Ollier's Disease with Myelodysplastic Syndrome
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ABSTRACT
Ollier’s disease also known as enchondromatosis is a rare skeletal disorder that is usually sporadic, non-hereditary, and characterized by abnormal bone development (skeletal dysplasia). While this disorder may be present at birth (congenital); it may not become apparent until early childhood with more obvious symptoms, such as deformities or improper limb growth. It carries high risk of skeletal, visceral and brain malignancy seen in approximately 25% of patients. Occurrence of Ollier’s disease with myelodysplastic syndrome has never been reported in the literature. The different types of myelodysplastic syndromes are diagnosed based on certain changes in the blood cells and bone marrow characterized by one or more cytopenias despite a relatively hypercellular bone marrow. We hereby report the case of a 14 years boy who presented with painless finger swelling and hepatosplenomegaly. Radiological and bone marrow findings confirmed the diagnosis of Ollier’s disease with Refractory Anemia and Excess Blasts (RAEB-1).

Key Words: Ollier’s disease. Enchondromas. Refractory anemia with excess blasts.

INTRODUCTION
Ollier’s disease is a rare intraosseous, usually benign cartilaginous tumors (enchondroma), that occur close to the growing plate cartilage. The estimated prevalence of Ollier’s disease is 1/100,000.¹ It remains uncertain whether the disorder is caused by a single gene defect or by combinations of (germ-line and/or somatic) mutations. The diagnosis is based on clinical and conventional radiological evaluations. Histological analysis has a limited role and is mainly used if malignancy is suspected.

It is usually associated with granulosa cell tumor of the ovary, chondrosarcomas and other malignancies.²⁻⁴ Coexistence with refractory anemia and excess blasts is very unusual and reported for the first time at the time of submission.

CASE REPORT
A 14 years boy presented to the Hematology/Oncology Department of The Children’s Hospital, Lahore through emergency with 5 months history of pallor. There was history of progressive swelling and deformity of finger joints since one year of age which had never been investigated. There was no family history of similar illness. He was developmentally normal, born to non-consanguineous parents and has 6 other healthy siblings. He never attended school due to bony deformities.

On examination, he was a pale looking child having maxillary prominence and dental overcrowding. Vital signs were stable and growth parameters were on the 75th centile (height = 135 cm, weight = 35 kg). Systemic examination revealed liver of 3 cm below right costal margin (total span = 10 cm) and spleen palpable 8 cm below left costal margin. Local examination revealed marked bony deformities with hard swellings in both the hands with functional limitation (Figure 1). Bone age corresponded with chronological age; Sexual Maturity Rating (SMR) was stage 1. Abdominal ultrasound showed grossly enlarged spleen (13.6 cm) with normal liver and other viscera. Complete blood counts showed haemoglobin level at 5.0 gm/dl, WBC count of 1.2 x 10⁹/l, (neutrophils 40%, lymphocytes 56%) platelet count was 180 x 10⁹/l, no atypical cells were seen. Baseline renal and liver function tests, coagulation profile, and screening for hepatitis B and C were all negative. Skeletal survey showed multiple lytic lesions expanding the metacarpals and phalanges bones of both hands (Figure 2) with lesions in the distal metaphysis of radius.
Ollier’s disease with myelodysplastic syndrome

and ulna along with shortening of ulna bilaterally. Similar, lesions were also noted in the proximal metaphysis of humeri and glenoid cavity, proximal femoral epiphysis. The calvarium showed widening. Thoracolumbar spine was normal. Bone marrow trephine and aspiration biopsy showed trilineage hematopoiesis with 10% blasts. Dysplastic changes were present in myeloid series and in Megakaryocytes. Findings were suggestive of Myelodysplastic Syndrome (MDS), specifically Refractory Anemia with Excess Blasts (RAEB 1). Parents were counselled and they opted for palliative therapy.

DISCUSSION

Ollier’s disease is a very rare condition that is characterized by multiple enchondromas throughout the skeleton principally located in the metaphyseal regions. The typical distribution is in small tubular bones of the hands and feet in 50% of cases and in large tubular bones e.g. femur, tibia, humerus in the remaining. Radiologically enchondromas appear as small lytic lesions with narrow zone of transition, sharply defined scalloped margin. Expansion of the overlying cortex may be present but there should not be cortical break unless fractured. Ring and arched chondroid calcifications may be present without any soft tissue mass or periosteal reaction.

The exact cause of Ollier’s disease is not known yet but is believed to be a random spontaneous mutation. A mutation (p.R150C) in PTHR1 (3p22-p21.1) has been reported. It shows no gender predilection, and usually becomes apparent by early childhood. Clinical presentation is usually with deformity. If pain occurs, this should raise the concern of malignant transformation or pathological fracture. Enchondromas in Ollier's disease present a risk of malignant transformation into chondrosarcomas.

There is an increased risk of chondrosarcoma occurring later in life. The risk has been reported to be up to 25-30% at 40 years. Multiple enchondromatosis associated with soft tissue hemangiomas is known as Maffucci syndrome. Until now Ollier's disease has only occurred in isolated patients. There has been an association with astrocytoma, glioma, and Gilbert syndrome. The diagnosis is based on clinical and conventional radiological evaluations. Thus, this patient was unique in that he presented with pallor and bony deformities of hand. On skeletal and bone marrow examination was subsequently diagnosed as having Ollier’s disease with MDS, which has never been reported in the literature. MDS are heterogeneous group of closely related clonal hematopoietic disorders, characterized by ineffective hematopoiesis with dysplastic changes in the marrow.

There is no medical treatment for enchondromatosis. Surgery is indicated in case of complications (pathological fractures, growth defect, malignant transformation). The prognosis for Ollier's disease is difficult to assess. As is generally the case, an early onset appear to be more severe. The diagnosis of Ollier's disease with MDS further complicates the prognosis.

REFERENCES


Figure 2: Bilateral hand X-rays showing multiple expansile lytic lesions.