Ollier’s Disease: 1 Case Report & Literature Review

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Abstract
Ollier’s disease is a rare tumor of unknown etiology. The main manifestation of this disease is a non-ossifying chondrocyte mass or hamartomatous growth of a chondrocyte in the metaphysis. A few cases can develop into chondrosarcoma or osteosarcoma. The present study describes the case of a 16-year-old female patient in King George Hospital, VSKP, Andra Pradesh, India seeking medical help for calf muscle cramps, stiffness, pain and non-progressive swelling of all fingers for the past 1 year. The incidence of Ollier disease is low, and thus, it is not well-known and due to its malignant transformation rate 20-30%, patients need to be followed up at regular intervals.

Introduction
Ollier in 1889 described a condition of multiple, typically unilateral enchondromas associated with deformity of the extremity referring to the condition as dyschondroplasia, a term implying that it resulted from a developmental defect related to abnormal growth of cartilage. The etiology of Ollier’s disease has not been identified, however somatic mosaic mutations of isocitrate dehydrogenase (IDH1) and (IDH2) is known to be related, Amary et al. The diagnosis is solely based on clinic-radiological evaluations. It is sporadic in nature and not hereditary. It mainly manifest as local pain, swelling, deformity and palpable bone mass. If multiple cavernous hemangiomas are associated, Maffucci syndrome is made as diagnosis.

Case Report
A 16 year old girl had initially developed and obvious shortening of finger and toes and frontal bossing of forehead at the age of 10 years; however no special attention and osteopathic intervention was advised.
She was born full term without intra or peripartum complications. Her menstrual cycle are normal. There was no limitation in movement of her fingers and no obvious swelling is seen over zones proximal to Zone 2. The swelling of the digits were hard and tender. She also has frontal bone prominence with exophthalmosis. The patient came to King George Hospital, Vishakapatnam, Andhra Pradesh, India on 10th May, 2023 due to local pain of all her fingers and pain over her calves. The presenting complaints has impaired her daily lifestyle for the past 1 year before which there was no daily activity impairment. Since the onset of pain, the patient did not have any symptoms of blurring of vision or persistent headache. She has no history of chemical or radiation exposure, and no history of same complaints in family. She does not indulge in alcohol and tobacco consumption.

Figure B (bony swelling, clubbing with short phalanx)

A physical examination upon admission revealed that there were no limb length discrepancy, fusiform swelling of phalanges and clubbing which were tender on palpation without erythema. Range of motion were with normal limits and there was also mild tenderness along bilateral foot which she did not complaint of in the presenting complaints. Gait was normal and pelvic compression was negative. There was no appreciable purpura, telangiectasia or ecchymosis. An x-ray examination of skull, hand, arm, leg and foot revealed multiple osteopathies of B/L hand, B/L Foot and B/L Tibia. MRI examination was then followed following which the clinic-radiological diagnosis of Ollier Disease was made. MRI spine indicated no spinal stenosis.

Figure C (frontal bossing)
1) Right foot- multiple osteolytic lesion at distal end of tibia, fibula, proximal phalanx of 1st, 2nd, 3rd toe, middle phalange of 2nd toe. Similar osteolytic lesions of 1st metatarsal bone- findings sequelae to Ollier’s disease

2) Left foot- Multiple small enchondromas involving distal end of tibia on medial aspect, proximal phalanx of 1st, 2nd, 3rd and 5th toes and also similar lesion at distal phalanx of great toe suggestive of Ollier’s disease

3) Hand with wrist- multiple osteolytic lesion with internal septations noted on all phalanx suggestive of multiple enchondromas involving all the phalanges of hand, involvement of 1st metacarpal bone and distal radius and ulna.

**Ultrasonography of Chest and abdomen reveals no abnormality**

The blood reports of the patient are presented in the table1.

Following Wrist block, biopsy from the 1st Phalange Rt Middle finger was attempted with 14g Trucut biopsy needle, however there was no substance to be taken and the wound was closed with sterile dressing and no further attempt was made due to the fear of pathological fracture.

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**Fig D – Xray of skull and proximal leg**

**Fig D – Xray of B/L Foot**
Table 1 – Lab reports of the patient

<table>
<thead>
<tr>
<th>LABS</th>
<th>VALUES</th>
<th>NORMAL RANGE</th>
</tr>
</thead>
<tbody>
<tr>
<td>HB</td>
<td>10.5 g%</td>
<td>14 - 18 g% (male)</td>
</tr>
<tr>
<td></td>
<td></td>
<td>12 – 16 g% (female)</td>
</tr>
<tr>
<td>TLC</td>
<td>4500 cells/cu.mm</td>
<td>4000 – 11000 cells/cu.mm</td>
</tr>
<tr>
<td>ESR</td>
<td>20</td>
<td>&lt;20 mm/h (female)</td>
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<tr>
<td></td>
<td></td>
<td>&lt;15 mm/h (male)</td>
</tr>
<tr>
<td>CRP</td>
<td>Negative</td>
<td></td>
</tr>
<tr>
<td>PLATELET COUNT</td>
<td>3.1 lakhs/cu.mm</td>
<td>1.5 – 4.5 lakhs/cu.mm</td>
</tr>
<tr>
<td>INR</td>
<td>1.0</td>
<td>&lt; 1.2</td>
</tr>
<tr>
<td>SERUM CALCIUM</td>
<td>11.1 mg/dl</td>
<td>8.5 – 10.5 mg/dl</td>
</tr>
<tr>
<td>SERUM PHOSPHORUS</td>
<td>5.6 mg/dl</td>
<td>3.4 – 4.5 mg/dL</td>
</tr>
<tr>
<td>FREE T3</td>
<td>277 pg/dL</td>
<td>260 – 480 pg/dL</td>
</tr>
<tr>
<td>FREE T4</td>
<td>1 ng/dL</td>
<td>0.7 -1.8 ng/dL</td>
</tr>
<tr>
<td>TSH</td>
<td>2.1 mIU/L</td>
<td>0.5 - 4.5 mIU/L</td>
</tr>
<tr>
<td>SERUM ALP</td>
<td>98 U/L</td>
<td>45 – 115 U/L</td>
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<td>TRIPLE SEROLOGY</td>
<td>NEGATIVE</td>
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<tr>
<td>SERUM UREA</td>
<td>21 mg/dl</td>
<td>20- 40 mg/dl</td>
</tr>
<tr>
<td>SERUM CREATININE</td>
<td>0.9</td>
<td>&lt; 1.4 mg/dl</td>
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Discussion

(8) Multiple enchondromatosis is a rare heterogeneous skeletal disease, which can be divided into six subtypes as follows:

1) Ollier disease
2) Maffucci syndrome
3) Metaphyseal chondromatosis
4) Metatarsal chondromatosis
5) Spinal interstitial chondrodysplasia
6) Spinal interstitial chondromatosis.

Since these lesions can be associated with brain, pancreatic, pelvic or skull base diseases attention should be paid to lesions various systems(9-15). In addition, due to of the high risk of malignant transformation, patients should undergo close and long-term follow-up.

<table>
<thead>
<tr>
<th>PARAMETRES</th>
<th>OBSERVED VALUES</th>
<th>NORMAL RANGE</th>
</tr>
</thead>
<tbody>
<tr>
<td>WEIGHT</td>
<td>45 KG</td>
<td></td>
</tr>
<tr>
<td>STANDING HEIGHT</td>
<td>1.49 metres / 149 cms</td>
<td>&gt; 124cms</td>
</tr>
<tr>
<td>BMI</td>
<td>20.17 kg/m.m</td>
<td>18.5 – 24.9 kg/m.m</td>
</tr>
<tr>
<td>TRUNK LENGTH (SITTING HEIGHT)</td>
<td>25 inch</td>
<td></td>
</tr>
<tr>
<td>LOWER LIMB LENGTH</td>
<td>33.8 inch</td>
<td></td>
</tr>
<tr>
<td>LTR ( LEG-TRUNK RATIO)</td>
<td>1.35</td>
<td>LOW &lt; -0.675 (&lt;25%)</td>
</tr>
<tr>
<td></td>
<td></td>
<td>MEDIUM &gt; -0.675 - 0.675 (&lt;75%)</td>
</tr>
<tr>
<td></td>
<td></td>
<td>HIGH &gt; +0.675 (&gt; +0.675)</td>
</tr>
</tbody>
</table>

(Reference- Standing height to rule out achondroplasia, LTR ratio indicating higher leg length for a given body height. High LTR ratio inversely proportional to the development of hypertension in adulthood- http://academic.oup.com/jpubhealth/article/38/4/6 88/2966969)

Diagnosis

The diagnosis of Ollier disease is based on clinical manifestations and a routine radiological evaluation. A histological analysis is mainly used to identify malignant tumors.

The first onset of Ollier disease usually appears within the first 10 years of life, appearing as a single or multiple bone mass. With the increase in age, corresponding limb deformities appear in
Ollier disease, deformities, which affects the growth and development of children.

Clinical description:
Ollier disease mainly manifests in the short bones of the hands and feet, followed by the tibia, femur, humerus, radius, ulna. Chondroblastomas have also been found in the skull, maxilla, temporal bone, ribs, pelvis, hands, proximal femur, patella, talus, calcaneus, and throughout the spine. Multifocal benign chondroblastomas have been reported. Nonepiphyseal locations in the long bones have been described. Neurologic deficits can occur if the vertebrae are involved.

In the present study, it was found that this patient had abnormalities and unbalanced growth and development during pre-adolescent and adolescent stages.

Imaging Diagnosis
Radionuclide bone imaging is the most effective tool for the diagnosis of multiple bony lesions as it can guide the clinical and prognostic analysis of Ollier disease. An X-ray is the most basic tools for the diagnosis of skeletal diseases. Bone abnormalities are usually more extensive than the physical examination would suggest. In the long bones, enchondromatosis is recognized as radiolucent longitudinal streaks that involve the metaphysis and extend down into the diaphysis. Epiphyses are usually not affected but may be involved. The cortex overlying the enchondroma is usually thin, and calcification within the lesion is common. Significant shortening and angular deformity are frequently noted in the involved long bones, whether in the hands, feet, or limbs. CT scan is important in evaluating soft tissue invasion with lesion of spine and abdomen. The clinical diagnosis of Ollier disease is relatively simple, and no sex-associated trend has been observed. The first onset of Ollier disease usually appears within the first decade of life. Fine-needle aspiration yields satisfactory material for interpretation and confirmation of the diagnosis. However, in our case we could not attain material for interpretation through trucut, void cavity to blood aspirates.

Sarcomatous Change
The incidence of secondary chondrosarcoma in patients with Ollier disease is approximately 25% to 30% by 40 years of age. Those patients with Maffucci syndrome have a similar or higher likelihood of development of malignant degeneration. Schwartz and associates reported a nearly 100% expectation of malignant degeneration. Over the long term, periodic surveillance of the brain and abdomen for occult malignant lesions is indicated in patients who have enchondromatosis. Increased localized growth of a lesion in an extremity accompanied by pain is the hallmark of possible malignancy. In such a situation, biopsy of the lesion is indicated.

Hematopoietic malignant diseases (acute lymphoid leukemia and chronic myeloid leukemia) have been described in association with both Ollier disease and Maffucci syndrome.

Treatment
Because of the extent of the disease, multiple enchondromatosis cannot be cured by curettage and bone grafting. The numerous deformities that often accompany multiple enchondromatosis require repeated operative interventions over several years to correct the angular deformities and achieve similar limb lengths at maturity. These procedures include osteotomies, limb lengthenings, and epiphysiodeses. In the older child, use of the Ilizarov apparatus is the best method to achieve both angular correction and equalization of limb lengths. Use of multiple wires or half-pins allows sufficient purchase in the enchondromatous bone so that lengthenings can be successfully achieved. It has been reported that there is no specific drug treatment for Ollier disease. For patients with Ollier disease without significant functional impairment, a long-term follow-up is warranted.
The treatment of complications associated with Ollier disease remains predominantly surgical, and is mainly considered in the following cases:

1) Bone lengthening surgery can correct deformities and can maintain balanced limb development as much as possible
2) For patients with pathological fractures, the traditional surgical treatment is usually adopted, involving the curettage of lesions and internal fixation with bone grafts
3) Surgical resection remains the primary treatment for chondrosarcoma associated with Ollier disease

References


34. Loder RT, Sundberg S, Gabriel K, et al: Determination of bone age in children with cartilaginous dysplasia (multiple hereditary osteochondromatosis and

