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Now you see it, now you don't – a case of "Vanishing Bone"

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ABSTRACT

Vanishing bones – no, not the movie fantasy of the "Invisible Man" but a case of a rare disease which causes massive osteolysis or progressive bone dissolution. The idiopathic disease, which causes bone to be replaced by angiomatous tissue, has resulted in the disappearance of this patient's right clavicle and glenoid. Diagnostic evaluation can sometimes prove difficult but ironically in this age of higher technology, plain radiography has demonstrated to be more effective for a definitive diagnosis than Nuclear Medicine, MRI, CT, etc. The rate of progress of the disease is as unpredictable as the prognosis. While a variety of treatments have been tried, the results have been inconsistent and as yet there is still no consensus on successful patient treatment.

Table 1– Osteolysis Syndromes[1]

Syndrome	Age of onset	Major site of osteolysis	Patterns of inheritance	Associated features
Acro-osteolysis of Hajdu and Cheney	Second decade	Distal phalanges; rarely tubular bones,mandible, acromioclavicular joints	Dominant or sporadic	Generalised bone dysplasia, fractures, osteoporosis
Massive Osteolysis of Gorham	Child, young adult	Variable; pelvic or shoulder girdles	Sporadic	Slowly progressive extreme dissolution
Carpal-tarsal osteolysis: Multicentric Osteolysis with Nephropathy	Infant, child	Carpal and tarsal areas, elbows	Sporadic; occasionally dominant	Osteoporosis, deformity, hypertension, renal failure, death
Hereditry Multicentric Osteolysis	1-5 years	Carpal and tarsal areas, elbows, digits	Dominant; occasionally recessive or sporadic	Progressive deformity
Neurogenic Osteolysis	Childhood	Phalanges	Dominant or Recessive	Sensory neuropathy, skin ulcerations
Acro-osteolysis Of Joseph	Childhood	Distal phalanges	Recessive	Otherwise healthy
Acro-osteolysis Of Shinz	Second decade	Phalanges	Dominant	Skin ulcerations, no neurologic defect
Farber's disease	Infancy	Elbows, wrists, knees, ankles	Sporadic	Subcutaneous nodules
Winchester's Syndrome	Infancy	Carpal and tarsal areas, elbows	Recessive	Osteoporosis, joint contractures, thick skin, corneal opacities
Osteolysis with Detritic synovitis	Adulthood	Widespread	Sporadic	Progressive

Table 1 above shows the list of nine Osteolysis Syndromes showing the ages of onset, most prevalent anatomical sites affected, heredity patterns and the prominent pathological features of each Syndrome.

INTRODUCTION

Moviemakers have long since captivated audiences imaginations by making a man invisible. In real life, it is possible to witness, radiographically, an act of invisibility within the human body – in particular that of vanishing bones. Unlike the movie version, Gorham's disease, which causes this process, is not reversible

Gorham's Disease is a rare syndrome of "massive osteolysis", one of nine recognised, different Osteolysis Syndromes.[1] As can be seen in Table 1, these Syndromes can collectively appear at any age and attack any part of the bony skeleton.[1] In 1938, Jacobsen first reported a case of spontaneous absorption of

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bone.[2] In 1955, Gorham and Stout reviewed twenty four similar cases and emphasised the angiomatous nature of the of the osteolytic lesions i.e. the affected bone is replaced by angiomatous tissue, which consists of fibrous tissue comprised of thin walled, ectactic capillary like vessels, containing either blood or lymph.[3] Although the angiomata (haemangiomata/ lymphangiomata) are histologically benign, the disease is characterised by a progressive course of bone destruction.[2]

Aetiology and Pathogenesis

The term "massive osteolysis" is based on the typical radiological findings such as increased translucency and loss of bone density. Gorham's disease has a plethora of synonyms including Vanishing Bone Disease, Gorham – Stout Disease, Phantom Bone Disease, Massive Osteolysis of Gorham and lymph or haemangiomatosis.[4,5,6] This abundance of terms reflects the poor understanding that persists nearly 50 years after Gorham and Stout established this process as a distinct syndrome.[6] The aetiology of the disease is unclear and there are only approximately 150 reported cases in the medical literature which makes it an extremely uniqu , interesting and mysterious condition.

Gorham's disease usually occurs in childhood but 30 per cent of patients are over 35 years old. It is indiscriminate to age, sex, culture and not genetically linked, as was first thought.[6] The disease can occur in any bone[2] but mainly affects bones that develop by intra- membranous ossification eg shoulder girdle, pelvis, mandible, ribs or spine.[7] Initially the disease progresses slowly to affect a single bone but adjacent bones may be affected as the disease spreads.[2] Extra skeletal sites may also be affected by a generalised lymphangiomatosis eg the spleen, and skin.[6]

CASE PRESENTATION

Classically, the disease occurs in the shoulder or pelvis, usually starting with a pathologic fracture and consequent deformity.

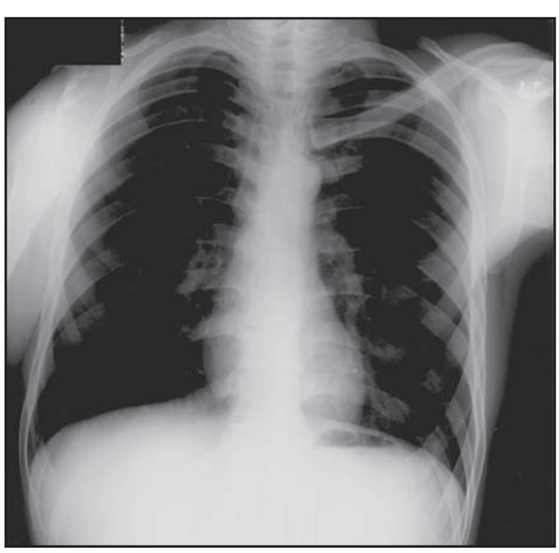

Figure 1: Chest X-Ray taken in 2001, demonstrating complete absence of the right clavicle and glenoid with subluxation of the right humerus. The chest and remaining bony skeleton is otherwise normal.

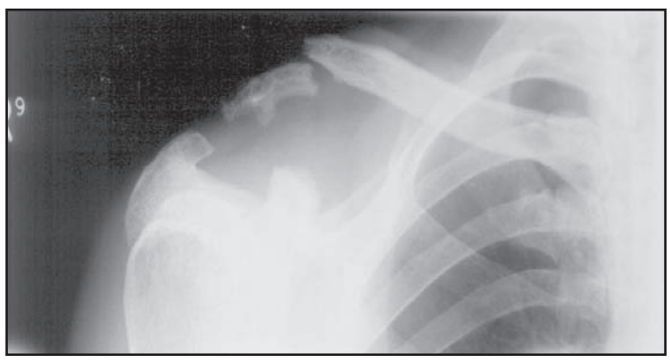

Figure 2: Right shoulder X-Ray taken in 1996, demonstrating a fracture of the outer one third of the right clavicle with A.C. Joint dislocation. Remaining bony skeleton appears normal.

This case involves a 40-year-old indigenous male from a remote North Queensland settlement who presented at his local hospital in 2001,with pain and weakness in his right shoulder. A Chest X-Ray demonstrated complete absence of the right clavicle and glenoid with subluxation of the right humerus .The lung fields were clear. See Figure 1. Looking back to 1996,the patients initial X-Ray showed a fracture of the outer one third of his right clavicle with dislocation of the acromio-clavicular joint. The outer fragment has the appearance of a loss of bone volume. See Figure 2. A Chest X-Ray taken in 1998, demonstrated absence of the outer half of the right clavicle, the residual medial half displayed a "sucked candy" appearance with increased translucency, compared to the left clavicle .The lung fields were clear. See Figure 3. Over a five-year period, this patient's right clavicle and glenoid have completely vanished while the remaining bony skeleton appeared normal.

The patient was then referred to Townsville General Hospital in 2001 for investigation and diagnosis. A Nuclear Medicine Bone scan was performed. This showed moderate

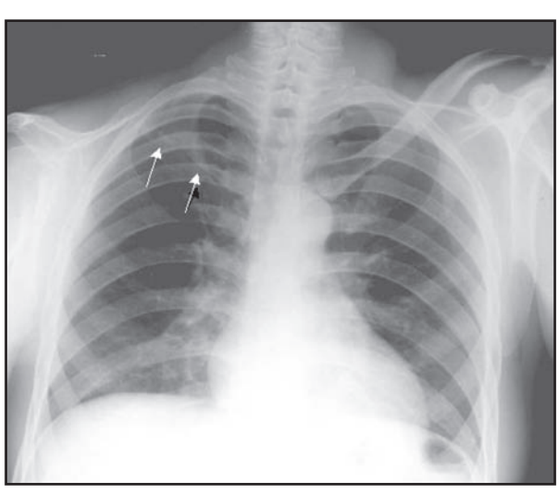

Figure 3: Chest X-Ray taken in 1998, demonstrating absence of the outer half of the right clavicle, the medial half (see arrows) displaying a sucked candy appearance and increased translucency compared to the left clavicle. Chest and remaining bony skeleton is normal.

increased blood pooling in the right shoulder particularly in the glenoid and spine area of the scapula. While there is less blood pooling than expected for an infective process, the appearance is not specific. This correlates well with the increased vascularity associated with angiomatous spread. The other focal abnormality noted in the ankle was traumatic in origin. See Figure 4. An MRI was requested but never performed as the patient has literally 'disappeared' back into his community and no further examinations or treatment have been recorded for him at this Hospital.

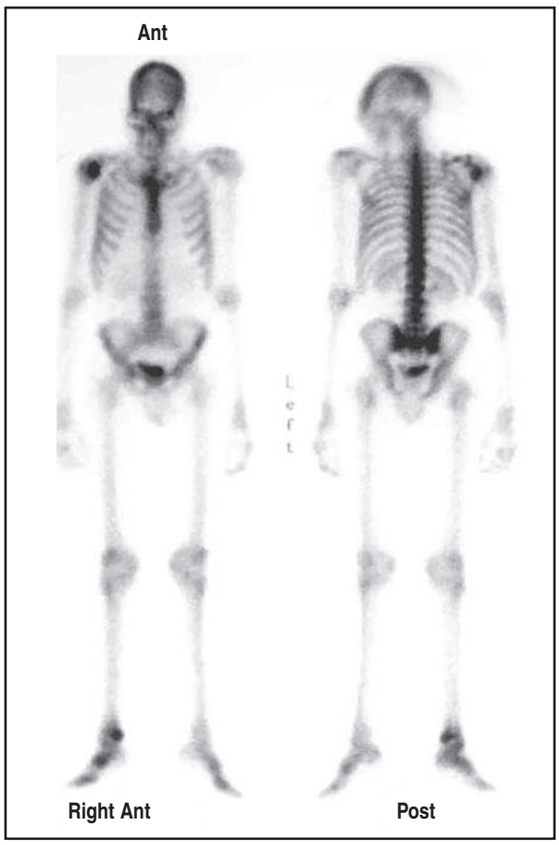

Figure 4: Anterior and Posterior images from a Whole Body N>M scan taken in 2001. Increased blood pooling is shown in the right glenoid and spine of scapula. The appearance is not specific but shows the extent of Gorham's disease. Increased uptake at the right ankle is probably traumatic.

Radiologic Features

This is dependent on the stage of the disease – initially radiolucent subcortical and intramedullary focii may be present, succeeded by expanding intramedullary lytic areas with no sclerosis. The characteristic absence of osteogenesis is striking-the above features are best demonstrated on plain X-Rays. In tubular bones, usually following fracture, tapering of the fragment ends will result in a "sucked candy" or cone shaped appearance. Massive osteolysis usually progresses and in the late stages the bones may disappear completely. Phleboliths may be observed in the soft tissues suggesting associated angiomatous elements.[4]

Diagnosis

Diagnosis in the early stages may be difficult with the appearance of lytic areas in the cortex or medullary cavity. The pointed bone ends, after a pathological fracture, may suggest the pseudarthrosis of neurofibromatosis or fibrous dysplasia.[4] The long indolent course of the disease, five years in this case, along with relevant clinical and osteolytic radiological features, which are particularly dramatic as in "classic" advanced cases, usually provides the diagnosis. Rather fortunately, the Senior Radiologist on duty had seen a single case of Gorham's disease in the spine, many years beforehand and issued a diagnosis almost immediately.

If doubt still exists, conclusive pathological evidence from biopsy of the medullary structures will demonstrate vascular proliferation but no osteoblastic activity.[4,5,7]

CT, MRI, Ultrasound and Nuclear Medicine have all been used but none have the ability for a definitive diagnosis. These modalities can best show the extent of the disease ie as in this case Nuclear Medicine scan shows the disease limited to the scapula.[6] Angiography has not been helpful, although it can sometimes show an increase in blood flow, it rarely shows a pattern suggesting an angiomatous malformation.[4]

Prognosis

The rate of progression of the disease is unpredictable and the prognosis can be difficult.[4]

This current condition of this patient is unknown and he has the possibility of further massive osteolysis of his scapula and adjacent bones, having already eroded the glenoid .His right arm has been rendered useless due to his dislocated humerus and can not be surgically repaired. The disease can either:

1) Stabilise after a number of years.
2) Go spontaneously into remission; or
3) Prove fatal – this is particularly so in patients who develop chylothorax from a lymph obstructed thoracic duct and die of pulmonary complications due to massive effusions.[1,2,6] Involvement of the spine and skull base may also lead to death from neurological complications. Recurrence of the disease can occur.[4]

Treatment

Only palliative treatment was recorded for this patient in 2001

There is no consensus on treatment.[7] This is mainly palliative but radiotherapy and chemotherapy have both been tried with inconsistent results. Surgical amputation or resection with joint prothesis, internal fixation or bone grafting have been used but recurrence is not infrequent and further surgery has been necessary.[4] Other treatment regimes are:

1) Pharmaceutical Agents that inhibit bone resorption – bisphosphonates and calcitonin as well as
2) Alpha Interferon and fractionated Radiotherapy which are known to inhibit angiogenesis.[6]

RESEARCH

Recent research on the disease by Aviv, McHugh and Hunt, 2001 suggest that angiomatoses are a spectrum of disease processes incorporating lymphangiomatosis and Gorham's Disease where the clinical, radiological and histological features often make distinction difficult.[6]

CONCLUSION

Gorham's Disease is a rare disorder of massive osteolysis, with

the dramatic radiological feature of vanishing bone. The patient who presented here in this case suffered massive osteolysis of the right clavicle and scapula, which has been recorded radiographically. No treatment was recorded other than palliative care and his condition is currently unknown. A legacy of its rarity and unknown aetiology is the lack of effective treatment, which offers a mixed prognosis ranging from spontaneous remission to death.

ACKNOWLEDGEMENT

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