

CHAPTER 18

GENETIC AND METABOLIC DISEASES

I. GENETIC DISEASES:

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Osteogenesis Imperfecta
Osteopetrosis
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Mandibulofacial Dysostosis (Treacher Collins Syndrome)
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Clefts of the Lip and Palate
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Sickle Cell Anemia
Thalassemia
Hemifacial Hypoplasia
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II. METABOLIC DISEASES:

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Hypoparathyroidism
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Hypopituitarism
Hyperthyroidism
Hypothyroidism
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Rickets and Osteomalacia
Hypophosphatasia
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Scleroderma
Infantile Cortical Hyperostosis

ECTODERMAL DYSPLASIA

Ectodermal dysplasia is a syndrome which is transmitted as an X-linked recessive disorder and occurs almost exclusively in males. This hereditary developmental disturbance affects the ectodermal tissues like hair, fingernails, sweat glands and teeth. Patients exhibit soft, smooth, thin, dry skin with partial or complete absence of sweat glands. Since these patients cannot perspire normally because of the agenesis of sweat glands, they are unable to endure warm temperatures. The hair of the scalp and those of the eyebrows tend to be fine, scanty, blonde and resemble lanugo. Body hair is sparse or absent. The finger and toe-nails are small and deformed or absent. The bridge of the nose may be depressed and the supraorbital ridges may be very prominent (frontal bossing). Oral manifestations include total anodontia or hypodontia. The teeth which are present are often malformed; frequently they are cone-shaped.

Fig. 18-1 Ectodermal dysplasia. Patient exhibiting hypodontia.

CHERUBISM (Hereditary fibrous dysplasia of the jaws, Familial intraosseous swellings of the jaws, Familial multilocular cystic disease of the jaws)

Cherubism is a hereditary condition that produces firm, painless swellings that occur bilaterally in the jaws, especially over the mandibular angles. This autosomal dominant disease is usually detected between the ages of 2 and 7 years. The mandible is more frequently affected and produces full, round lower face similar to those of cherubs portrayed in Renaissance religious paintings. Often the eyes are upturned to reveal the white sclera beneath. In some cases, where the maxilla is involved, the skin of the cheeks appears to be stretched. This benign fibro-osseous disease does not affect the other bones of the body. The bony defects contain soft tissue that is histologically similar, if not identical, to a giant cell granuloma.

On a radiograph of the mandible, the expansion of the buccal and lingual cortical plates is seen on occlusal and postero-anterior views. The mandibular body and rami are frequently involved. Enlargement of the maxilla is at the expense of the maxillary sinuses. The lesion consists of multiple cyst-like radiolucencies which are located bilaterally in the jaws. These bilateral multilocular radiolucencies are a characteristic feature of cherubism. The teeth may be unerupted and displaced. Some of the teeth appear to be floating in the cyst-like spaces. This benign, self-limited disorder tends to regress after puberty, thus eliminating the need for surgical intervention. Treatment, if necessary, consists of recontouring the bone for cosmetic reasons after the lesions have stabilized in size.

Fig. 18-2 Cherubism. Panoramic radiograph showing extensive multilocular radiolucent lesions of the mandible and maxilla.

OSTEOGENESIS IMPERFECTA ("Brittle bones" disease, Fragilitas ossium, Lobstein's disease)

Osteogenesis imperfecta is a hereditary disorder of connective tissues, that is, there is an inborn error of collagen metabolism which produces an abnormality in the organic matrix of bone. Some cases occur as autosomal dominant traits whereas others occur as spontaneous mutations (idiopathic). The development of bones is poor and, therefore, results in their fragility, porosity and proneness to fracture. Although the fractures heal, the new bone formation is of imperfect quality. An exuberance of callus may be formed at the sites of repeated fractures. Bowing of the limbs and angulation deformities occur at previous fracture sites. A characteristic clinical feature is that of a patient having pale blue sclera caused by the pigmented choroid coat showing through the thinned sclera. Early hearing loss in a patient or a member of a family with a history of fragile bones is highly suggestive of the disorder.

The oral findings are similar to those seen in dentinogenesis imperfecta in which the crowns of teeth are of opalescent hue. Like bone, the dentin is poorly calcified and, therefore, the crowns fracture easily. The exposed dentin undergoes rapid attrition. Radiographic appearance is that of teeth constricted near their cervical portions and having thin, short roots. The pulp chambers and canals are calcified and obliterated. There are types I, II, III and IV of osteogenesis imperfecta, each having specific clinical features. It is the most common inherited bone disease and owing to its variable clinical expression, many mildly affected patients remain undiagnosed.

Fig. 18-3 Osteogenesis imperfecta. The dental changes are similar to those of dentinogenesis imperfecta: teeth constricted near the cervical portions, thin and short roots, calcification of pulp chambers and root canals. Clinically, the crowns are of opalescent hue.

OSTEOPETROSIS (Albers-Schönberg disease, Marble bone disease)

Osteopetrosis, also known as Albers-Schönberg disease or marble bone disease, is a rare disease of unknown etiology. In most cases heredity seems to play a role. The infantile type (malignant form) is inherited as an autosomal recessive trait whereas the adult type (benign form) is inherited as an autosomal dominant trait. Although both forms are congenital in origin, the adult type may not be noticed until later in childhood or adulthood. Osteopetrosis is characterized by skeletal, hematologic, and neurologic abnormalities. In skeletal abnormalities, there is an increased calcification and thickening of cortical and spongy portions of the entire skeletal (osseous) system. The formation of bone is normal but the resorption of bone is reduced owing to a reduction in osteoclastic activity. The bones become fragile and susceptible to fracture. In osteopetrosis, multiple fractures result from excessive mineralization; whereas in osteogenesis imperfecta, the fractures result from bone resorption. In hematologic abnormalities, secondary anemia occurs due to a reduction of available marrow because of extensive deposition of dense bone in the bone marrow. There may be enlargement of spleen and liver (hepatosplenomegaly). In neurologic abnormalities the excess bone formation narrows the various foramina in the base of the skull; this may produce blindness, deafness, diabetes insipidus, facial palsy, and neuralgic pain, depending on the nerve being compressed. The relative avascularity of the bone makes it vulnerable to infection, especially at the time of tooth extraction and results in osteomyelitis. Another complication that may result at the time of extraction is that the bone may fracture easily. In the infantile type of osteopetrosis, most patients die before age 20 as a result of anemia or infection.

In the clinically benign adult form, the life expectancy is usually normal, the disease may not be diagnosed until the third or fourth decade. Osteopetrosis can be diagnosed solely on the basis of appropriate radiographic findings. The bones of the body show greatly

increased density; their medullary cavities being replaced by bone. A lateral skull radiograph shows an increased radiopacity due to increased thickness of the bones in the skull, calvarium and all the structures at the base. Dental defects include delayed eruption, early loss of teeth, missing teeth, malformed roots and crowns.

Fig. 18-4 Osteopetrosis. Lateral skull radiograph shows dense calcification of jaws and skull, resulting in loss of trabeculae. All the sinuses are obliterated with bone.

CLEIDOCRANIAL DYSOSTOSIS

Cleidocranial dysostosis is an inherited skeletal anomaly which affects primarily the skull, clavicle and dentition. Some of the cases occur spontaneously but most are inherited in autosomal dominant mode. The skull findings are brachycephaly (reduced anteroposterior dimension but increased skull width), delayed or failed closure of the fontanelles, and the presence of open skull sutures and multiple wormian bones. There is pronounced frontal, parietal and occipital bossing. Underdeveloped maxilla and paranasal sinuses result in maxillary micrognathia. The mandible is not involved, however, the maxillary hypoplasia gives the mandible a relative prognathic appearance. The syndrome is notable for aplasia or hypoplasia of the clavicles. Some patients have hypermobility of shoulders and can bring them forward until they meet in the midline. The neck appears long and narrow, and the shoulders markedly drooped. Oral manifestations exhibit a high narrow arched palate. Often there may be a true or submucosal cleft palate. Crowding of teeth is produced by retention of deciduous teeth, delayed eruption of permanent teeth, and the presence of a large number of unerupted supernumerary teeth. The supernumerary teeth are mainly found in the premolar and molar regions. The affected individuals do not need any treatment for the syndrome and often enjoy normal life.

Fig. 18-5 Cleidocranial dysostosis. In the absence of clavicles, the patient can bring the shoulders forward towards the midline. Note the underdeveloped maxilla.

Fig. 18-6 Cleidocranial dysostosis. Postero-anterior skull radiograph shows delayed closure of sutures and fontanelles, and presence of multiple wormian bones. Multiple supernumerary teeth are present.

CRANIOFACIAL DYSOSTOSIS (Crouzon's disease)

Craniofacial dysostosis, also known as Crouzon's disease, is an inherited anomaly characterized by a variety of cranial deformities. In this autosomal dominant disorder, the skull reveals early closure of all cranial sutures. Premature closure of these sutures (craniosynostosis) can initiate changes in the brain secondary to increased intracranial pressure. The resulting increased intracranial pressure produces malformation of the skull, including frontal bossing. Facial malformation consists of hypoplasia of the maxilla with a relative mandibular prognathism and a high arched palate. The maxillary hypoplasia produces the characteristic facies often described as "frog-like". The eyes are set wide apart (hypertelorism) and protrude (exophthalmos) with divergent strabismus. The deformities of the cranial bones and orbital cavities are the result of the fusion of sutures and increased intracranial pressure. The life expectancy for the patients is, in general, normal. Many have progressive visual impairment and a few may have mental retardation. Surgical intervention may be necessary to relieve intracranial pressure. Radiographic examination of the skull reveals the absence of cranial sutures and the presence of multiple radiolucent cranial markings (digital impressions) covering the inner surface of the cranial vault.

Fig. 18-7 Cruzon's disease (craniofacial dysostosis). Facial malformation shows hypoplasia of maxilla with mandibular prognathism. Eyes exhibit hypertelorism, exophthalmos and divergent strabismus. (Courtesy, Dr. William Ledoux)

Fig. 18-8 Cruzon's disease (craniofacial dysostosis). Lateral skull radiograph shows early closure of all cranial sutures. Note the prominent digital markings. (Courtesy, Dr. William Ledoux)

MANDIBULOFACIAL DYSOSTOSIS (TREACHER COLLINS SYNDROME)

Mandibulofacial dysostosis, also known as Treacher Collins syndrome, is a rare hereditary developmental anomaly which is transmitted by an autosomal dominant mode of inheritance; some are spontaneous mutations. In the fully expressed syndrome, the facial appearance is characteristic and is often described as "bird-like" or "fish-like". The syndrome is characterized by hypoplasia of the facial bones, particularly the zygomatic bone and the mandible. The underdeveloped zygomaticomaxillary complex leads to a clinically severe midface deficiency. The external ears are malformed (deformed pinnas) and sometimes the middle and inner ears are also affected, resulting in partial or complete deafness. A striking feature is the antimongoloid obliquity or downward slanting of the palpebral fissures and notched or linear colobomas of the outer third of the lower eyelids. Macrostomia may be present. The palate is usually high and sometimes clefted. Dental malocclusions are very common. On radiographic examination, the zygomatic bone (cheek) often appears to be underdeveloped or completely absent. There is also underdevelopment of the paranasal sinuses. The mastoid bone is often sclerotic, that is, the mastoid air cells are absent or reduced. The life expectancy of these patients is normal. Some patients may need ear surgery and hearing aids.

Fig. 18-9 Treacher Collins syndrome (mandibulofacial dysostosis). Note the characteristic facial appearance: downward slanting of palpebral fissures, colobomas of outer third of lower eyelids, depressed cheek bones, receding chin, and a nose that appears relatively large.

ACHONDROPLASIA

Achondroplasia is a hereditary condition (autosomal dominant) producing disturbance in the formation and development of bones ossified in cartilage, that is, the normal process of cartilaginous bone growth is retarded and ceases prematurely. As a result, the bones derived from cartilage are short but of normal width. Ossification of bones derived from membrane (intramembranous ossification) proceeds normally. Thus, the skull vault (derived from membrane) seems large in proportion to the base (derived from cartilage) and the vertebral column long in proportion to the limbs. The lack of growth at the epiphyses of long bones produces extremities which are short in comparison with the torso.

An achondroplastic dwarf is quite short, with short muscular extremities, brachycephalic skull and bowed legs. The elbows cannot often be straightened because the joints exhibit limitation of motion. They are endowed with normal intelligence, unusual strength and agility. Radiographs of the skull show that the maxilla is retruded because of the restriction of growth of the base of the skull. The retrusion produces a relative mandibular prognathism. The disparity in size of the two jaws produces obvious malocclusion and a saddle-nose profile. The dentition is usually normal, but may show congenitally missing teeth and disturbance in the shape of those present. Achondroplastic dwarfism must not be mistaken with pituitary dwarfism; in the latter, the size of the limbs is in proportion to the size of the body.

Fig. 18-10 Achondroplasia. Patient shows extremities which are short in comparison with the torso. Achondroplastic dwarfism must not be mistaken with pituitary dwarfism; in the latter, the size of the limbs is in proportion to the size of the torso.

CLEFTS OF THE JAW BONES

Clefts of the jaw bones result from complete or partial failure of the developmental processes to fuse during embryonic life. The Japanese have a higher frequency of cleft palate than do whites and the incidence is lowest in American blacks. The incidence of cleft lip or cleft palate is about 1 in 800 infants that are born. Clefts of the jaws cause severe functional problems of speech, mastication, and deglutition. Cleft lip causes difficulty with suckling whereas cleft palate causes problems with regurgitation of food and liquid into the nose, as well as problems with speech. Clefts can involve the lip, hard palate, soft palate, uvula, alveolar process or all of them. When the alveolar ridge is involved, it is usually in the region of the maxillary lateral incisor. Clefts may be unilateral or bilateral. A combination of cleft lip and palate is the most common of the various clefts. Cleft lip generally occurs at about the sixth to seventh week in utero whereas cleft palate occurs at about the eighth week in embryonic development. Isolated cleft lip is more common and more severe in males whereas isolated cleft palate is more common in females. The occurrence of mandibular lip or jaw clefts are extremely rare. Radiographs are useful in determining the exact site of the cleft, the amount of bone present, and the position of the teeth near the cleft. Both the dentitions, deciduous and permanent, may be affected. Supernumerary, missing and malpositioned teeth are often associated with clefts. Cleft lips are usually repaired surgically at about one month of age. Cleft palates are usually repaired after 18 months of age, that is, after speech patterns are well established and it is late enough for the growth centers to be disturbed.

Fig. 18-11 Cleft of the maxilla situated between the maxillary lateral incisor and canine.

PIERRE ROBIN SYNDROME

In Pierre Robin syndrome, mandibular retrognathia and hypoplasia is the primary malformation. Along with the severe micrognathia, the other clinical presentations include cleft palate and glossoptosis (posteriorly placed tongue which falls back into the airway). Respiratory and feeding problems are prevalent and may result in episodic airway obstruction, with infant hypoxia and malnutrition. The associated respiratory difficulty, if not recognized, may result in death.

Fig. 18-12 Pierre Robin syndrome. Infant exhibiting severe micrognathia of the mandible. (Courtesy, Dr. Jim Weir)

SICKLE CELL ANEMIA

Sickle cell anemia is an inherited disorder which occurs almost exclusively in black individuals. It is a chronic hemolytic anemia characterized by abnormal hemoglobin, which under low oxygen tension results in sickling of the red blood cells. Thus, the red blood cells have a reduced capacity to carry oxygen to the tissues. They also adhere to the vascular endothelium and clog the capillaries. During a mild attack, the patient complains of weakness, easy fatigability, shortness of breath, muscle and joint pain. Patients may exhibit general signs of anemia and jaundice. During the crisis state there is severe abdominal, muscle and joint pain; high temperature; and may result in circulatory collapse. The disease occurs in children and adolescents. Many patients die of complications before the age of 40 years, although some may have a normal life span. Radiographic appearance is principally caused by hyperplasia of bone marrow of several bones in the body. The skull demonstrates a widening of the diploic spaces and thinning of the inner and outer tables of bone. In some cases there may be a loss of the outer table of bone which may give rise to a "hair-on-end" appearance. Osteoporosis may be observed in the mandible with coarse mandibular trabeculae giving a "stepladder" appearance.

Fig. 18-13 Sickle cell anemia. Lateral skull radiograph shows thicker than normal cranial vault and linear markings of "hair-on-end" appearance.

Fig. 18-14 Sickle cell anemia. Enlarged marrow spaces with the trabeculae giving a "stepladder" appearance.

THALASSEMIA (Cooley's anemia, Mediterranean anemia, Erythroblastic anemia)

Thalassemia, also called Cooley's anemia, is a hereditary chronic anemia which results from abnormal red blood cells having difficulties in hemoglobin synthesis. The red blood cells thus have a reduced hemoglobin content and a shortened life span. Thalassemia was once more prevalent in people living in or originating from the lands around the Mediterranean Sea. This disease is now widely distributed. Radiographic examination shows the expansion of the bone marrow spaces with generalized osteoporosis. The skull exhibits a wide diploe. Sometimes the loss of the outer table of bone may give rise to the characteristic "hair-on-end" appearance of the skull.

Fig. 18-15 Thalassemia. Postero-anterior skull radiograph shows the characteristic "hair-on-end" appearance. (Courtesy, Drs. Jan Hes, Isaïc van derWaal and Kommer de Man)

Fig. 18-16 Thalassemia. Panoramic radiograph shows generalized rarefaction, thinning of cortical bone, and enlarged marrow spaces with thin trabeculation. (Courtesy, Drs. Jan Hes et al, Oral Surg Oral Med Oral Pathol 1990, 69(2):185-190)

HEMIFACIAL HYPOPLASIA

Hemifacial hypoplasia may have its onset at birth or at any time up to the cessation of facial growth. There is progressive failure of growth of the affected side with the result that there is a reduced dimension of the involved side of the face. There may be malocclusion on the affected side but the teeth are of normal size and shape. Radiographic examination, reveals a reduction in the size of the bones on the affected side, especially the condyle, coronoid process and the overall dimension of the body and ramus of the mandible.

Fig. 18-17 Hemifacial hypoplasia. The affected side of the face is smaller than the normal side. Note the crumpled and distorted pinna of the external ear on the involved side. (Courtesy, Dr. William Ledoux)

HEMIFACIAL HYPERTROPHY

Hemifacial hypertrophy begins during youth, sometimes at birth, and usually continues throughout the growing years. It may be characterized by gross body asymmetry or may involve only a specific region of the body. There is progressive growth of half of the face and jaws to unusual proportions. The condition is often associated with other abnormalities including mental deficiency, skin abnormalities (excessive secretions by sebaceous and sweat glands) and compensatory scoliosis. Caniofacial findings include asymmetry of the frontal bone, maxilla, palate, mandible, alveolar process, condyles and associated soft tissue. The pinna of the ear is often remarkably enlarged. Dental malocclusion is common owing to asymmetric growth of the maxilla, mandible, and alveolar process. Radiographic examination of the skulls of these patients reveals enlargement of the bones on the affected side.

Fig.18-18 Hemifacial hypertrophy. Facial asymmetry resulting from progressive growth of half of the face.

GARDNER'S SYNDROME

Gardner's syndrome is an inherited autosomal dominant disorder. This syndrome complex is characterized by skeletal, gastrointestinal, dermatologic, and dental manifestations. The skeletal manifestations include multiple osteomas, especially of the skull and jaws. The gastrointestinal manifestations include multiple intestinal polyps of the colon and rectum but rarely involve the small intestine. The dermatologic manifestations include multiple epidermal cysts and fibromas of the skin. The dental manifestations include multiple impacted supernumerary teeth, failure of permanent teeth to erupt and tendency to the formation of odontomas. Patients afflicted with Gardner's syndrome should be diagnosed and treated in the early stages otherwise, they will develop and die of colonic carcinoma.

Fig. 18-19 Gardener's syndrome. Postero-anterior skull radiograph shows multiple osteomas (arrows). (courtesy Dr. Jim Cade).

PAGET'S DISEASE (Osteitis deformans)

Paget's disease, also called osteitis deformans, is a disease of unknown etiology. The incidence increases in older individuals, especially those over the age of 40 years. Pain in a bone may be mistaken for arthritis. The initial lesion is one of destruction by resorption; later an excessive amount of bone is deposited in a haphazard fashion with a diminution of vascularity of the lesion. The new bone is of poor quality and may result in increased bone fragility and a tendency to fracture. In fact, pathologic fracture is one of the most common complications of Paget's disease. The neurologic symptoms in Paget's disease develop gradually and consist of bone pain, severe headache, deafness, loss of sight, dizziness, facial paralysis, mental disturbance and weakness. The disease may be monostotic or polyostotic. When polyostotic, the bones most prominently affected are those of the axial skeleton which include the skull, vertebral column, extremities and maxilla. There is progressive enlargement of the skull, bowing deformity of long bones and dorsal kyphosis (spinal curvature). The patient develops a waddling gait. The jaws are involved in only 20% of cases; the maxilla is more frequently affected than the mandible (3:1 ratio). In some instances, both jaws are involved. Ultimately the alveolar ridge widens, with a relative flattening of the palatal vault. The enlargement of the maxilla and/or the mandible results in migration, spacing of the teeth and malocclusion. Dentures may have to be remade periodically to accommodate the increase in jaw size. An important diagnostic feature of Paget's disease is that the serum alkaline phosphatase level is increased to extreme limits although serum calcium and phosphate levels are normal. The disease often proceeds with exacerbations and remissions. During remissions the value of alkaline phosphatase is usually normal.

On a radiograph, in the early stages, the density of bone is decreased. As the disease progresses, osteoblastic activity is more than osteoclastic activity; so that apposition

exceeds resorption of bone. The osteoblastic areas appear as patchy radiopacities and give the characteristic "cotton-wool" appearance similar to that of florid osseous dysplasia (chronic diffuse sclerosing osteomyelitis). The lamina dura around the teeth in the involved regions may be absent. The teeth may be hypercementosed only after the bony changes in the jaw are manifested. The most serious complication of Paget's disease is osteogenic sarcoma and occurs in 10% of the patients. On extraction of teeth in an affected part of bone, the wound healing is disturbed and may result in suppurative osteomyelitis. There is no specific treatment for Paget's disease.

Fig. 18-20A Paget's disease. Lateral skull radiograph shows patchy radiopacities giving the characteristic "cotton-wool" appearance.

Fig. 18-20B Paget's disease showing a "cotton-wool" appearance on a postero-anterior projection of the skull.

Fig. 18-21 Paget's disease. Panoramic radiograph shows multiple radiopaque masses producing the characteristic "cotton-wool" appearance similar to that of florid osseous dysplasia.

Fig. 18-22 Paget's disease. Periapical radiographs show patchy radiopacities of the jaws, spacing of teeth, loss of lamina dura and some hypercementosed teeth.

HYPERPARATHYROIDISM

Hyperparathyroidism is an endocrine abnormality in which there is an excess of circulating parathyroid hormone (PTH). The PTH mobilizes calcium from the skeleton and decreases renal tubular reabsorption of phosphate. Primary hyperparathyroidism is caused by a benign tumor or hyperplasia of the parathyroid gland, producing an excess quantity of PTH hormone. In primary hyperparathyroidism, the serum calcium is elevated beyond its normal 9-11 mg % range by resorption of calcium from bones and decreased renal excretion of calcium. The serum phosphorus level is decreased. Secondary hyperparathyroidism is the result of certain types of kidney diseases that cause hypocalcemia, thereby stimulating the parathyroid glands to secrete excess PTH in an attempt to elevate the serum calcium level. The serum calcium is normal to decreased and serum phosphorus is increased.

Hyperparathyroidism is a common cause of generalized rarefaction of the jaws. The skeleton undergoes generalized osteoporosis and is seen on a radiograph as having a ground glass appearance with loss of trabecular bone pattern. In a small percentage of patients there is loss of lamina dura around all the teeth. The lost lamina dura returns after successful treatment of the disorder. Late in the disease, a small number of cases develop central giant cell lesions known as "brown tumors." These "brown tumors" appear radiographically as ill-defined radiolucencies. There is a tendency for the patients to develop renal stones. The serum alkaline phosphatase level is elevated. Increase in serum alkaline phosphatase level occurs in systemic and bone diseases whenever there is significant bone resorption or turnover.

(Note: "Brown tumors" are histologically identical with the central giant cell granuloma of the jaws. For this reason, hyperparathyroidism should always be ruled out in a patient with a giant cell lesion of the jaws).

Fig. 18-23 Hyperparathyroidism. Panoramic radiograph shows generalized disappearance of lamina dura and reduction in radiographic bone density in both jaws.

Fig. 18-24 Hyperparathyroidism. Osteoporosis of bone is seen on the radiograph as having a ground glass appearance with loss of trabecular bone pattern. Also, there is loss of lamina dura.

Fig. 18-25 Hyperparathyroidism. Central giant cell lesions known as "brown tumors" produce ill-defined radiolucencies and disappearance of lamina dura.

HYPOPARATHYROIDISM

Hypoparathyroidism is a relatively rare disease caused by an accidental (or purposeful) removal of the parathyroid glands during surgery. The clinical syndrome is characterized by a reduction in the secretion of the parathyroid hormone. There is decreased renal tubular reabsorption of calcium and increased renal tubular reabsorption of phosphate. The resulting hypocalcemia leads to increased neuromuscular excitability with tetany. Tetany is manifested in the form of carpopedal spasm. When hypoparathyroidism occurs in children, it may cause hypoplasia of enamel and dentin, underdevelopment of the roots of forming teeth, and delayed eruption. If not treated, hypoparathyroidism may cause laryngospasm and death.

HYPERPITUITARISM

In hyperpituitarism there is increased production of growth hormone, causing an overgrowth of all tissues in the body which are still capable of growth. If hyperpituitarism occurs in childhood, the result is gigantism; if it occurs in adulthood, it results in acromegaly.

In gigantism, there is relatively uniform overgrowth of soft tissues and bone, producing a fairly well-proportioned but abnormally large individual with heights of 7 feet or more. Sexual maturity is at a young age; however, impotency occurs at an early age. Radiographic examination reveals normal size teeth which are widely spaced because of increased jaw size. The posterior teeth may be hypercementosed due to functional and structural demands. The trabecular pattern is normal. A lateral view radiograph of the skull shows an enlarged cranium, an enlarged sella turcica, a prominent frontal sinus, and a prognathic mandible.

In acromegaly (hyperpituitarism of adult onset), there is no further increase in height because the adult bones are incapable of increased growth because the epiphyses of bones are fused with their shafts. The sutures of the craniofacial bones are fused. However, there is subperiosteal deposition of bone, resulting in increased thickness of arms and legs. The main feature is the enlargement of the mandible, producing a Class III skeletal malocclusion. An increase in dental arch length does not occur (as in gigantism); however, anterior teeth may be spaced because of the pressure effects of macroglossia. The teeth are of normal size. The posterior teeth may be hypercementosed due to functional and structural demands. The trabecular pattern is normal. The nose, lips, tongue, and soft tissues of the hands and feet

overgrow, sometimes to an abnormally large size. A lateral view radiograph demonstrates an enlarged mandible with a prognathic bite, enlarged sella turcica, frontal sinus and occipital prominence.

Fig. 18-26 Hyperpituitarism producing acromegaly. The main feature is the enlargement of the mandible, producing a Class III skeletal malocclusion.

HYPOPITUITARISM

In hypopituitarism there is reduced secretion of pituitary hormones. It results in a condition called "pituitary dwarfism" in which the person is of small stature and proportionate in body.

Mental development is normal. Jaw growth is retarded, resulting in a small vertical dimension and a small arch length. The teeth are of normal size. Crowding of teeth and malocclusion results because of disproportion in the sizes of the jaws and teeth. Radiographs of extremities show marked retardation of osseous development. Periapical radiographs show retention of deciduous teeth, delayed eruption of permanent teeth, and crowding of the dentition. A lateral skull radiograph shows small hypophyseal fossa and small sinuses incompletely pneumatized.

Fig. 18-27 Pituitary dwarfism resulting from hypopituitarism. The individual is of small stature and proportionate body. Pituitary dwarfism must not be mistaken with achondroplastic dwarfism; in the latter, the extremities are short in comparison with the torso.

HYPERTHYROIDISM (Graves' disease, Exophthalmic goiter, Thyrotoxicosis)

In hyperthyroidism, there is excessive secretion of thyroxin by the thyroid gland. This excess thyroxin causes a generalized increase in the metabolic rate of all body tissues and results in tachycardia, nervous irritability, rapid pulse, fatigability, muscle weakness, heat intolerance and emaciation. There is protrusion of the eyeballs (exophthalmia) and enlargement of the thyroid gland (goiter). Radiographic findings include early eruption of permanent teeth after early exfoliation of deciduous teeth. Infants born to mothers with this condition may have several teeth at birth. Adults may show a generalized osteoporosis.

Most of the cases can eventually be controlled with treatment consisting either of administration of thyroid-suppressive drugs (or radioactive iodine) or of surgical removal of part of the thyroid gland. Use of certain drugs such as epinephrine and atropine is contraindicated because they may precipitate a potentially fatal complication called a "thyroid storm".

Fig. 18-28 Hyperthyroidism. Enlargement of the thyroid gland in a patient with hyperthyroidism. (Courtesy Dr. Jim Cade).

HYPOTHYROIDISM

In hypothyroidism, there is deficient secretion of thyroxin by the thyroid gland. The congenital form is called cretinism and is marked by retarded physical, sexual, and mental development. The individual will have short, fat, puffy features, sparse hair, delayed fusion of all bone epiphyses and fontanelles, and an extremely large tongue causing separation of teeth. There is delayed eruption of teeth. The paranasal air sinuses show partial pneumatization.

The acquired form of hypothyroidism (in adults) is called myxedema and is marked by facial changes resulting in swollen lips and a thickened nose but no dental or skeletal changes (unlike cretinism). The symptoms in myxedema are lethargy, constipation, and cold intolerance. Treatment consists of exogenous replacement of thyroid hormone. The prognosis is good with treatment although finding the proper dosage of thyroxine often requires a trial and error approach. Without treatment, the disease is fatal.

Fig. 18-29 Cretinism resulting from hypothyroidism. The individual has short, fat, puffy features, and an extremely large tongue causing separation of teeth.

DIABETES MELLITUS

Diabetes mellitus results from an absence or decreased production of hormone insulin by the pancreas in response to elevated blood glucose. Predisposing factors include genetics, obesity, increasing age, pregnancy and stress. The condition is characterized by hyperglycemia, glycosuria, polyuria and polydipsia. Diabetes mellitus in itself does not cause periodontal disease; however, diabetic patients tend to have an increased incidence and severity of periodontal disease. There may be generalized loss of alveolar bone and a wide destruction of the lamina dura. Treatment includes a diet of low calories and low carbohydrates, hypoglycemic agents (which stimulate insulin production), and insulin injections (for severe cases). Death is usually due to myocardial infarction, renal failure, stroke or infection.

Fig. 18-30 Diabetes mellitus. Uncontrolled diabetes mellitus shows loss of alveolar bone.

CUSHING'S SYNDROME

Cushing's syndrome is caused by hypersecretion of adrenal cortical hormones by the adrenal glands. Clinical signs and symptoms include central obesity, "moon face" (puffiness of face), "buffalo hump" (adipose tissue above the upper portion of the trunk), altered hair distribution (masculinizing effects in females and in male children), wasting of extremities, hypertension, and a tendency to develop diabetes mellitus. On a radiograph, Cushing's syndrome exhibits generalized osteoporosis due to excess cortical activity. The osseous demineralization may lead to pathologic fractures. Patients undergoing exogenous steroid therapy should discontinue or reduce the dosage to cure the disease. When the cause is a cortisone secreting tumor, it should be removed.

RICKETS AND OSTEOMALACIA

Rickets and osteomalacia result from a failure of new bone to properly calcify. The person has a nutritional deficiency of vitamin D combined with a lack of exposure to ultraviolet light.

Rickets occurs in infants and children, whereas osteomalacia occurs in adults. In rickets, poor bone calcification results in bowing of long bones and increased widening of their epiphyses. Greenstick fractures may occur in these bones. Enamel hypoplasia is fairly common when rickets occurs before the age of 3 years. The teeth may show retarded apical closure and abnormally large pulp chambers. In osteomalacia the teeth are not affected because they are fully developed before the onset of the disease. Metastatic calcification occurs in many tissues, for example, renal stones.

In osteomalacia, the pain is localized in the bones, however, back pain is not as common as in osteoporosis. Spontaneous bone fractures may result. Radiographic changes are similar to those of osteoporosis and include generalized rarefaction and cortical thinning of bones. The lamina dura may be less prominent or completely absent. Treatment includes increased dosage of vitamin D and calcium supplements.

Fig. 18-31 Rickets. Periapical radiograph shows rarefaction of bone and disappearance of lamina dura.

Fig. 18-32 Osteomalacia. Panoramic radiograph shows osteoporosis of bone and disappearance of lamina dura.

HYPOPHOSPHATASIA

Hypophosphatasia is an hereditary disorder in which there is a deficiency of serum alkaline phosphatase. The clinical features include enlarged pulp chambers of deciduous teeth, alveolar bone loss with a predisposition for the anterior regions of the jaws, hypoplastic enamel, and hypoplasia or aplasia of cementum over the root surface.

Fig. 18-33 Hypophosphatasia. Teeth show thin enamel, thin root dentin, thin cementum, and large pulp chambers.

OSTEOPOROSIS

Osteoporosis is a generalized rarefaction of bone without any change in size. The decreased mass is the result of increased porosity of compact bone and loss of trabeculae in cancellous bone. It is the most common form of metabolic bone disorder and is probably the most common cause of backache in elderly persons. Osteoporosis can occur from a number of causes, however, there is a greater tendency for it to occur in old age, postmenopausal women, Cushing's syndrome, patients receiving cortisone therapy, and hyperthyroidism. The bones are more prone to fracture. On a radiograph, osteoporosis is evident as a rarefaction of maxilla and mandible. The cortical borders of bone and anatomic cavities such as nasal fossa and maxillary sinus are thinner and less dense. Individual trabeculae are thin and fine. There is a reduction in the overall quantity of trabeculae in the cancellous bone. Treatment includes estrogen therapy, calcium supplements, protein, vitamin D, and fluoride.

Fig. 18-34 Osteoporosis. Panoramic radiograph of an elderly female shows a reduction in the overall quantity of trabeculae in the cancellous bone. The cortical bone is thin and less dense.

SCLERODERMA

Scleroderma is a systemic autoimmune disease of collagen. It is a generalized connective tissue disease that produces hardening and sclerosis of the skin. The age of onset of this rare disease is in the young to middle-aged adults. It is more common in females than in males (10:1). There is progressive fibrosis of all organs like the gastrointestinal tract, heart, lungs and kidneys, and may result in serious complications and death. The facial skin and oral mucosa are rigid and the patient may be able to manage only limited mouth opening. There is marked thickening of the periodontal ligament spaces, especially around the posterior teeth. This periodontal ligament space thickening is about double the normal width. Another striking finding in 25% of scleroderma patients is the resorption of the mandibular condyles and coronoid processes. Treatment consists of immunosuppressants such as corticosteroids. The disease is debilitating but rarely fatal except over a long period of time. Death may occur due to progressive fibrosis of vital organ.

Fig. 18-35 Scleroderma. Radiographs show generalized abnormal width of periodontal membrane space.

INFANTILE CORTICAL HYPEROSTOSIS (Caffey's disease)

Infantile cortical hyperostosis is a disease of unknown etiology. It is characterized by unusual cortical thickening of bones. The age of onset is usually in the first three months of life but may not be diagnosed until the age of 2 or 3 years. The mandible, clavicles, pelvis and extremities are most commonly affected. Soft tissue swellings occur over areas where bones will later be thickened. The symptoms are those of localized swelling, malocclusion with jaw involvement, and increased alkaline phosphatase. The radiographic appearance is that of a laminated outer surface of bone caused by deposition of new bone under the periosteum. Most cases are self-limiting and regress without treatment within few months or few years. Rare cases produce permanent deformity.

Fig. 18-36 Infantile cortical hyperostosis. Lateral jaw radiograph shows the subperiosteal deposition of bone at the lower border of the mandible (arrow), giving it an onionskin appearance. (Courtesy Drs. Wuehrmann and Manson-Hing).

GENERALIZED RAREFACTIONS	LOSS OF LAMINA DURA
Osteoporosis	Hyperparathyroidism
Hyperparathyroidism	Paget's disease
Osteomalacia	Osteomalacia
Hyperthyroidism	Fibrous dysplasia
Cushing's syndrome	Cushing's syndrome
Prolonged cortisone therapy	
Sickle cell anemia	
Thalassemia	
Leukemia	

SERUM VALUES			
Disorder	Calcium	Phosphorus	Alkaline Phosphatase
Osteoporosis	Normal	Normal	Normal
Primary Hyperparathyroidism	Increased	Decreased	Increased
Secondary Hyperparathyroidism	Normal to Decreased	Increased	Increased
Hypoparathyroidism	Decreased	Increased	Normal
Osteomalacia	Decreased	Decreased	Increased
Paget's disease	Normal	Normal	Increased
Multiple myeloma	Normal to Increased	Normal	Increased