managing the patient's behaviour. Share your ideas with them, and **find out what motivates the patient**.
- Gaining the patient's **trust** is the key to successful treatment.
- Nitrous **sedation** may be useful in some cases.
- Early settlement of a **Dental Home** (ideally by age 1) helps.

Oral care for children with Down Syndrome. Focus on:

- **Record** in the patient's chart strategies that were successful in providing care. Note your patient's preferences and other unique details that will facilitate treatment, such as music, comfort items, and flavour choices.

- Comfort people who resist oral care and **reward cooperative behaviour** with compliments throughout the appointment.

Oral care for children with Down Syndrome. Focus on:

- Maintain a **clear path for movement** throughout the treatment setting.
- Determine the **best position** for your patient in the dental chair and the safest way to move his or her body, especially the head and neck.
- Talk with the physician or caregiver about ways to **protect the spinal cord**. Use pillows to stabilize your patient and make him or her more comfortable.
- Use **immobilization** techniques only when absolutely necessary to protect the patient and staff during dental treatment—not as a convenience.
- Obtain consent from your patient's legal guardian and choose the least restrictive technique that will allow you to provide care safely. **Immobilization should not cause physical injury or undue discomfort**.

Oral care for children with Down Syndrome. Focus on:

- Plan a **step-by-step** evaluation, starting with seating the patient in the dental chair. If this is successful, perform an oral examination using only your fingers. If this, too, goes well, begin using dental instruments.

- **Prophylaxis** is the next step, followed by dental radiographs. Several visits may be needed to accomplish these tasks. Try to be consistent in all aspects of providing oral health care.

- Use the same staff, dental operatory, appointment times, and other details to help sustain **familiarity**.

- The more **consistency** we provide for our patients, the more likely that they will be cooperative.

Important for the Paediatric Dentist

- Examine a child by his or her first birthday and **regularly** thereafter to help identify unusual tooth formation and patterns of eruption.

- Consider using a **panoramic radiograph** to determine whether teeth are congenitally missing. Patients often find this technique less threatening than individual films.

- When permanent teeth are missing maintain primary teeth for as long as possible. Consider placing space maintainers where teeth are missing. Consider **orthodontic interception** whenever possible.

- Talk to patients and their caregivers about **preventing oral infections** with regular dental appointments and daily oral care.

Important for the Paediatric Dentist

- Advise patients taking medicines that cause xerostomia to **drink water** often.
- Suggest taking **sugar-free medicines** if available and rinsing with water after dosing.
- Recommend **preventive** measures such as topical fluoride and sealants. Suggest fluoride toothpaste, gel, or rinse, depending on your patient's needs and abilities.
- Emphasize **non-cariogenic foods** and beverages as snacks. Advise caregivers to avoid using sweets as incentives or rewards.
- Use **lip balm** during treatment to ease the strain on patient's lips.

Home Oral Care

- Some people with Down syndrome can brush and floss independently, but many need help.
- **Encourage independence** in daily oral hygiene. Ask patients to show how they brush, then follow with specific advice.

- Recommend adapted brushing methods or toothbrush **adaptations**. Involve patients in hands-on demonstrations of brushing and flossing.
- **Talk** to the caregivers about daily oral hygiene. Do not assume that all caregivers know the basics; **demonstrate** proper brushing and flossing techniques.

Home Oral Care

- A **power toothbrush** or a **floss holder** can simplify oral care.
- Use own experience with each patient to demonstrate sitting or standing positions for the caregiver.
- Some patients benefit from the daily use of an antimicrobial agent such as **chlorhexidine**. Recommend an appropriate delivery method based on your patient's abilities. Rinsing, for example, may not work for a person who cannot expectorate. Chlorhexidine applied using a spray bottle or toothbrush is equally efficacious.
- If use of particular medications has led to gingival hyperplasia, emphasize the importance of daily oral hygiene and frequent professional cleanings.
- Emphasize that a **consistent approach to oral hygiene** is important—caregivers should try to use the same location, timing, and positioning.

Important for the Orthodontist

In and of itself, Down syndrome is not a barrier to orthodontic care.

BUT

- Orthodontics should be carefully considered in people with Down syndrome. Some may benefit, while others may not.

- The ability of the patient or caregiver to maintain good daily oral hygiene is critical to the feasibility and success of treatment.

SO

Setting **realistic objectives** is crucial in order to avoid disappointment and/or frustration for patient, family and the dental team.