Throughout my short time in this Oral Pathology course, it has become fairly known that most pathology whether a lesion, tumor, syndrome or disease is usually named as an eponym; in the honor of an individual who described or died from the disease. However, every now and then we come across diseases/disorders that are named after the characteristics most closely associated with that particular disease. In the case of Cherubism, it was first described in 1933 by a Canadian radiologist named William A. Jones as “having full round cheeks and an upward cast of the eyes which gave the children a peculiarly cherubic appearance.” Medically, Cherubism is a rare bone disorder that occurs specifically in the bones of the jaw; both in the maxilla and mandible.

Cherubism, also known as multilocular cystic disease of the jaws is an autosomal dominant condition that takes place as a result of multiple mutations in the SH3BP2 gene on Chromosome 4. This gene acts as a protein-coding gene and is responsible for transmitting certain signals in certain immune system cells as well as cells involved in bone remodeling; replacement of old bone tissue with new bone tissue. In Cherubism specifically, both the maxilla and the mandible become enlarged as bone is resorbed resulting in numerous cavities. These cavities begin to fill with fibro-osseous tissues that are constantly expanding. Over time, these lesions are capable of invading the eye sockets and can displace the eyes and push down on the eyelids. Since Cherubism is autosomal dominant, it can be inherited and often run in families. However, as with most diseases/disorders, Cherubism can also be seen on individuals who do not have any known family history.

Clinically, Cherubism can have variable expressivity but is mostly characterized by bilateral symmetric swelling or enlargement of the jaw. Individuals may also present with a round face, broad cheeks, hypertelorism, maxillary enlargement, prognathism,

enlarged submandibular and cervical lymph nodes as well as swelling in the zygoma and the condyles. If the lesions progress into the area of the eye, the sclera below the iris can become visible which will result in an upward gazing look. Subjectively, individuals with Cherubism can have painless, swollen cheeks, a prominent lower face, enlarged jaw, abnormal teeth and thick gums. Other manifestations of this disorder can include severe enlargement causing visual, respiratory, speech, mastication and swallowing complications, lower lid retraction, exophthalmos or diplopia. Additionally, Cherubism is frequently accompanied by a significant number of dental abnormalities both in the arch and the teeth individually.

Cherubism affects males and females equally and has been reported in individuals of all racial and ethnic backgrounds. Individuals with Cherubism appear normal at birth. Swelling of the jaw begins to take place anywhere from 2-7 years of age. Because of the associated hormonal tweak, the swellings continue on, increasing in size and number until puberty. The progression of these fibro-osseous tissue filled cavities slowly comes to an end and are gradually replaced with normal bone. At approximately 30 years of age, the facial abnormalities subside and are usually no longer visible. However, during pregnancy, there may be a possibility of not only recurrence, but a 50/50 likelihood of passing the mutation of the SH3BP2 gene onto the child.

Because there is high variable expressivity and uniqueness associated with Cherubism, many believe that grading systems are not appropriate in determining the degree of Cherubism in an individual. Instead, clinicians direct their focus mainly on the lesions; both their size and rate of growth, dental abnormalities and functional deficits. Microscopically, there is an increased number of multinucleated giant cells present. Radiographically, there are extensive multilocular radiolucencies involving all four quadrants. In some cases, teeth may be unerupted, displaced and/or floating in cyst-like spaces.

The most important goal in the management and treatment of Cherubism should be emphasized on slowing down the disease as much as possible. Treatment options consist of intralesional steroid injections, systemic calcitonin administration, curettage of the lesions and recontouring of the jaw and face. Children are often referred to craniofacial clinics and are treated by a wide variety of clinicians with experience in pediatrics such as surgical teams, geneticists, dentists, orthodontists, ophthalmologists and social workers.

Because Cherubism is self-limiting and is said to improve over time, there are no well established treatment protocols. Treatment is often customized to the individual’s needs. Mild forms do not usually require any form of treatment. Instead, managing mild cases consists of long term observation and follow ups. Treatment of more severe cases focus additionally on the prevention of secondary complications such as early orthodontia and jaw reconstruction as well as long term follow up and the evaluation of relatives at risk.

The diagnosis of Cherubism is usually based on a combination of microscopic, radiographic and clinical findings including patient age, family history as well as the option to undergo gene testing. The combination of all findings is necessary because microscopically, Cherubism presents as an abundance of multinucleated giant cells in a stroma of fibrous connective tissue. The same is seem microscopically in Central Giant Cell Granuloma, Peripheral Giant Cell Granuloma, Brown Tumor of Parahyperthyroidism and Aneurysmal Bone Cysts. Gene testing allows for Giant Cell Granuloma to be ruled out. Clinical data allows for Aneurysmal Bone Cysts amongst others to be ruled out. Analysis of parathormone levels, calcium, phosphorous and alkaline phosphate concentrations allows for Hyperparathyroidism to be ruled out. Differentiation between all possible diagnoses is important because all of the diseases/syndromes mentioned behave differently, vary in aggressiveness and require different courses of treatment.

This disorder is relevant to me as a dental hygiene student mainly because the majority of individuals who present with Cherubism are children with parents that will be concerned about at least one of the long list of dental abnormalities that result as a consequence of this disorder. In fact, if the child began attending dental appointments at the appropriate age, the dental hygienist may be one of the first clinicians to note any substantial changes. Dental abnormalities range depending on the time of onset and severity of lesions. As a dental hygiene student, it is important to recognize abnormalities that can occur in the primary dentition including a disrupted arrangement, malocclusion, undeveloped molars, missing teeth and premature exfoliation. In the secondary dentition there are commonly malocclusion, absent teeth, abnormally shaped teeth, delayed eruption, resorbed roots, impactions and teeth that are “free floating” in cherubism lesions. As a dental hygiene student, the focuses lie on encouraging the individual to undergo restoration of any carious teeth and on ensuring thorough professional oral prophylaxis as well as an effective customized oral hygiene routine.

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Cherubism

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