Huge Secondary Chondrosarcoma of the Scapula – A Delayed Presentation.

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Abstract: Multiple Hereditary Exostoses (MHE) produces bone deformities, shortened stature, bony growths, limb length discrepancies, and tumors. Forty percent of persons with MHE have a shortened stature due to the disorder, and twenty percent have a knee deformity. Discrepancies in limb length, as well as wrist and hand deformities are also common. The incidence of secondary chondrosarcoma developing in hereditary multiple exostoses is 1-5\% (4). The survival rate depends on the time interval at the time of presentation and the histological type. At times, the tumour can attain a very large size (9). We report such an unusually huge tumour, arising from an exostosis over scapula.

Keywords: Diaphyseal aclasis; chondrosarcoma; scapula

INTRODUCTION

The disorder hereditary multiple osteochondromas (HMO), previously called Hereditary multiple exostoses (HME), is characterized by growths of multiple osteochondromas (benign cartilage-capped bone tumors that grow outward from the metaphyses of long bones). HMO is inherited in an autosomal dominant manner. The two genes in which mutations are known to cause HMO are EXT1 and EXT2. Monitoring of the size of exostoses in adults may aid in early identification of malignant degeneration. The most serious complication of HMO is sarcomatous degeneration of an osteochondroma. Axial sites, such as the pelvis, scapula, ribs, and spine, are more commonly the location of degeneration of osteochondromas to chondrosarcoma [Porter et al 2004]. Rapid growth and increasing pain, especially in a physically mature person, are signs of sarcomatous transformation, a potentially life-threatening condition. The rates of sarcomatous degeneration averages approximately 2\%-5\%.

CASE REPORT

A 30 year old young male patient presented with multiple bony swellings since early childhood. Initially, his parents noticed swelling over left scapula. Subsequently, he developed multiple swellings over right scapula, right thigh & right leg. Since 2 years, the right scapular swelling had started to grow rapidly in size and had become painful. For the past few weeks, there was watery discharge from the swelling at 2-3 sites. There was no history of fever. Sleep was quite disturbed as he could not lie down comfortably supine due to the size of the swelling.

Clinically, his general condition was fair. There was no pallor and vital signs were stable. There were no systemic abnormalities. He had multiple bony swellings of varying sizes over long bones of all four limbs. He had stunted growth with limb deformities. The right scapular bony swelling measured about 30cm.x20 cm.s, bony hard, nontender and with areas of skin breakdown (Figures 1 & 2).
A clinical diagnosis of Hereditary multiple exostoses (Diaphyseal aclasis) with secondary chondrosarcoma was made. X-ray showed features of chondrosarcoma of the scapula (Fig.3). A Ct scan of thorax and right scapula showed features suggestive of diaphyseal aclasis with chondrosarcomatous changes.

Patient was advised to undergo open biopsy to confirm the diagnosis and to know the histological grade & type of tumour. However, patient declined any form of invasive procedures and hence was discharged at request.

**DISCUSSION**

Multiple osteochondromas (MO) is characterized by development of two or more cartilage capped bony outgrowths (osteochondromas) of the long bones. The prevalence is estimated at 1:50,000, and it seems to be higher in males (male-to-female ratio 1.5:1). Osteochondromas develop and increase in size in the first decade of life, ceasing to grow when the growth plates close at puberty. They are pedunculated or sessile (broad base) and can vary widely in size. The number of osteochondromas may vary significantly within and between families, the mean number of locations is 15–18.

Hereditary multiple osteochondromas (HMO) is a rare genetic disorder characterized by multiple benign (noncancerous) bone tumors that are covered by cartilage (osteochondromas), often on the growing end (metaphysis) of the long bones of the legs, arms, and digits. The number of multiple osteochondromas varies from two to hundreds. These osteochondromas usually continue to grow until shortly after puberty and may lead to bone deformities, skeletal abnormalities, short stature, nerve compression and reduced range of motion. Hereditary multiple osteochondromas is inherited as an autosomal dominant genetic condition and is associated with abnormalities (mutations) in the EXT1 or EXT2 gene. These growths vary in size and number among affected individuals. In many cases, no treatment is required. If the exostoses are small, they may have little or no effect on the patient. The risk for development of malignant (cancerous) tumors, mostly chondrosarcomas, is approximately 1 to 5% (3 & 4). Rapid growth and increased pain are signs of a possible malignant change (5). The suspicion of secondary chondrosarcoma is indicated by growth of the tumour after puberty, the presence of pain, or a thickness over 1 cm of the cartilaginous cap in adults (2). Persons who develop malignant chondrosarcomas are usually in their twenties to their fifties.

Recommendations for treatment of chondrosarcomas include the following (6, 7):

- Resectable low-grade and intracompartmental lesions: Wide or intraleisional excision, with or without adjuvant therapy
• Pelvic low-grade tumors, large tumors, and high-grade (II, III), clear cell, or extracompartmental lesions, if resectable: Wide excision (preferred)
• Wide excision to achieve negative surgical margins by either limb-sparing surgery or amputation
• Postoperative proton and/or photon beam radiation therapy for tumors in an unfavorable location (e.g., skull, axial skeleton)
• Consideration of radiation therapy for unresectable tumors

CONCLUSION

The incidence of secondary chondrosarcoma is low in bony exostoses. We report a gigantic sized growth over scapula in a young man, who has been an interesting case from teaching and academic point of view. Sadly, patient did refuse any investigations or treatment, hence could not be followed up.

REFERENCES

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