Neurofibromatosis I

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Overview

Neurofibromatosis I (NSF1), also known as Von Recklinghausen's neurofibromatosis, is a common genetic autosomal dominant hereditary disorder. According to a case report on NSF in the Journal of Pigmentary Disorder, "this is the most common type of neurofibromatosis, accounting for 90% of all cases having a prevalence of approximately 1 in 3,000".

Etiology

Neurofibromatosis I is a hereditary disorder. If a birth parent has the abnormal gene, it likely they can pass the genetic defect onto their newborn. It also possible for genes to mutate on their own, leading to the disorder. Neurofibromatosis I effects both sexes, with no predisposition, and all races equally.

Clinical Presentation

Neurofibromatosis I is not limited to one particular cell in the effected body; it is expressed in all of the bodies cells. Because of this, the clinical presentations of NSF1 can be cutaneous, ophthalmological and neurological. Neurofibromatosis I may therefore lead to serious esthetic issues, learning difficulties, intellectual defects and even psychological and social problems.

According to the National Institute of Neurological Disorders and Stroke, to diagnose NF1 a doctor looks for two or more of the following:

- Six or more "café-au-lait" spots, measuring more than 5 millimeters in diameter in children or more than 15 millimeters across in adolescents and adults;
- Two or more neurofibromas, or one plexiform neurofibroma
- Freckling in the area of the armpit or the groin;
- Two or more growths on the iris of the eye
- A tumor on the optic nerve (called an optic nerve glioma)
- Abnormal development of the spine (scoliosis), the temple (sphenoid) bone of the skull, or the tibia (one of the long bones of the shin); or
- A parent, sibling, or child with Neurofibromatosis I

In NF1 "bone abnormalities are reported between 50% to 70% and oral manifestations in 70% of cases. The most common oral lesions are increased fungiform papillae, intraosseous cystic lesions, branches of the mandibular canal, and enlarged foramen and mandibular canal. Only 25% of patients with neurofibromatosis present intraoral neurofibromas. In addition, specific skeletal injuries of the jaws include coronoid process enlargement, condylar deformity, condylar neck stretching, irregularities in mandibular cortical, thinning and lateral curvature of mandibular ramus (Trindade, Elias, Migliolo, Nogueira, Ferraz, 2015, pg.1)

Demographic
Since Neurofibromatosis I is a genetic disorder, there is no specific demographic. Anyone who has a parent with the gene, or endures gene mutation during fetal growth for Neurofibromatosis I is at risk for developing the disorder.

Biopsy / Histology / Radiographs
The diagnosis of Neurofibromatosis I is based on clinical assessment and therefore "biopsy of asymptomatic cutaneous neurofibromas should not be undertaken for diagnostic purposes in individuals with clearcut NF1" (Ferner, Rosalie E et al, 2017, pg. 1)

The histological features of Neurofibromatosis I are that it is not encapsulated with a cellular fibrous appearance suggestive of nerves. No thickening of vessel walls but displays fibroblast and collagen.

The radiographic appearances of Neurofibromatosis I are very visible on radiographs. For NSF1 effecting the skull, chest, arms, and legs the abnormalities are well demarcated with a radiopaque appearance. As dental hygienists, we are likely to see: "enlarged orbits, enlargement of orbital margins, facial asymmetry, hypoplasia of the paranasal sinuses, mandibular abnormalities, mandibular hypoplasia with flattening of the external contour, thinning of the ramus, coronoid hyperplasia, widening of the lateral and medial coronoid spaces appear on the radiograph(s)" (Khan, 2016, pg. 2).

Differential Diagnosis
list all the pathology that this could reasonably be mistaken for:

Neurofibromatosis I does have very common clinical presentations that may present overlap with some other diseases. Some of the most common presentations that could be misleading are: "cafe au lait" spots, localized hyper growth syndromes, neurofibromas-like tumors and multiple endocrine neoplasia.

The types of pathology that Neurofibromatosis I may be mistaken for are:

- Other "cafe-au-lait" conditions: Isolated CAL, Familial CAL, Legius Syndrome, DNA Repair Deficiency Syndrome, NF1-Noonan Syndrome Piebaldism, Bloom Syndrome, Fanconi Anemia CAL or skin hypopigmentation; short stature; malformations of the thumbs, forearms; progressive bone marrow failure; autosomal recessive. (Rodrigues et al., 2014, pgs. 8,9)
- Skin disorders with CNS tumors: Tuberous Sclerosis Complex, McCune-Albright, Ataxia-telangiectasia, Gorlin Syndrome (nevoid basal cell carcinoma syndrome), Sturge-Weber syndrome (encephalotrigeminal angiomatosis). (Rodrigues et al., 2014, pgs. 8,9)
- Maculae conditions misdiagnosed with "café-au-lait": LEOPARD Syndrome, Neurocutaneous melanosis, Peutz-Jeghers, Klippel-Trenauney-Weber Syndrome, Proteus Syndrome, Lipomatosis, Banayan-Riley-Ruvalcaba Syndrome, Congenital generalized fibromatosis, Endrocrine multiple neoplasias type 2B (MEN2B). (Rodrigues et al., 2014, pgs. 8,9)

Treatment
There is no one specific treatment that exists for Neurofibromatosis I. A combination of collaborative treatments and surgical therapies are utilized depending on the specific treatment and present anomalies.

Surgery is regularly recommended to remove tumors that become symptomatic and may become cancerous, as well as for tumors that cause significant cosmetic disfigurement (including the reduction of optic nerve gliomas are bone malformations). Radiation or chemotherapy are also possible treatment methods.

The treatment for other conditions associated with Neurofibromatosis I are designed to control or relieve symptoms (ex. headaches and seizures are treated with medications).

Prognosis
Neurofibromatosis I is a progressive disorder, where most symptoms will worsen over time, although it is possible that some people may have symptoms that remain constant.

Although the prognosis for someone with Neurofibromatosis I varies, overall patients with neurofibromatosis I have a life expectancy approximately half that of non-affected individuals. This is likely due to the individual succumbing to a malignancy that has developed in their tumor(s) tumors or from cardiovascular complications. However, most people with Neurofibromatosis I will only develop mild to moderate symptoms.

Professional Relevance
An individual's oral health condition is threatened in patients with neurofibromatosis I. There are several conditions that a patient with Neurofibromatosis I may present with, such as: gingival enlargement, impacted or supernumerary teeth, cemental dysplasia, nodular lesions that appear on the tongue, buccal musoca or even the floor of the mouth. The pathology dental health professionals are not limited to the ones previously listed, but we are to expect diminished oral health in any patient who is immunocompromised. It our duty as dental health care professionals to know the proper way to care for patients with Neurofibromatosis I and to help them maintain a positive oral health status as to not contribute to the progression of their disorder.

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