Evaluation of the Surgical Management of the Skeleton in Patients with Ollier's Disease and Mafucci Syndrome

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INTRODUCTION: Both Ollier's disease and Mafucci syndrome are rare and have a high rate of skeletal abnormalities. Due to the rare nature of these diseases, the risk of fracture and incidence of surgery performed is not well defined. In addition to surgery for fracture and malignancy, many patients have procedures for deformities and limb-length discrepancies. The purpose of this study is to delineate the number and type of surgical procedures performed in this patient population.

METHODS: After obtaining IRB approval, an online survey containing 25 questions was distributed through online support groups to patients with Ollier's disease and Mafucci syndrome. Both the patient and the immediate family member/caregiver were allowed to respond to the survey.

RESULTS: A total of 86 surveys were obtained from 5 continents including North America (52), Europe (27), Australia (4), Africa (2), and South America (1). Seventy-three patients had Ollier's disease and 13 had Mafucci syndrome. The average age of the patient was 30.1 (3-81). Five patients never had surgery on their bones. Fifty-two had at least one fracture which first occurred at 8.4 years of age on average (0.75-29). Thirty-eight had surgery due to an enchondroma causing pain or fracture, whereas 53 had surgery to correct a limb-length discrepancy or deformity. The average age when they first had surgery was 9.0 years (0.5-41) and each patient had an average of 8.1 surgeries (1-49). The most common location for surgery for an enchondroma was the hand, followed by the femur and tibia, and plate fixation was the most common implant. For limb-length discrepancy, the tibia/fibula was the most common surgical site followed by the femur and radius/ulna. Implants included external fixators (29), plates (22), and intramedullary nails (9). Deformity correction was most commonly performed on the tibia/fibula followed by the femur, hand, and forearm with external fixation (26), plate fixation (24), and intramedullary nails (12) as the most common implants. Seventeen patients developed cancer at an average age of 27.1 (11-46) and 14 were chondrosarcomas. Six patients developed cancer at a site where a previous surgery was performed and 3 of these patients developed metastatic disease. Five of the initial surgical procedures were to correct a deformity or limb-length discrepancy and 2 of the 3 patients who had intramedullary nails placed developed metastatic disease. The remaining patient who developed metastatic disease had 4 procedures on the same finger with no implant placed.

DISCUSSION AND CONCLUSION: Surgery is very common in patients with Ollier's disease and Mafucci syndrome, with most patients requiring multiple procedures. The majority of patients have surgery for deformity correction and limb-length discrepancies. Caution should be taken in the skeletally mature patient undergoing these procedures, particularly with an intramedullary nail, to ensure no malignancy is present.