Genu Valgum Associated With Short Stature

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History

A 7-year, 9-month-old Hispanic female presented for evaluation of knock knees and short stature. The knock knee deformity was noted when the child started walking and appeared to progress over the two years prior to presentation. There was an associated weight gain which was thought to be an important contributing factor. Because of the impression that the child was also short in stature, radiographs and metabolic workup were performed, the radiographs suggesting rickets. However, thyroid function tests were normal, and serum phosphorus was reported as low normal, with a normal serum calcium, and a high normal serum alkaline phosphatase.

The patient was adopted, and hence the family history was unknown. She had attained her developmental milestones at the normal time, and had been completely healthy through her infancy and childhood. Physical examination showed bilateral genu valgum measuring 25° valgus deformity clinically (Fig. 1). The remainder of the orthopedic examination was unremarkable, including range of motion of the hips, knees, and ankles, with no deformity noted in the upper extremities or spine. The facies was felt to be normal, and the patient's neurological examination was completely normal, including deep tendon reflexes, motor strength, and gait. Repeat radiographs demonstrated metaphyseal changes consistent with rickets (Fig. 2), and repeat serum chemistries showed a calcium of 4.8 meq/l, phosphorus 2.9 mg/l, alkaline phosphatase 160 units (normal 38 to 138), and a completely normal urinalysis. The patient's height was 115 cm (below the fifth percentile), and weight was 33 kg (90th to 95th percentile). Because of the normal serum chemistries, and the completely normal physical examination other than short stature, the lower extremity deformity was treated with a night splint comprised of long leg polypropylene splints with a rigid spacer between the knees, combined with a Dennis-Browne bar with sufficiently small intramalleolar distance to produce a varus moment at the knee. No medical therapy was instituted.

In follow up 2.5 years later, there was essentially no change in the valgus alignment (Fig. 3). There had been no treatment for the rachitic changes, and there had been no progression of the disease clinically. The patient's skeletal age at this time of 8 years, 6 months was a full 2 years behind her chronological age of 10 years, 6 months. Because of the lack of improvement in the genu valgum, stapling of the medial distal femoral epiphysis bilaterally was performed (Fig. 4), anticipating staple removal when correction had been achieved. Follow up radiographs 16 months later demonstrated mild overcorrection into varus, particularly on the left (Fig. 5), and the staples were removed. Biopsy of the proximal fibular physis was performed at this time for diagnosis, with histological evidence of "metaphyseal dysostosis" being found.

The patient was followed after staple removal, with increasing varus being noted on the left side, both clinically and radiographically. By age 15, radiographs demonstrated 8° varus on the left (Fig. 6), and the patient complained of pain on the medial side of the left knee on a regular basis. At this time, height
was 144 cm (markedly below the fifth percentile), and weight was 50 kg (25th percentile), at skeletal maturity. Arthroscopic examination of the left knee demonstrated chondromalacia of the medial femoral condyle in the weight bearing area, with a pristine lateral compartment. The patient underwent high tibial osteotomy to correct her varus deformity (Fig. 7), with complete resolution of symptoms and return to normal activity 6 months postoperatively.

Discussion

Metaphyseal chondrodysplasia, formerly known as metaphyseal dysostosis, is one of the more common skeletal dysplasias presenting in childhood, because of the frequent orthopedic deformity of non-resolving angular deformities of the lower extremities associated with short stature. The group of disorders known as metaphyseal dysostosis originally was described by Jansen in 1934, and has subsequently been subclassified into several types, depending on severity of involvement and other associated systemic conditions.1-5. Jansen's original report described patients with severe short stature and severe radiographic metaphyseal changes reminiscent of classic rickets, showing significant longitudinal widening of the physeal plate, which may be

Fig. 1: Clinical appearance, age 8.

Fig. 2A: Standing AP radiograph at presentation. Note the "widening" of the physis, especially of the distal femur. There is no cortical osteopenia, however.
best described as “vacuolated” streaking. In 1949, Schmid reported a milder variety of a short-statured patient with the same radiographic findings typical of rickets, but with a milder degree of shortness, and much less skeletal involvement. Schmid-type metaphyseal chondrodysplasia is characterized by an adult height of 130 cm to 160 cm, an autosomal dominant inheritance, and anomalies basically limited to the lower extremities, with the most common being genu varum or valgum, and coxa vara in many patients. Intelligence is normal, facies is normal, and skull and spine radiographs are normal, with no other associated systemic anomalies. Other than the rachitic radiographic findings, these patients are otherwise healthy and normal, as exemplified by the patient reported here.

The Schmid-type chondrodysplasia is reported to have a prevalence of three to six per million, and as suggested in the history, the major differential diagnostic problem is to separate this skeletal dysplasia from metabolic or renal rickets. Since all serum chemistries are normal, the workup of such patients can usually cease once the basic screening chemistries have been determined. Bone maturation is normal, with the rachitic changes seen radiographically disappearing in the adult, and there is no osteopenia associated with the metaphyseal changes as is usually present in classic rickets (Fig. 2, 3). Differential
diagnosis from other types of metaphyseal chondrodysplasia (Jansen, McKusick, etc) can usually be made on the basis of the inheritance pattern, the severity of shortening and metaphyseal radiographic changes, and the presence of other systemic involvement, such as hair involvement, or pancreatic insufficiency.3 5 7

Histopathological study of metaphyseal chondrodysplasia shows a disorder of chondrocyte maturation.8 There is a failure of the orderly alignment of the chondrocytes in the physis; instead of orderly columns, the chondrocytes are clumped in clusters, and are surrounded by a dense collagen matrix, which may prevent the chondrocytes from aligning in the proper longitudinal orientation. Ossification appears to be normal with normal vascular invasion into the hypertrophic zone of the physis, but there appears to be a paucity of vascular invasion, with resultant continued migration of clusters of hypertrophic chondrocytes into the metaphysis. This probably explains the radiographic widening of the physeal plate, and the vacuolated streaking of the metaphysis in the more severe Jansen-type of chondrodysplasia. Electron microscopic studies suggest a possible storage problem in the involved chondrocytes,9 but the etiology of metaphyseal chondrodysplasia is essentially unknown, other than to refer to it as a rickets-type syndrome with no biochemical abnormalities identified.

As noted above, the usual complaint at presentation is one of non-resolving varus or valgus deformity at the knee, or a waddling gait, due to the presence of coxa vara. Depending on the age of the child, short stature may or may not be appreciated. Because of dominant inheritance, family history will be extremely helpful in the initial evaluation. In this particular case, family history was not obtainable,
but with the repeatedly normal serum chemistries and the entirely negative review of systems, the diagnosis of rickets was never seriously entertained, and no metabolic treatment for the radiographic changes was undertaken.

Spontaneous improvement of angular deformity, particularly genu varum, has been reported, but the natural history of varus or valgus deformities at the knee in metaphyseal chondrodysplasia is far from agreed upon. Most authors have described bracing as an initial treatment, similar to the treatment of infantile tibia vara. Surgical correction by either distal femoral osteotomy or stapling of the epiphyseal plate on the medial side of the distal femoral epiphysis has been recommended. In addition, correction of coxa vara and arrest of the trochanteric apophysis to prevent trochanteric overgrowth has been described. Indications for any of these procedures are not well delineated. Although such deformities of hip and knee may usually be corrected purely on the merits of deformity or symptoms, there is no evidence that either coxa vara or residual angular deformity at the knee in adult chondrodysplastics is symptomatic, as many reports describe asymptomatic adults with various degrees of residual coxa vara, trochanteric overgrowth, and mild varus deformity at the knee (Schmid-type involvement).

The decision to treat this patient’s genu valgum was based on functional, as well as cosmetic difficulties related to significant, non-resolving valgus deformity in a patient whose legs were not expected to get much longer, and hence improve with growth. There are well documented symptoms in adolescents with this angular profile of knee discomfort, mechanical rubbing of the inner thigh and knee during gait, and foot and ankle symptoms associated with the mechanical axis of the extremity falling medial to the ankle and foot. The selection of epiphyseal stapling as a simple treatment for this problem is often cited in the literature, but critical appraisal of this patient’s treatment would suggest that the stapling was carried out at too early a skeletal age. Epiphyseal stapling must be considered, for all practical purposes, to be a form of permanent epiphyseal arrest, and the intention to remove staples after correction is obtained with the expectation of resumed growth is unpredictable. Because this patient underwent stapling at a skeletal age of around 8 years, 6 months, the eventual overcorrection into varus by persistent lateral physeal growth in the presence of an effectively arrested medial physis in the distal femur cannot be regarded as unexpected.

In the hands of its inventor, stapling of an epiphysis seemed an ideal method of temporary growth arrest, with few complications and minimal surgery. Subsequent experience with the technique to obtain growth arrest for equalization of limb lengths revealed a high incidence of complications, including asymmetric growth arrest producing deformity, ineffective growth arrest due to staple extrusion, and a host of other problems. Because of the high incidence of complications, formal bony epiphysiodesis is the recommended method to obtain complete growth arrest for the purpose of equalization of limb lengths. However, for the correction of angular deformity, especially genu valgum, epiphyseal stapling has enjoyed significant, though only sparsely reported, success, and is often recommended. The major problem, according to
Fig. 6: AP radiograph, age 15.

Fig. 7: AP radiograph, status post left high tibial osteotomy.

Blount, is the amount of time staples may be left in situ with the assurance of resumption of growth after removal.\textsuperscript{12} It was originally suggested that the interval be limited to 2 years, though most investigators have found that correction is obtained long before this time, with the staples being removed at the time of correction or slight overcorrection.\textsuperscript{16,17} The other major clinical problem is the age of the patient at which time stapling is most efficacious. For idiopathic adolescent genu valgum, the usual time for stapling is between 11 and 14 years chronological age.\textsuperscript{16,17} Using the Green-Anderson growth tables, Howarth recommends stapling be done according to the skeletal age, and using calculations based on the ratio of leg length to epiphyseal plate width, estimates how many inches of intramalleolar distance can be corrected by stapling.\textsuperscript{17} There is obviously an implicit inaccuracy in the measurement and control of angular correction using measurements based on
distance between the medial malleoli. Additionally, several months may be required before effective epiphysial growth arrest occurs following stapling, because of the need of some longitudinal growth at the physeal plate before the arms of the staples can exert a compressive, growth-retarding pressure on the epiphyseal plate. Hence, the timing of epiphyseal stapling is subject to an unpredictable lag time before inhibition takes effect.

Hence, there is no accurate method to determine how much correction can be obtained by stapling due to variables of skeletal age, rapidity of onset of growth inhibition, and the rate of growth resumption following staple removal. As previously noted, stapling should probably be considered a method of permanent growth arrest, because of the uncertainty of resumption following staple removal. In this patient, removal of the staples at the time of slight radiographic overcorrection, the recommended time for removal, did not result in resumption of effective growth, with gradual increase in a varus deformity, which over time became symptomatic by the production of medial compartment chondromalacia. Although staple removal has been reported to be a benign procedure, some consideration must also be given to possible surgical damage to the physeal plate during removal, which would lead to complete arrest, and resumption of growth on the previously stapled physis would not be observed. Hence, this patient's varus deformity at maturity required correction by high tibial osteotomy, with resolution of symptoms over the short term, presumably by relieving excessive medial compartment joint reaction force which had led to the cartilage damage observed arthroscopically.

The final outcome will, of course, require long-term follow up into adulthood, but, because of the otherwise normal joints in Schmid-type chondrodysplasia, this patient's articular cartilage should behave in a normal manner, i.e., the mechanical axis at the knee should be maintained in neither excessive varus nor excessive valgus, but should be corrected to as close to a neutral mechanical axis as possible to avoid late degenerative arthritis.

References


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