the Child Dental Patient with

Neurofibromatosis

ASOCIAȚIA NAȚIONALĂ DE STOMATOLOGIE PEDIATRICĂ
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Definition

- a group of three different tumor disorders including:

neurofibromatosis type I (NF1)
(von Recklinghausen's disease)

neurofibromatosis type II (NF2)

neurofibromatosis – type III (NF3)
(schwannomatosis)

- show different gene origination and clinical characteristics but exhibit the same autosomal dominant mode of inheritance.

Etiology

- NF1 - the gene locus is 17q11.2
- NF2 - gene mutations in a specific region of chromosome 22q
- NF3 - mutations of the genes SMARCB1 and LZTR1 of chromosome 22q

NF1+ NF2: approximately 50% of cases - new mutations

NF3 : about 10% are familial

Epidemiology

- **NF1** – the most common (about 90% of cases of NF)

Prevalence

- NF1 – 1:25,000
- NF2 – 1:60,000
- NF3 - ?

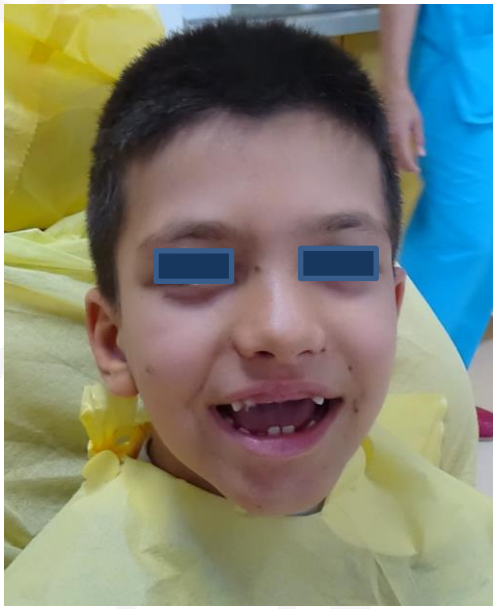
Incidence

- NF1 – 1: 3,000-4,000
- NF2 – 1:25,000-40,000
- NF3 – 1: 40,000- 1,700,000

NF1 – signs and symptoms

- **Multiple cafe au lait spots**

(flat, light brown spots on the skin) – begin in early childhood, increase in size and number with age.



- **Axillary and inguinal freckling -** appears in late childhood, occurs in clusters in skin folds.
- **Lisch nodules** (pigmented hamartomas of the iris) - don't affect vision.
- **Multiple neurofibromas (benign complex tumors from peripheral nerve sheaths) –** can be localized **/**plexiform .

NF1 – signs and symptoms (2)

- **Bone deformities -** scoliosis or bowed lower leg.
- **Optic glioma -** appears by age 3, rarely in late childhood and adolescence, and almost never in adults.
- **Learning and writing disabilities -** usually mild;
- **ADHD** - is common.
- **Epilepsy**
- **Macrocephaly**
- **Short stature**

NF1

- One of the most frequent human genetic diseases, with no sex or race predilection
- Occurs in 1:200 inhabitants with mental retardation
- Variable expressivity:
  - 60% - mild cases
  - 20% - moderate
  - 20% - severe
- Progressive condition:
  - Increase in size and number of localized neurofibromas with age (especially during puberty and pregnancy)
  - plexiform neurofibromas – increase in size (can cause disfigurment) and have risk of malignancy!!!

NF1 – head and neck findings

- Multiple localized neurofibromas – scalp, cheek, neck and oral cavity
- Facial asymmetry – due to plexiform neurofibromas on the face
- Facial disfigurement – due to hypoplasia or hyperplasia of the jaws, the zygomatic bone and the temporomandibular joint.
- Exophthalmia - caused by sphenoid wing dysplasia.

NF1 - oral findings

- Lengthening of fungiform papillae – the most frequent finding
- Localized oral neurofibromas
  - asymptomatic nodules covered by normally colored mucosa
  - most commonly in tongue
  - may determine impairment of motor function of nerves VII or IX or the sensitivity of the nerve V
  - may cause malposition of the teeth (partial or total retention)
- Supernumerary/missing teeth
- Macroglosia

Oral radiographic findings

- Enlarged mandibular canal, mandibular foramen and mental foramen.
- Well demarcated unilocular, but occasionally multilocular, radiolucent lesions (intraosseously developed neurofibromas)
- lengthening, narrowing and rarefaction of coronoid and articular process
- deepening of sigmoid notch

Important to know

- Oral manifestations – are frequent (about 70% of NF1 patients)
- NF1 can determine esthetic and functional problems (mastication, speech)
- Orthodontic treatment can be performed
- Gingival neurofibromas can be confused with periodontal disease and can cause subsequent periodontitis
- Difficulties in oral surgical procedures and in the maintenance of teeth
- Longterm follow-up is mandatory because of the potential for local complications and the risk of malignant tumors.

NF2 – signs and symptoms

- Generally appear in the late teen and early adult years
- Multiple schwannomas and meningiomas, which bilaterally affect the vestibular nerve, potentially leading to hearing loss and deafness.
- Initial symptoms:
  - hearing loss frequently unilateral
  - tinnitus
  - dizziness, loss of balance.
- In later stages of the disease - vomiting or true vertigo.

NF2 – signs and symptoms

- headaches
- seizures

} intracranial meningioma

- pain
- muscle weakness
- paraesthesia

} spinal tumour

- cutaneous tumour
- reduced visual acuity due to cataract, optic nerve meningioma, extensive retinal hamartomas

NF2 – oral findings

- in cases of impaired vision:
  - high prevalence of caries (due to poor oral hygiene)
  - dental traumatic injuries in permanent and primary dentition

Dental management

- Hearing impairment can cause:
  - problems in communications during dental appointments
  - increased dental fear and anxiety
- Special communication methods in achieving patient's cooperation during the dental treatment:
  - ”Tell-show-do” technique
  - Tactile finger spelling
  - Use of laptop and tablet - voice command, “text-to-speech” operating systems, and high or reverse contrast

NF3 – signs and symptoms

- usually appear in early adulthood
- multiple schwannomas (nerve sheaths tumors) of the peripheral nervous system without concomitant involvement of the vestibular nerves
  - there are throughout the body or in isolated regions
- chronic pain – the most common symptom
  - ranges from mild to severe
- dysesthesia and paresthesia may also be present.
- common localizations: the spine, peripheral nerves, and the cranium.

NF3 – oral features

- Only 1% of all schwannomas are located in the oral cavity
- The treatment of choice for solitary schwannomas is conservative surgical excision.