

DOWN SYNDROME

Down syndrome comprises a number of genetic disorders, Trisomy 21 being the most common of these. Such genetic disorders give rise to learning and physical disabilities. Down syndrome was first described by John Landon-Down, who originally called the condition "mongolism".

Epidemiology

Down syndrome occurs in all areas of the world and among all racial groups. The prevalence is 1 in 650-800 live births. As such, Down syndrome is the most common chromosomal abnormality.

Aetiology

Approximately 95% of Down syndrome cases are due to Trisomy 21. A trisomy is the presence of three, instead of the normal two chromosomes.

Clinical features

Physical characteristics of neonates with Down syndrome include:

- Brachycephaly — 63-98 %
- Oblique (up-slanting) palpebral fissures — 70-98 %
- Gap between first and second toes — 44-97 %
- Loose skin on nape of neck — 17-94 %
- Hyper-flexibility — 47-92 %
- Single palmar crease — 42-64 %

Other characteristics of neonates with Down Syndrome include:

- Congenital cardiac defect (highly specific for Down syndrome) — 40-50 %
- Transient myelodysplasia (highly specific for Down syndrome) — 10 %
- Duodenal atresia (highly specific for Down syndrome) — 5-8 %

Medical conditions associated with Down syndrome

[From: Desai (1997);¹ Saenz (1999);⁴ Gorlin *et al.* (2001);⁵ Cooley and Graham (1991);⁶ Myers and Pueschel (1995);⁷ Pueschel *et al.* (1995);⁸ Pilcher (2001)⁹]

Cardiovascular anomalies

About 40-50% of neonates with Down syndrome exhibit some type of cardiac abnormality. Approximately 20% of these are the cause of death of the patient.

Immune anomalies

Patients with Down syndrome often exhibit impaired immunity. This is due to a variety of potential defects, including [Desai (1997);¹ Sasaki *et al.* (2004)¹⁰]:

- defective and short-lived neutrophils
- impaired cell-mediated immunity
- irregular serum immunoglobulin patterns .

As a result, Down syndrome children may have a higher rate of infection, including respiratory, gastrointestinal, mucosal and dermal infections. From an oral perspective, these children often exhibit periodontal disease, and there is an increased prevalence of aphthous ulcers, oral Candida infection and acute necrotizing ulcerative gingivitis [Pilcher (2001)⁹].

Haematologic anomalies

Children with Down syndrome are at greater risk of developing leukaemia.

Musculoskeletal anomalies

Children with Down syndrome commonly exhibit musculoskeletal anomalies. These include atlanto-axial instability, mid-facial hypoplasia and relative prognathia of the mandible.

The implications of atlanto-axial instability pertain to the risk of its dislocation and injury to the spinal column. As such, extreme care is required during intubation, orientation of the head by the paediatric dentist during general anaesthesia and head tilt during emergency management of the collapsed patient.

Nervous system anomalies

Children with Down syndrome often exhibit delayed motor function and reduced muscle tone. This manifests as delayed coordination and reduced strength.

Mental retardation

The IQ of persons with Down syndrome rarely exceeds 60.

Gastrointestinal anomalies

GIT anomalies associated with Down syndrome include [Kallen *et al.* (1996)¹¹]:

- Oesophageal Atresia
- Pyloric stenosis
- Duodenal atresia

Approximately 8% of Down syndrome patients die as a result of their gastrointestinal anomalies.

Ophthalmologic anomalies

Ophthalmologic anomalies associated with Down syndrome include:

- Congenital and acquired cataracts
- Nystagmus
- and many others

Speech anomalies

Expressive language in Down children is more delayed than receptive language.

Endocrine anomalies

Down syndrome patients exhibit a higher prevalence of diabetes and hypothyroidism [Kieser (2003)¹²].

Respiratory

Children with Down syndrome often have chronic respiratory problems.

Oral and craniofacial manifestations/associations of Down syndrome

A variety of characteristic oro-facial anomalies and problems present in children with Down syndrome. The most common of these are [McDonald *et al.* (2004)¹³]:

1. Skeletal anomalies
2. Periodontal disease
3. Delayed eruption
4. Developmental defects of enamel
5. Hypodontia
6. Supernumerary teeth
7. Atypical patterns of eruption
8. Ectopic eruption
9. Abnormal dental morphology
10. Orthodontic problems
11. Bruxism

These are discussed below in their various categories.

Skeletal

The primary skeletal anomaly affecting the orofacial structures in Down syndrome is hypoplasia of the mid-facial region, with the bridge of the nose, bones of the mid-face and maxilla being relatively small in size.

Many children with Down syndrome children exhibit a narrow palate, probably due to the sides of the palate being abnormally thick. This narrowness may sometimes give the impression of a high palatal vault; however the height is generally not significantly greater than normal [Desai and Flanagan (1999)¹⁵].

Cleft lip and/or palate are reported in 3%-5% of Down syndrome patients [Gorlin *et al.* (2001);⁵ Kallen *et al.* (1996)¹¹].

Soft tissues

Tongue

Down syndrome patients give the impression of macroglossia. However, this is generally a relative macroglossia; true macroglossia is very rare. The relative macroglossia is attributed to the oral cavity being reduced in size, due to the mid-facial hypoplasia and narrow palate [Desai and Flanagan (1999)¹⁵]. The implications of this include difficult access for oral treatment and difficulty with speech and mastication.

Dentition

Dental features associated with Down syndrome are varied, and sometimes contrasting. They include [Gorlin *et al.* (2001)⁵]:

- Microdontia
- Hypodontia
- Supernumerary teeth
- Spacing between teeth
- Taurodontism
- Variation in crown morphology
- Developmental defects of enamel
- Delayed eruption
- Ectopic eruption

Caries

A reduced prevalence of caries is reported in Down syndrome compared to children with other forms of mental retardation and unaffected children. Earlier epidemiologic reports of dental caries in patients with Down syndrome have reported extremely low rates [Barnett *et al.* (1986)¹⁷]. However, these have since been identified as biased, due to the fact that most study groups were children in institutions where their diet was strictly controlled. More recent studies have show that while there is still a reduced prevalence of dental caries in Down syndrome children, it is not as uncommon as once thought. Proposed reasons for the lower prevalence of caries include delayed eruption of the primary and permanent teeth, increased spacing between teeth or possible increased proportion of protective salivary components in Down syndrome children [Sasaki *et al.* (2004)¹⁰].

Developmental defects of enamel

Developmental defects of enamel, primarily enamel hypoplasia and enamel hypocalcification are relatively common in Down syndrome children [Gorlin *et al.* (2001);⁵ Cohen and Winer (1965);¹⁸ Cohen and Cohen (1971)¹⁹]. This may involve both the primary and permanent dentition.

Erosion

Dental erosion and tooth wear is more frequent in children with Down syndrome, with both attrition and erosion being contributing factors. Gastric reflux and vomiting are reported in over 20% of children with Down syndrome. These children are also more likely to grind their teeth [Bell *et al.* (2002)²⁰].

Orthodontic

Hypoplasia of the mid-facial region is common in Down syndrome children. In many instances this causes a relative prognathic Class III occlusal relationship which may contribute to an open bite [Vitteck *et al.* (1994)¹⁴].

Other common orthodontic features of Down syndrome include irregular alignment of the teeth, posterior crossbite, anterior open bite, crowding of teeth, and widely spaced teeth [Cohen and Cohen (1971)¹⁹].

Down syndrome patients tend to have teeth with small conical roots. This is an important factor when considering orthodontic tooth movement and also contributes to early tooth loss in periodontal disease [Pilcher (2001)⁹].

Eruption

The eruption of teeth is delayed in 75% of cases of Down syndrome. The delay in eruption may be as long as two to three years.

Hypodontia

Hypodontia in both the primary and permanent dentition has been reported in 23%-47% of patients.

Morphology

Between 35% and 55% of patients with Down syndrome present with microdontia in both the primary and permanent dentition [Desai (1997)¹]. Clinical crowns are frequently conical, shorter and smaller than normal.

Periodontium

Early, severe periodontal disease is often seen in patients with Down syndrome. The prevalence of periodontal disease in Down syndrome adults has been reported as 90% [Gorlin *et al.* (2001);⁵ Shapira *et al.* (1991)²²]. Onset of the condition usually occurs in the mid teens; however, severe periodontal involvement has been frequently reported in children 6 yoa and older.

Dental management of patients with Down syndrome

General management

Cooperation

Down syndrome children vary widely in the degree of intellectual impairment. Most have IQ's in the mild to moderate range and are able to be treated in a normal setting. Patients with Down syndrome are characteristically gentle, tolerant and eager to please (some may exhibit anxiety and stubbornness). As such, age- and intellect-appropriate conventional behaviour management techniques may be employed with success when treating Down syndrome patients [Pilcher (2001)⁹].

Communication

Despite the fact that there is often a delay in language development, it is generally perceived that patients with Down syndrome tend to understand more than their verbal skills would belie. Notwithstanding this, the primary caregiver should always be encouraged to be present for any consultation or treatment to facilitate consent, cooperation and communication.

General anaesthetic

General anaesthesia is frequently employed to facilitate treatment of paediatric dental Down syndrome patients. It is essential to note that the physical and medical associations of Down syndrome may have important implications for general anaesthesia. Peri-operative morbidities result mainly from problems with the respiratory system (Down syndrome children tend to have chronic respiratory conditions) and spinal cord damage from atlanto-axial subluxation. Comprehensive examination and consultation with the anaesthetist is essential prior to general anaesthesia [Roizen (2002)³].

Prevention

As with all medically compromised children, prevention is the key to any treatment approach. This incorporates extensive and regular oral hygiene motivation, topical fluoride, fissure sealants and cleaning [Pilcher (2001);⁹ Hennequin *et al.* (1999)²⁹].

Children with Down syndrome often exhibit delayed motor function and reduced muscle tone. This manifests as delayed coordination and reduced strength. Oral hygiene should be performed by the primary caregiver until the patient acquires adequate skill. Those patients who exhibit significant intellectual impairment represent a greater challenge to maintaining oral hygiene.

Restorative

All restorative treatment is indicated. Attention to the presence of erosion and developmental defects of dentine is essential. In such cases, full coronal coverage may be preferred.

Periodontal disease

Prevention is the key to management of periodontal disease. This includes optimal oral hygiene, routine review, routine prophylaxis and use of medicaments such as chlorhexidine as indicated. Notwithstanding this, the immune anomalies seen in Down syndrome children may circumvent even the best preventive regimen. In such cases, early, aggressive periodontal treatment is indicated.

Orthodontic

Orthodontic management of malocclusion in children with Down syndrome is considered problematic. This is due to the fact that compliance is perceived to be low [Backman *et al.* (2003)³¹] due to an inability to understand the need to wear appliances and the inability to adequately maintain oral hygiene. This leads to a high risk of treatment failure, caries and periodontal disease.

In some cases, microdontia and hypodontia may alleviate crowding. In other cases, selective extractions following orthodontic workup, may be beneficial.

Pulp therapy

Avoid pulp therapy in the primary dentition of children with Down Syndrome, who have a congenital cardiac defect.

Medical considerations

Dental management must include adequate investigation of and precautions for any concomitant medical conditions.