Ollier's Disease with Parotid Involvement

Ramesh Mundle [1], Manjusha Mardikar [2], Nalini Humney [2],
Sharad Dhurve[3], Nitin Wadaskar [4], Debjani Goswami [4], Sony Thomas [4].

ABSTRACT

Ollier's disease is a rare, developmental nonhereditary skeletal disorder of mesodermal dysplasia characterized by multiple asymmetric distribution of cartilaginous tumors, enchondromas, with a predilection for unilateral distribution. This condition primarily affects the long bones and cartilage of the joints of arms and legs, usually in the region where rate of growth is most marked i.e. metaphysis. They lead to skeletal deformities and limb length discrepancies. The pelvis is often involved but ribs, sternum and skull are seldom affected. Very rarely it involves the soft parts, as in the salivary glands, testes of men and the breast of women. The Prevalence is 1/100000. The disorder is usually recognized in childhood. The cause is not known and there is no medical treatment. Surgery is indicated where complications arise, like pathological fractures, growth defects and malignant transformation to chondrosarcoma. Few Indian cases of Ollier's disease have been reported. This report describes a case of 32 yrs young male of Indian origin who presented with multiple enchondromatosis with bilateral parotid enlargement which is very rare and reported for the first time in India.

Introduction:

Ollier's disease, discovered by and named after the French surgeon Louis Xavier Le'opold Ollier (1830-1900) (1), is a disorder in which the ends of bones grow improperly and multiple cysts form all over the body. These large cartilaginous cysts form benign, painless bone tumors which are not life threatening. (1) Ollier's disease is most common in the hand, femur, humerus and tibia. Most cases are sporadic. The cartilage affects the inner parts of the periosteum. Symptoms are usually discovered during first decade of life. Involvement of parotid glands and testes is described though very rarely. (2,3)

Address for correspondence

1. Professor 2. Associate Professor 3. Lecturer, 4. Residents

Department of Medicine, N.K.P.Salve Institute of Medical Sciences, Digdoh hills, Hingna, Nagpur.

Case History:

History of Presentation

A 32 years, young male presented to us with progressively increasing painless swelling of the distal phalanx of left middle finger since 1 year, progressively increasing painless swelling on left forearm just above the left wrist joint medially since 3 months and bilateral parotid enlargement since 3 months. He had lower limb length discrepancy, the left lower limb being longer than the right limb. He had no major illness in past. His two other siblings and parents were normal.

Physical Examination:

On Examination: his pulse was 84/min, regular; all peripheral pulsation well felt, BP-110/80 mm Hg, there was venous varicosity on the left calf, bilateral parotid enlargement was present, left more than the right. The glands were soft, multilobulated, nontender with a discharging sinus on the superior

aspects of left parotid. There was a single lymph node palpable in the posterior cervical region on left side. It was discrete, mobile, nontender, 0.5 cm x 3 cm in size. Bilateral multiple, small, nontender, discrete inguinal lymph nodes were palpable. Exostosis was present at the distal end of the left ulnar bone. There was swelling on the left middle finger 3cm X 3 cm X 2 cm in size at the distal interphalangeal joint. The swelling was soft, nontender, lobulated, encircling the finger with increased vascularity. A punctum was present at the medial aspect of the swelling. Talipus Equinus deformity was present in both feet. Length of the left lower limb was longer than the right limb. The respiratory, cardiovascular, abdomen and central nervous system examination was normal. There was no haemangioma anywhere in the body.

Relevant investigations:

Hb: 11.7gm%

P.Smear: Microcytes +, Normocytes +, Mild hypochromia, Platelets adequate

TLC/DLC: normal

Blood Sugar Random-84mg%

Serum Creatinine: 0.69 mg%

HIV I and II- Negative

Urine Routine- Normal

X-ray both hands with wrist joint. (AP and oblique view)

- A large geographic ill defined lucent lesion seen in the epimetaphyseal region of the ulna on left side. Lateral margins of the lesion were smooth. The lesion was showing few internal septations. The lesion was causing thinning and expansion of the cortex. The lesion was purely lytic and margins also did not reveal sclerosis. Another ill defined heterogeneous lytic lesion seen in right radius was showing septae.
- Multiple irregular lucent areas were seen in the distal end of right second to fourth metacarpals. The lesions were of varying sizes.
- Similar morphology lesions were also seen in phalanges.

X ray of Middle Phalanx of 3rd finger:

- It showed irregularity of the cortex with associated soft tissue component. The soft tissue swelling was showing increased vascularity upon limited Doppler assessment.

X ray both feet AP view

- Few well defined lucent lytic areas were seen in the 1st and 2nd metatarsals on right side. No marginal sclerosis/ calcification were seen.

X ray Both Knee Joint: AP and Lateral view

- Ill defined lucent lytic areas were seen in tibia on both sides. A round heterogeneous predominantly sclerotic lesion having peripheral zone of sclerosis and lucencies interspersed within was seen in the lower end of femur medially. Imp: These findings were suggestive of Multiple Enchondromatosis. (Ollier's Disease)

FNAC from left parotid swelling:

Smears showed sheets of small lymphocytes with few acinar cells in the background. No other cell type seen in smear studied. Cytological features suggested chronic inflammatory lesion.

FNAC from Middle Finger Swelling:

Sparsely cellular smears. Smears showed few small cells entangles in fibrin strands.

Background is haemorrhagic. Histopathology study recommended for conclusive diagnosis. Impression : Possibility of **Enchondromas**.

Treatment Given:

Patient was counselled about the nature of

the disease and its complication and was given various surgical options along with importance of regular follow up. Patient was not willing for any surgical intervention or further histopathological studies. Hence he was given symptomatic treatment and discharged. He was advised to follow up at 6 monthly intervals or if there were any complaints or pain in the existing swellings.

Discussion :

Chondromas are bening tumors of hyaline cartilage divided primarily into ecchondromas or the hyperplastic forms and the enchondromas or the heteroplastic forms (3). Ecchondromas proceed from the permanent cartilage and enchondromas occur in places where cartilage should not be formed, as in the bones, where it is most frequently observed and also in the soft parts as in the salivary glands, in the testes of men and the breasts of women (3).

When multiple enchondromas are present, the condition is called the enchondromatosis also known as Ollier Disease (WHO terminology) (4). The estimated prevalence of the disease in 1/100000. No racial predilection is known and it occurs equally in males and females. Clinical manifestations often appear in the first decade of life (s).

Etiology : Endocondral bone ossification requires progression of undifferentiated mesenchymal cells into hypertrophic chondrocytes and subsequent replacement of a cartilageneous matrix by mineralized bone (6). Enchondromas/Ollier Disease may result from abnormalities in signalling pathways controlling the proliferation and differentiation of chondrocytes leading to intraosseous cartilagenous foci. Olliers disease is thought to occur due to postzygotic somatic mutation resulting in mosaism. Whether single gene or multiple gene mutations occur is not known. Formation of cancerous tumors from enchondromatosis come from varying expression of parathyroid hormone receptor protein PTHrP or PTHRI (1).

Clinical Features : Enchondromas frequently affect the long tubular bones, particularly the tibia, the femur and or the fibula. On physical examination the enchondromas present on the extremities are usually visible as masses embedded within phalanges, metacarpal and metatarsal bones. Lesions are asymmetrically distributed and exclusively or predominantly affecting one side of the body (1), though they may at times be present bilaterally.

Enchondromas are by no means as innocent as Johann Muller thought. From the extension into the soft parts masses have been carried into lymph and blood vessels and metastatic forms have been noticed. Among the glands, it is the salivary and the sexual, which are chiefly affected, and among the former, the submaxillary and the parotid. The enchondroma may be either diffuse, affecting the whole of the gland or lobular affecting only certain lobes. The former is seldom seen in the parotid, most common form in the submaxillary. The sexual glands in the female are much less frequently affected than in men. In the testicles it generally occurs complicated with some other kind of tumor (3).

Radiography : Enchondromas on x-ray are seen as multiple, radiolucent, homogenous lesions which run parallel with long bone axis. They are frequently assembled in clusters, thus resulting in epiphyseal widening. Lesions are localized in the metaphysis of long bones and in the small bones of hands and feet. In the hands, the lesions almost never affect all metacarpal bones and phalanges (7).

Diagnosis: of Ollier disease is based on clinical and conventional radiological evaluations. Additional investigations like scintigraphy, ultrasound. MRI are not useful. Histology has limited role. **Differential diagnosis** may include-Hereditary multiple

exostosis HME, polyostotic fibrous dysplasia, klippel-trenaunay Syndrome, Weber-parks syndrome.

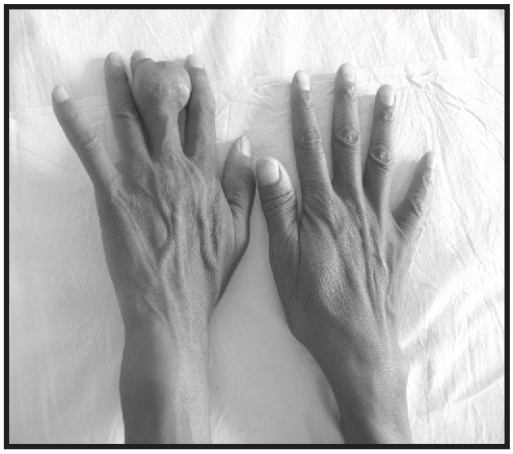
Complications may arise in the form of pathological fractures or malignant change as chondrosarcoma and osteosarcoma. About 25% of cases undergo malignant change by the age of 40 (8).

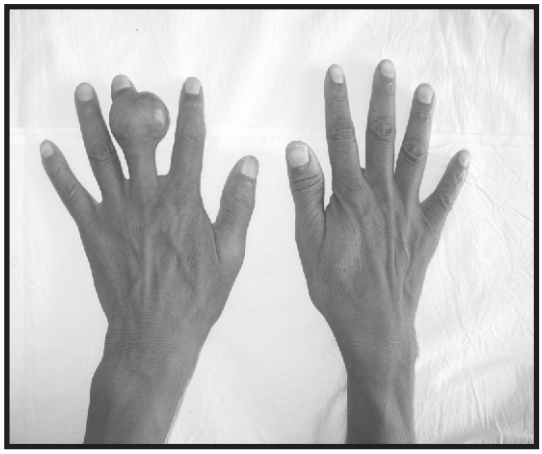
Treatment : There is no medical treatment and surgery is indicated in enchondromatosis complicated by pathological fractures, growth defect or malignant transformation. Prognosis is difficult to assess (1).

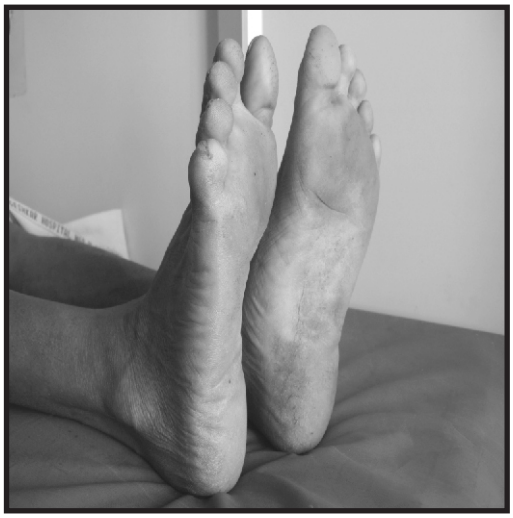
Conclusions: The patient who presented to us had symptoms for the first time in the third decade of life. Though clinical examination revealed disease on the left side, radiological features were seen bilaterally. Also the right lower limb was shorter than left suggesting disease early on the right side. Bilateral parotid involvement was again a rare presentation in him.

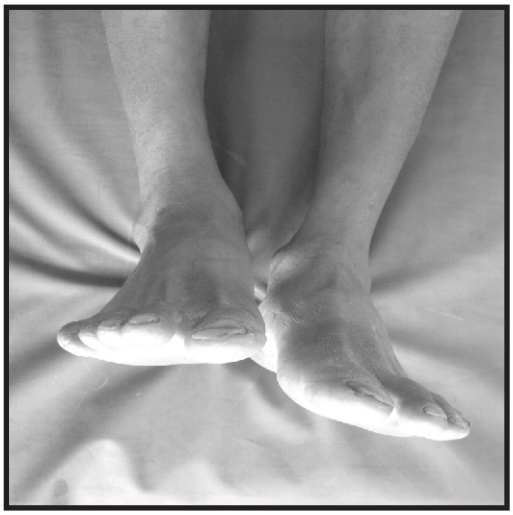
References :

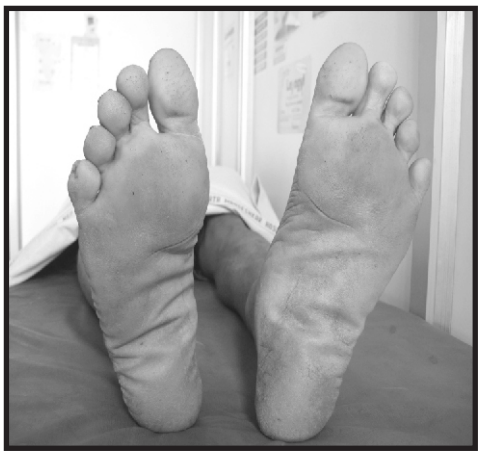
1. Silve C. Jupner H (2006). "Ollier disease". Orphanet J Rare Dis 1:37.
2. Matt and Leela Probert 1993-2010. The Probert Encyclopedia of Medicine, Southampton, United Kingdom.
3. The American Journal of Medical Sciences Vol 49. Virchow lectures on morbid tumors No : XLV II-Jan 1865, Issue -97, Page 175-177. By William Merrick Sweet, Southern society for clinical investigation (U.S.), Daniel and Eleanor Albert collection.
4. Fletcher CDM, Unni K, Mertens F. (Ed) World Health Organization, Classification of Tumor Pathology and genetics. Tumors of soft tissue and Bone. Lyon: IARC Press 2002, Page 427.
5. Maroteaux P, Le Merrer M. Les Maladies osseuses de l'enfant. Paris : Medicine-Sciences, Flammation, 2002
6. Kronenberg H M. Developmental regulation of the growth plate. Nature. 2003; 423:332-336.
7. O Adam, ES Boia. Ramona Mandrusca R. Mejdi. Enchondromatosis-Ollier Disease-Case report. Jurnalul Pediatrului Year XII, Vol XII Nr 47-48, July-December 2009.
8. Schwartz HS, Zimmerman NB Simm MA, Wroble RR. The malignant potential of enchondromatosis. Bone joint Surg Am. 1987; 69: 226-274 (Pubmed).

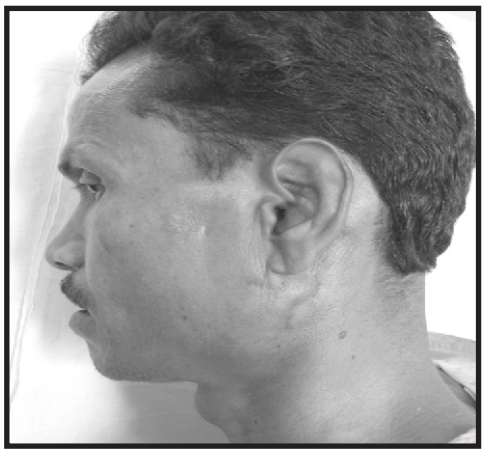


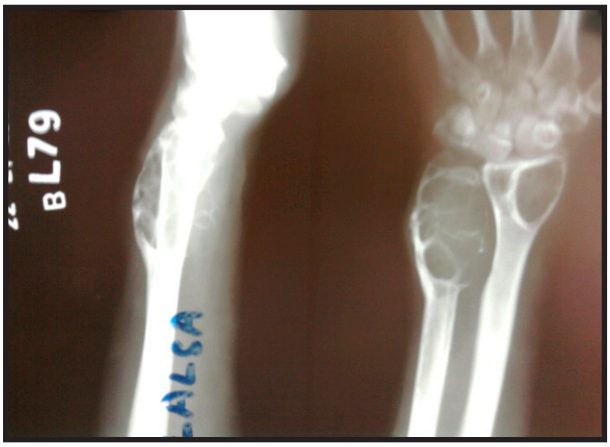


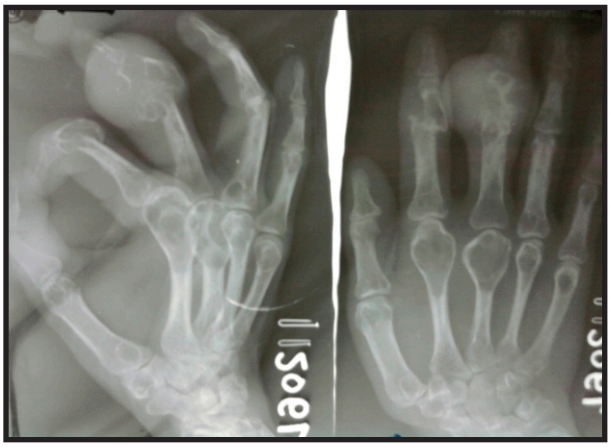


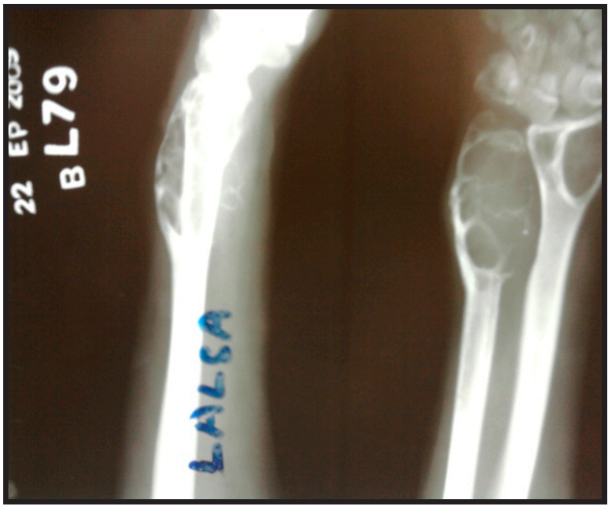







BL79
LALSA




22 EP 2009
BL79
LALSA

Instructions To Authors

"Vidarbha Journal of Internal Medicine" official publication of the "Association of Physicians of India-Vidarbha Chapter.

The Journal will consider for publication manuscripts suitable as original papers, brief reports, reviews, current perspective articles, case reports, letters to the editor, selected summaries from important articles published elsewhere, calender of conferences, and other special category articles of interest to the physicians. The editorial specially encourages submission of rare case-reports, presented in the various clinical meetings of all other affiliated chapters of API.

Manuscripts should be addressed to Dr. S. R. Tankhiwale, Chief Editor, Vidarbha Journal of Internal Medicine, Ashwini Nursing Home, W. H. C. ROAD, Nagpur -440 010. E-mail:

Statements and opinions expressed in the articles published in the Journal are those of the authors and not necessarily of the Editor, Neither the Editor nor the Publisher guarantees, warrants or endorses any product or service advertised in the Journal. If accepted for publication, the manuscript may be edited. Without altering the meaning to improve clarity and understanding. Two or more referees will scrutinize every original or review article submitted to the "Vidarbha Journal of Internal Medicine".

Guidelines for Manuscript Preparation:

1. Original Articles:: The manuscript should be prepared in accordance with the 'Uniform requirements for manuscripts submitted to biomedical journals' compiled by the International Committee of Medical Journal Editors. (NEJM 1997; 336: 309-315). (Vancouver style)

Three complete sets of the manuscript should be submitted; typed in double-space on one side of the page throughout (including references, tables and legends to illustrations).

The manuscript should be arranged as follows: Covering letter. Title page. Abstract. Introduction. Methods. Results. Discussion. Acknowledgments. References. Tables, and Legends to Illustrations.

Three sets of illustrations in three separate envelopes should be attached at the end. The pages of the manuscript should be numbered consecutively beginning with the title page.

Please also submit an electronic version (CD), along with the printed copies to facilitate processing.

Covering Letter: The covering letter should explain if there is any deviation from the standard IMRAD (Introduction. Methods. Results and Discussion) format and should outline the importance and uniqueness of the work. All authors must sign the covering letter with the understanding that:

1) All authors have reviewed the article and agree with its contents.

2) The article is entirely original.

3) No portion of the article is under consideration elsewhere for publication or has been previously published. (This restriction does not apply to abstracts published in connection with scientific meetings.)

Title page (Page 1): This should contain the Title, Names of all the Authors written in continuation.

Name(s) of institution(s) they are affiliated to. Names (first name, initial of middle name followed by surnames)

of all the authors (without degrees or diplomas). Name and full location of the department / institution / laboratory where the work was performed.

The Name. Telephone number. Fax number, e-mail and Postal Name and Address of the Corresponding Author and Acknowledgment of financial support if any.

Structured Abstract (Page 2): Should be typed on a separate sheet

The abstract (250 words maximum) should be structured and divided into sections namely Background/ Rationale, Methods and Results, Discussion and Conclusions.

Also, provide 3 to 5 keywords. Terms from the medical subjects headings (MeSH);list of Index Medicus should be used.

Text (Page 3 onwards): The text should be divided into Introduction. Methods. Results and Discussion.

Give generic rather than trade names of drugs.

Avoid the use of full stops in between abbreviations (TMT. not T.M.T.).

Introduction: The introduction should state why the study was carried out and what were its specific aims.

Methods: These should be described in sufficient detail to permit evaluation and duplication of the work. This section may be divided into subsections, if required. Ethical guidelines followed by the investigators should be described. Statistical details should be provided, where applicable.

Results

Discussion: should consist of review of literature and relate the major findings with other publications.

References: in Vancouver style and should be numbered consecutively in the order in which they first appear in the text.