

11. Bones / Orthopedics

Bone structure

Radiographic findings in children with Progeria

Osteoarthritis of the hip



Children with Progeria face many problems with bone growth and development. Skeletal abnormalities can sometimes be seen at birth, but often develop as the children age.

Bone structure

Children with Progeria have smaller bones compared to their age-matched peers, but their bone mineral density is usually mildly low to low-normal after accounting for differences in bone size. However, because the bones are smaller, they are relatively weaker than age-matched children without Progeria. Spontaneous bone fractures are unusual and children with Progeria do not appear to suffer from broken bones any more frequently than children without Progeria. When fractures do occur, the bones heal appropriately.

In general, weight-bearing activities (i.e., walking, running, jumping) are good for maintaining bone mineral density and should be encouraged. Reasonable care should be taken when playing with larger peers, since friends who weigh more than children Progeria can inadvertently cause an injury during play.

In order to maintain the best possible bone health, it is important that children receive adequate calcium and vitamin D in their diets. The goal for calcium intake is 1000-1200 mg per day (3-4 cups of milk or other calcium-rich foods or beverages). To facilitate the absorption of dietary

Though low bone density may develop, rate of bone fracture is no higher than in the general pediatric population.



Child on the DXA scanner; this machine measures bone density and body composition

calcium for proper bone growth, it is recommended that children ingest at least 400 IUD of vitamin D per day. Since it can be difficult to get adequate vitamin D in food alone, supplementary vitamin D (eg., children's multi-vitamin tablet) is recommended by the American Academy of Pediatrics.

> Dual energy x-ray absorptiometer

Yearly bone density measurements by dual energy x-ray absorptiometer (DXA) are recommended to track progress of bone status. Scans of the spine (for density) and whole body provide the most helpful measurements in a child. A whole body scan is particularly helpful because it provides an assessment of body composition in addition to the bone measures. Hip measures are less reliable for bone density, due to the unusual femoral bone findings in Progeria. DXA is available at most hospitals. For accuracy, adjust bone density Z scores for small size. The Z scores that are automatically generated are for larger age-matched children and will appear deceptively low, often in the osteoporotic range. When adjusted for size (i.e., using height-age), Z scores increase, usually to the osteophytic or even the normal range.

> Quantitative computed tomography

Quantitative computed tomography (QCT) may be performed to assess bone structural geometry to assess fracture risk. QCT is not available in many hospitals, but is a three-dimensional analysis of bone structure that can aid in assessing bone status regardless of bone size. There is little pediatric normal control data in the literature at present, so following changes over time (i.e., annually) for a particular child is most helpful to assess status.

Radiographic findings in children with Progeria

> Abnormal findings

- Acroosteolysis (resorption of bone at distal pharynx) is found as early as infancy, but it is observed in all children in later years; it becomes progressively severe with increasing age; it is not usually found in every finger; externally, the fingertips become bulbous; there are no painful sequences associated with acroscleriosis
- Maldevelopment of the mandible; the mandible is small with an increased obtuse angle to its shape
- Clavicular resorption
- Thinning and tapering of ribs
- The thorax develops a pyramidal configuration with the ribs having a “drooped” appearance resulting in narrowing at the apex
- Coxa valga (femoral head-neck axis in excess of 125 degrees) leads to a “horseriding” stance and a wide based gait; it predisposes to hip joint instability and subluxation
- Coxa breva (shortened femoral neck)
- Coxa magna (broadened femoral head)
- Acetabular dysplasia (relatively shallow acetabulum) progressing to hip subluxation resulting in loss of hip joint motion, osteoarthritis, and pain with weight-bearing
- Avascular necrosis of the femoral head
- Long bones: Slender diaphyses, large, broadened epiphyses with atypical demineralization; cortical bone at the diaphysis has normal thickness and mineralization; cancellous bone at the metatheses and epiphyses has decreased mineralization
- Flared proximal humeral metaphysis
- Enlarged capitellum of the distal humerus
- Flaring of the distal femoral metaphysis/epiphysis and the proximal tibial metaphysis/epiphysis

Many x-ray findings develop later in life, so most are not used for diagnosis. The earliest finding is usually acroosteolysis.

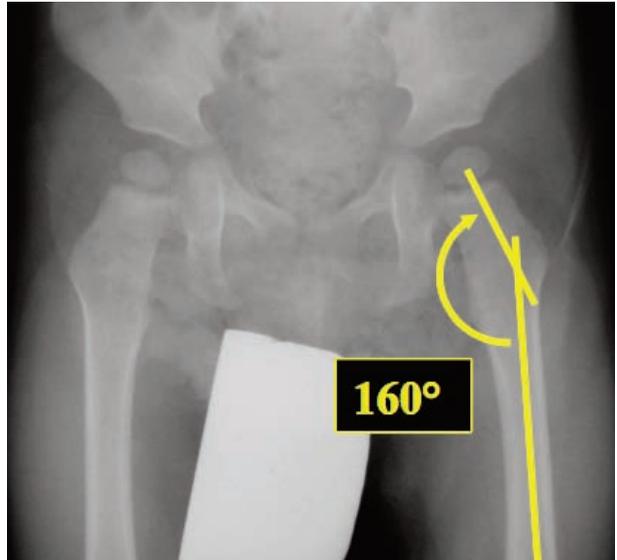
> Normal findings

- Bone age is variable; it can be normal, slightly delayed, or slightly ahead at any chronological age
- Growth plates are normal
- Cranial sutures are usually normal
- Normal configuration of bony pelvis
- Normal joint spaces at the wrists, ankles, knees, and elbows

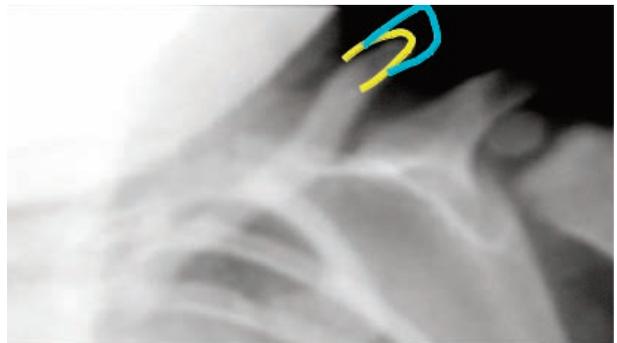
> Radiographic findings



Acroosteolysis



Coxa valga



Clavicular resorption

Osteoarthritis of the hip

Osteoarthritis (OA) is a painful, chronic, incurable, non-inflammatory arthritis that affects diarthrodial joints by progressively breaking down hyaline cartilage. The syndrome is characterized clinically by pain, deformity, and limitation of motion, and pathologically by focal erosive lesions, cartilage destruction, subchondral bony sclerosis, cyst formation, and marginal osteophytes. While many etiologic factors have been postulated, the pathologic changes observed in patients with OA result from some form of mechanical injury. In children with Progeria, OA is likely the result of joint instability from anatomic misalignment and persistent articular surface incongruity related to dysplasia both of the femoral head (coxa magna) and acetabulum. There is a mismatch between the oversized femoral head trying to articulate with the undersized socket resulting in mechanical instability, impingement with range of motion, focal joint space narrowing, and subchondral sclerosis.

MRI using Thor or Gadolinium can be used to diagnose the earliest changes of osteoarthritis before irreversible changes are evident radio-graphically.

Treatment for osteoarthritis can help relieve pain and stiffness, but cartilage degradation may continue to progress. Initial treatment includes physical therapy to restore range of motion, muscle strength, and anti-inflammatory medications to relieve pain. To facilitate ambulation, children with advanced hip OA may require augmentative supports such as walkers. When children are unable to ambulate independently, they often require a wheelchair. As arthritic changes progress, surgical alternatives to reconstruct the involved joint to create a stable, congruent joint may be considered. However, there is little experience with these surgical interventions in children with Progeria. It is important to consider associated risk (i.e., complicated intubation, anesthesia) and medical conditions (i.e., cardiovascular disease) when considering these or any procedures in this high risk population.

Though most children eventually have radiographic evidence of OA, only a minority develop persistent, significant pain or permanent subluxation within their lifespans.