PATHOLOGICAL FRACTURES; A CONSIDERATION WITH METACHONDROMATOSIS AND DIFFERENTIAL DIAGNOSES

Osteochondromatosis and Gaucher's Disease

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**Abstract:**

**Background:** Metachondromatosis is a condition that causes gross conical metaphyseal expansion (sometimes irregular), cortical thinning, exostoses. Metachondromatous lesions occur mainly in the extremities and are roughly symmetrical. The lesions can involve the bones of the hand and all long bones in the arms and legs. The distribution in this case additionally involved the acromion process and ischia. The bone changes, although dramatic, can be confused with other types of metaphyseal dysplasia such as Gaucher disease and multiple exostoses.

**Objective:** This paper will review the literature with regard to Metachondromatosis, Gaucher disease and Osteochondromatosis due to their similarities. The case study serves as an example of these findings and documents a history of fractures secondary to the obvious bone changes.

**Discussion:** Clinical manifestations of these conditions and how they may present to the manual therapist are discussed. With respect to Metachondromatosis, the manual therapist needs to be mindful of pathological fractures that can occur with little trauma. Manual therapists are cautioned against using long bones as levers for spinal manipulation in these patients.

**Key Words:** Metachondromatosis, Osteochondromatosis, manual therapies, manipulation, chiropractic, contraindications, fractures, case report.

**INTRODUCTION**

Encondromatous lesions, formed from cartilaginous cells, are found in the long bones and flat bones (ribs, ilium etc) of the body. They usually appear near the epiphysis and extend along the shaft, often expanding the cortex, making it thin and susceptible to fracture. The inner surface of the cortex is often scalloped and the lesion is of a mottled radiolucent appearance. There are many conditions that have similar radiographic appearances, including Metachondromatosis, Osteochondromatosis and Gaucher disease. It is worthwhile understanding the similarities and differences between the characteristics of these differentials.

Osteochondromatosis and Metachondromatosis are two conditions that have many similarities. Metachondromatosis, that has had approximately 34 cases reported in the literature, has lesions including exostoses that tend to point towards the joints and enchondromatous lesions. The enchondromatous lesions may appear as metaphyseal streaking, a periarticular flowery appearance or lesions similar to that seen in lytic metastatic disease. The exostoses can spontaneously regress and disappear. The identified location of these lesions include the iliac crests, proximal femur, distal femur, proximal fibula, distal tibia, distal radius, proximal humerus and hands and feet. A common complication is nerve compression that results in mechanical problems secondary to exostotic growth. A scenario often requiring decompression surgery.

Osteochondromatosis is the most common bone tumour occurring in approximately 1-3% of the population. It has the distinguishing feature of having osteochondral bodies, formed from bone and covered by cartilage. They can be located within the articular joint spaces, and are usually associated with one articulation. The osteocartilaginous exostoses can be large enough to result in mechanical impingement of nerves. Malignant degeneration of these lesions is possible and although hereditary factors are present, the lesions have also been induced by radiation therapy. Pathological fractures can occur with the enchondromatous lesions in the metaphysis.

Gaucher disease is an autosomal recessive condition of lysosomal storage, resulting in uptake of lipids in macrophages, seen often in the cortex of bone. An accumulation of gluco-cerebroside occurs in the reticuloendothelial cells, due to a deficiency in the
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activity of glucosylceramide-β-glucosidase (glucocerebrosidase enzyme)\textsuperscript{19}. More uncommonly it is due to a deficiency of saposin C, a heat-stable cofactor required for normal catalytic function of glucocerebrosidase\textsuperscript{19}. The expression of the disease varies in individuals, depending on genetic components that may modify the presentation of the disease. It is possible to have some individuals who are essentially carriers, having the irregularity in genetic material but no symptoms\textsuperscript{13}.

There are three groups of Gaucher disease. Type I, the most common group, occurs at an incidence of 1/40,000-1/60,000 of the population\textsuperscript{20} and makes up between 95-99\% of all Gaucher disease affected people. People of Ashkenazi Jewish decent have an incidence of 100 times the average incidence of this type of Gaucher Disease\textsuperscript{20}. Type I is a chronic non-neuropathic group, the disease having a chronic and fluctuating pattern, from being asymptomatic at times to severe symptoms that include bone crises and pathological fractures\textsuperscript{21}. Type II, an infantile and acute neuropathic group is rare and usually lethal before 2 years of age\textsuperscript{22,24}. Type III, a juvenile and subacute neuropathic group suffer from neurological symptomatology such as convulsions\textsuperscript{16}. It is more common in Norrbottnian people of Sweden\textsuperscript{16,17,19}.

On a cell level in Gaucher disease, the monocytes or more specifically macrophages can be demonstrated to have membrane bound inclusions containing tubule-like precursors to Gaucher cells\textsuperscript{14}. Later, the accumulation of glucocerebrosidase occurs in reticuloendothelial cells\textsuperscript{14,18,24}. These cells tend to group together in the liver, spleen and bone marrow and are commonly referred to as Gaucher cells. Splenectomy is frequently performed in type I Gaucher disease patients\textsuperscript{25,27}.

The symptomatology of type I patients include bone crises (35-83\%)\textsuperscript{16,23}. The symptoms often last about 2 weeks, include severe pain that induces forced bed rest, some local redness, swelling and warmth of the affected area\textsuperscript{23,26}. The pain is thought to be due to localised infarction\textsuperscript{14} and associated with osteonecrosis\textsuperscript{17,18} and pathological fractures\textsuperscript{23,25,28}. Bone crises appear more common in spleenectomised patients\textsuperscript{22,28,29} and may follow episodes of gastroenteritis\textsuperscript{30}. Other possible related conditions include osteomyelitis\textsuperscript{16,24} and premature joint degenerative changes\textsuperscript{16}. It is important to note that these patients may have coagulation factor deficiencies (factor XI)\textsuperscript{19}, menorrhagia\textsuperscript{19} and a higher incidence of spontaneous abortion\textsuperscript{19,20} and post-partum bleeding and infection\textsuperscript{20}. There is a slightly greater chance of Gaucher disease patients developing Parkinson’s disease\textsuperscript{31}.

Clinical features can often be identified by distinct examination findings. Plain film x-ray is most revealing with Erlenmeyer-flask deformity\textsuperscript{17,29}, osteoporosis\textsuperscript{26,32}, thin cortex\textsuperscript{33}, osteosclerosis\textsuperscript{17} and pathological fracture\textsuperscript{15,17,29,30,33,34}, peristeal changes\textsuperscript{16}, flattening of the femoral head and a central area of reabsorption of the femoral head\textsuperscript{16}. Radiographic changes of the femoral neck may precede those of the head by a few months\textsuperscript{36}. The common locations of Gaucher cells in bone include the proximal and distal ends of the femur\textsuperscript{27,33}, proximal\textsuperscript{22,30,33} and distal ends of the tibia\textsuperscript{26}, proximal humerus\textsuperscript{27} and thoracic\textsuperscript{15,17,23,26,29,30,33,34} and lumbar spine vertebral bodies\textsuperscript{35}. Compression fractures of thoracic and lumbar vertebrae are commonly encountered\textsuperscript{21}. MRI, using T1-weighted sequences, reveals Gaucher cell anomalies\textsuperscript{36}. Computer tomography is ideal for defining new fractures but MRI is better for defining the extent of the lesions and checking on healing fractures\textsuperscript{36}. Bone scans using Tc-Sestamibi and Tc-labelled methylene diphonate tracers can show the infiltration of bone marrow\textsuperscript{33,37}. An increased metabolic activity is usually seen\textsuperscript{31}. Unfortunately it is difficult to identify individuals at risk of developing severe clinical skeletal complications in the early stages\textsuperscript{29}.

Many Gaucher disease patients are treated with enzyme replacement therapy (alglucreas)\textsuperscript{19,20,26,28}. It has a plasma half-life of 11 minutes and intracellular half-life of about 8 hours but can cause a decrease in hepatomegaly and splenomegaly and an increase in the patient’s height, weight and sexual maturity\textsuperscript{15}. The enzyme is best taken in low doses 3-7 times per week\textsuperscript{19,20,24}. Enzyme replacement therapy is often used during a bone crisis but may not remove the possibility of suffering pathological fractures\textsuperscript{26}. Vertebral compression fractures may cause cord compression and needs to be treated with decompression surgery using metal braces and screws to ensure stability\textsuperscript{5,28}. Gaucher patients often undertake arthroplasty surgery to replace degenerated or destroyed joints\textsuperscript{27}.

Other therapeutic approaches that have been described in the literature include crutches\textsuperscript{36}, radiation therapy\textsuperscript{35} and medication including prednisone (helps decrease pain by reducing inflammation)\textsuperscript{15,20,26}. Bed-rest is considered to increase osteopenia and therefore is unwise unless fractures of the lower limbs have recently occurred. Chiropractic therapies are diverse and for this article I will consider the suitability of only manual joint manipulation and its suitability for a sufferer of Gaucher disease. There appears to be no information about manual therapies being used on a patient with Gaucher disease.

On review it appears that there is considerably more information written about Gaucher disease than Osteochondromatosis or Metachondromatosis. A clinical presentation of an expansile metaphyseal lesion should alert the clinician to potentially consider all three disorders. Additionally, Ollier’s disease presents with multiple enchondromatous lesions and if there are exostoses to consider, the conditions of multiple exostoses and Langer-Gierdian syndrome need to be considered. Multiple exostoses however do not have the enchondromatous
lesions and Langer-Gierdian syndrome has distinct accompanying features such as mental retardation and facies (bulbous nose and fine sparse hair). Finally, Genochondromatosis should be considered as a differential diagnosis.

Genochondromatosis, even rarer than Metachondromatosis has had less than 10 cases recorded. It presents with symmetrical enchondromatous lesions, swollen metaphyses and a skeletal distribution which includes medial clavicle, proximal humerus, distal femur and proximal tibia. Exostoses do not appear in Genochondromatosis.

Written permission was obtained from the patient to publish her details in this case report prior to publication.

CASE STUDY: CLINICAL PRESENTATION

A 31 year old Caucasian female presented to a Chiropractor complaining of neck and shoulder pain with referral of pain into the thoracic spine. It was concluded at that time that she was experiencing a lower cervical facet joint sprain on the left side with diffuse myofascial pain and subsequent joint referral into the upper thoracic region. It was likely that the complaint was unrelated to her genetic based metaphyseal abnormalities.

She demonstrated a series of slight scoliotic curves on observation, with the left shoulder and iliac crest observed to be slightly higher and left cervicothoracic and left lumbar spine erector spinae more prominent than the right. She had distinct changes in posture from a lateral aspect, with marked anterior weight bearing of the head and extreme hyperlordosis of the lumbar spine and an almost horizontally placed sacrum. Both knees had marked genu valgus (especially right). She had a remarkable habit of straightening her knees, while the hips were flexed, to tie up her shoelaces. She demonstrated limitation of lumbo pelvic flexion but had very flexible hamstring muscles. She had normal facies and no torticollis was present.

Palpable bony prominences or expansions were noted on both wrists (radius and ulnar), hands including both ends of metacarpophalangeal joints and the bases of some proximal phalanges, both proximal humerus, proximal and distal tibia and fibula on both sides and proximal right femur. The function of the spine demonstrated slight limitation of movement in the mid thoracic spine in all ranges of motion. The left shoulder tended to elicit a crack on scapular excursion. On initial presentation, marked tenderness was present over C5-6 left facet joints, limitation and pain was elicited on left rotation, left lateral flexion and extension and there was accompanying left sided paraspinal muscle hypertonicity. These signs resolved quickly over the period of 7-10 days.

Plain film x-rays taken periodically as a child and teenager at the Royal Children’s Hospital in Melbourne, demonstrated diffuse metaphyseal deformity (extending into the diaphysis), cortical thinning (see figure 1) and pathological fractures. The fractures over the years have included the proximal tibia and fibula on the right side (on two occasions), the mid shaft of the right femur, the right proximal humerus (see figure 2) and recently the left ulnar styloid process. The fractures have healed well. She has had an osteotomy of the distal femur on the right side to help correct a valgus deformity and a graft to fuse tarsal bones of the right foot.

Figure 1: Diffuse metaphyseal deformity (extending into the diaphysis), cortical thinning and pathological fractures.
Cervical and thoracic spine plain film x-rays, taken within the last 5 years, were relatively normal. There was a slight to moderate levoscoliosis with its apex in the upper lumbar spine with early degenerative changes noted. There was generalised deformity of the bony pelvis including acetabula, inferior pubic rami and proximal femurs. The deformity involved patchy lucency and sclerosis as well as diaphyseal widening. Metastatic disease was a consideration but considered unlikely considering the other skeletal abnormalities.

The most recent plain film x-rays taken within the last 12 months demonstrated enchondromatous lesions in the left proximal humerus and acromion process, bilaterally at the wrists including lesions of distal radius, irregularity of the distal ulna and shortening of the ulna on both sides, bilaterally in the hands, especially involving all distal metacarpal metaphyses, and proximal phalanges in the proximal metaphyses. The femur was involved distally on both sides, right proximal tibia and fibula had metaphyseal changes. The femurs were also bowed, possibly due to a previous fracture or enchondromatous lesions. The distal tibia on both sides had enchondromatous lesions and there were two exostoses, one about 3-4 cm in length and the other, much smaller, from the distal tibia on the lateral side which pointed distinctly toward the ankle joint.

A working diagnosis of Metachondromatosis was made on the basis of enchondromatous lesions, their distribution in the body, exostoses and the absence of recognisable genetic markers that would indicate Gaucher disease. Although the bony lesions on radiographs were Gaucher like in appearance, the lesions were largely symmetrical, had an extremity distribution and there was no history of bone crises. An analysis of marrow in an affected area would definitively rule out Gaucher disease.

**TREATMENT**

The patient received manual manipulation of the cervical, thoracic and lumbopelvic regions but (primarily the cervical and thoracic spines). The manipulation of the lower back was performed only once, without incident. The manual manipulation involved a short lever contact and used a controlled force to cavitate the joint(s). The lower back manipulation was performed with the patient sidelying with mild torsion of the trunk. The thoracic manipulation was usually performed reclining the patient in a flexed position over a flat hand and the cervical manipulation was performed in a supine position with a combination of lateral flexion and rotational forces. Substantial specific massage techniques were performed in addition to the manipulation. The patient responded well to treatment. Her various symptomatic complaints, that consisted of minor joint sprains and muscular aching pains, were assisted by this regime of treatment. She found the pain resolved quickly over a period of 1 week in the initial presentation and she did have further minor injuries over the following 2 years. The author considers it unlikely that ongoing periodic treatments would have diminished the incidence of these minor sprains.

No attempts were made to do anything with the enchondromatous lesions and the exostoses. It was felt that the treatment did not have any significant effect on these bony lesions.

**DISCUSSION**

It is likely that a patient with metachondromatosis or related disorders will enter an osteopathic or chiropractic clinic in the future. The incapacity of this condition is slight, with the patient presenting with common, mostly unrelated musculoskeletal complaints that commonly present to the manual therapist. The clinical problem in metachondromatosis is whether the bone is strong enough...
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in the extremities, pelvis and spine to withstand manual manipulative techniques. The fractures that have been documented over the last 35 years in this patient with Metachondromatosis certainly indicate that bone strength is minimal in affected areas.

In the above case of metachondromatosis, the involvement can be almost symmetrical and widespread, involving all 4 limbs and shoulder and pelvic girdles. The conical metaphyses, similar to the Erlenmeyer-flask deformity in Gaucher disease and marked metaphyseal changes with irregular trabecular markings are visually striking. The fact that she had suffered 7 fractures that have been identified, would suggest the enchondromatous lesions to be fragile. On the last 2 occasions the trauma associated with fractures was minor, and on the last occasion the causative action could not be identified. The healing of such fractures in metachondromatosis, based on this case, appears to be reasonable. Fractures in metachondromatosis have not been described in the literature.

CONCLUSION

Multiple fractures have occurred with a patient suffering from metachondromatosis. Manual therapists need to evaluate using plain film x-ray for areas of enchondromatous lesions in the extremities and consider that all these lesions have the potential to fracture. In addition, the Manual therapist when manually manipulating the pelvis should consider that there may be possible involvement of the ischium or innominate bones and the proximal femur. Forces applied through these structures could bring about an unwanted pathological fracture. There appears to be little evidence for involvement of the spine and thus manipulation of the cervical and thoracic spine would appear to have no significant additional risk. The exostoses in metachondromatosis may be multiple and may compress neighbouring structures potentially resulting in compression syndromes of peripheral nerves. Such potential problems should be considered on a case by case basis after regional examination.

Importantly, misdiagnosis may lead to other complications as Gaucher disease often involves pathological weakness of the thoracic and lumbar vertebral bodies. Osteochondromatosis can involve osteochondral bodies within the joint spaces, potentially leading to mechanical impingement and sometimes malignant degeneration. Awareness of the possible negative outcomes to manual techniques and the sensible clinical management of this and other related conditions by manual methods is highlighted.

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