Heterotopic ossification after Patellar Tendon Repair presenting in Trisomy 8 Mosaicism: a case report and literature review

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ABSTRACT
Heterotopic ossification is the abnormal formation of lamellar bone in soft tissue. Its presence jeopardizes functional outcome, impairs rehabilitation and increases costs due to subsequent surgical interventions. We present a case of a 32-year-old male with trisomy 8 mosaicism who developed severe heterotopic ossification of his right extensor mechanism subsequent to repair of a patellar tendon rupture. To our knowledge there are no prior reports of heterotopic ossification as a complication of patellar tendon repair. This case may suggest an association between T8M and increased risk of heterotopic ossification.

INTRODUCTION
Heterotopic ossification (HO) is most commonly associated with musculoskeletal trauma, central nervous system disorders or injuries, severe burns, and elective surgery such as total hip arthroplasty (6). The clinical signs of HO include increased joint stiffness, limited range of motion, warmth, swelling, and erythema. Although its etiology is still unclear, important contributing factors include
hypercalcemia, tissue hypoxia, alterations in sympathetic nerve activity, prolonged immobilization, and imbalance between parathyroid hormone and calcitonin (9). The over expression of bone morphogenetic proteins (BMPs), among other systemic and local factors, also appears to play an important role in the pathophysiology of HO (9). HO occurs in 3-90% of lower limb joint replacement cases, though only 3-7% is clinically significant based on the Brooker Classification of HO (Grades 3 and 4) (10, 11). HO can also be hereditary; similar to fibrodysplasia ossificans progressiva, progressive osseous heteroplasia, and Albright’s hereditary osteodystrophy (10).

Complete somatic trisomy 8 is rarely compatible with life and often results in miscarriage (16). Trisomy 8 mosaicism (T8M) on the other hand, is a form of trisomy 8 in which some of the body’s cells have three copies of chromosome 8 while other cells still possess the normal two copies. T8M is an uncommon diagnosis affecting only 1 in every 25,000-50,000 live births. The timing and particular cell lineages in which nondisjunction occurs determine which tissues and cells are affected. Therefore, T8M can present with a wide range of clinical manifestations and extremely variable phenotype (1). Some of the common musculoskeletal features of T8M include joint contractures, long and narrow thorax with wide sloping ribs, hypoplastic glenoid cavities, symmetrical widening of the clavicles, abnormal sternum, narrow pelvis, and hip dysplasia (2-5).

CASE PRESENTATION

Our patient is a 32-year-old male with a history of T8M syndrome documented by chromosomal analyses at an outside hospital. His syndrome is characterized by dysmorphic facial features including saddle nose deformity and a large forehead as well as mild mental retardation. He presented to our clinic with complaints of right knee pain and inability to completely extend his right knee after injuring it several months ago. On examination of his right knee he was able to achieve full extension passively but was unable to actively perform a straight leg raise. On palpation, there was generalized tenderness and a high riding patella with a palpable gap beneath it consistent with a patellar tendon rupture. X-rays revealed marked patella alta with some mild heterotopic ossification in the distal quadriceps musculature. The patient was consented for right patellar tendon repair and possible excision of the HO.

During the operative repair, the patellar tendon was found to be avulsed off the inferior pole of the patella. A repair was accomplished by weaving sutures through the patellar tendon and drill holes in the patella. Postoperatively, the patient was placed in a long-leg cast. The patient was not given any therapy for HO prophylaxis.

Postoperative follow-up visits for the first six weeks revealed no obvious complications with proper wound healing and no complaints from the patient. At six weeks postoperatively, the patient’s cast was removed. Physical therapy was instituted at that time.

Follow-up visits for the next three months demonstrated a decreasing range of motion of the right knee. X-rays taken three months postoperatively revealed extensive HO within the quadriceps muscles as well as the patellar tendon
At four months postoperatively, the patient’s knee was completely fused at 45 degrees. Despite the deteriorating range of motion, plantar and dorsiflexion remained intact. Sensation was intact and there was brisk capillary refill.

At this time the patient was given the option of leaving his knee locked at 45 degrees or performing a second surgery to fuse the knee in more functional position. A total knee arthroplasty was not considered because the patient’s quadriceps mechanism had ossified thereby eliminating active knee extension. After several additional opinions, the patient and his mother decided to proceed with a knee fusion.

A second surgical procedure was undertaken. Compression arthrodesis of the knee was accomplished with an intramedullary interlocking nail from the hip to ankle (Stryker T-2 Fusion Nail System) after the distal femur and proximal tibia were transversely denuded of cartilage and subchondral bone. Images taken after the surgery revealed a successful procedure (Figure 3).

**DISCUSSION**

After delaying treatment for several months for unclear reasons, the patient presented with mild HO on his initial radiographs. The subsequent trauma of the primary surgery to repair his patellar tendon was most likely a catalyst for the extensive additional HO that crippled his right knee mobilization. There are no documented cases of HO secondary to patellar tendon repair. The few reported cases of patellar tendon HO are secondary to medial parapatellar tibial intramedullary nailing (17).

The aggressive nature of this patient’s HO may be attributable to his T8M diagnosis. Chromosome 8 has been linked to certain bone morphogenetic proteins (BMPs). BMPs are part of the transforming growth factor beta (TGF#) superfamily and play an important role in postnatal bone development (12). BMP-1, located at 8p21, may explain the presence of abnormal bone formation in our patient with T8M (13). BMP1 has a unique structure and may play a role in activating other BMPs (13). Extensive research is being conducted to better understand the biochemistry of these proteins.

Basic standards for HO prophylaxis have been relatively well established, but specifics are still debated. Current methods include non-steroidal anti-inflammatory drug (NSAID) treatment with indomethacin or localized radiation therapy. A recent study concluded that indomethacin is the gold standard for HO prophylaxis following total hip arthroplasty and is furthermore the only drug proven to be effective against HO following acetabular surgery (14). Although radiation therapy has been shown to be slightly more costly than NSAIDs, other studies suggest that morbidities and quality of life differences associated with NSAIDs are difficult to quantify, and radiation therapy may remain the preferred prophylaxis of HO after total hip arthroplasty (14, 15).

**CONCLUSION**

It is our opinion that this patient’s T8M status placed him at higher risk for developing HO postoperatively. There are no reports of HO as a complication of patellar tendon rupture or repair. A link between these pathological phenomena
could explain the extensive HO in our patient and allow us to anticipate similar outcomes in T8M patients.

Consent

Written informed consent was obtained from the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal.

Competing interests

I, nor any other authors involved in this case report declare that we have no competing interests.

REFERENCES


Figure legends:
1. A-P x-ray of right knee at the time of patient's presentation. Some heterotopic ossification (HO) is seen superior to the knee.
2. Lateral x-ray 3 months after patellar tendon repair showing marked progression of HO.
3. Lateral x-ray right knee after surgery to accomplish arthrodesis.