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Overgrowth Management in Klippel-Trenaunay-Weber and Proteus Syndromes

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Summary: Twenty-eight patients with limb overgrowth and the diagnosis of Klippel-Trenaunay-Weber or Proteus syndromes were evaluated retrospectively. These disorders are part of the phakomatosis spectrum of syndromes. The orthopedic problems consisted of asymmetric limb overgrowth, localized gigantism, angular deformities, scoliosis, vascular malformations, and skin anomalies. Systemic abnormalities are common and deserve full evaluation before treatment. Surgical treatment consisted of epiphysiodesis, osteotomies, debulking procedures, and amputation. Mixed results were obtained with surgery, and conservative or supportive treatment should be the primary mode of orthopedic care. **Key Words:** Asymmetric overgrowth—Judicious surgery—Klippel-Trenaunay-Weber syndrome—Proteus syndrome—Systemic manifestations.

Overgrowth syndromes in the pediatric population are either symmetric or asymmetric. The former generally involve endocrinopathies. Conditions with asymmetric or partial gigantism involve skeletal and soft tissues alike and are the focus of this study. Probably all of these entities belong to the phakomatosis family. These are a group of overgrowth syndromes that consistently have highly variable soft tissue and skeletal hyperplasia. The Greek term "phakos" refers to a spot birthmark or nevus. The phakomatoses typically have hamartomas, which are congenital tumorlike or developmental malformations with primary ectodermal origins, but may also be mesodermal or endodermal (17,18). The phakomatosis family consists of at least 50 different syndromes with varying clinical presentations. These have many common characteristics and are either monogenic or sporadic. The neurofibromatoses are the prototypes of these conditions and exemplify the monogenic state. Klippel-Trenaunay-Weber and Proteus are sporadic phakomatoses secondary most likely to somatic mutations or germline mosaicism (14,25,33). These two conditions have many common characteristics and may be part of the encephalocranio-cutaneous lipomatosis subgroup of phakomatoses as described by Kousseff and Madan (17). The asymmetric overgrowth requires stabilization and improvement of function.

The physical manifestations of Proteus and Klippel-Trenaunay-Weber syndromes have many common characteristics, with overlap of their stigmata. Clinical course and treatment methods are similar, but Proteus syndrome shows more frequent progression of the hamartomatous growth. For these reasons, in this article they will be considered a spectrum of the phakomatoses, dealing with orthopedic management of patients with these two syndromes.

MATERIALS AND METHODS

We made a retrospective review of 28 patients with Klippel-Trenaunay-Weber or Proteus syndromes. Patients with neurofibromatosis, Sturge-Weber syndrome, and unclassifiable asymmetric overgrowth conditions were omitted. Any condition with a Mendelian genetic inheritance pattern was excluded. No patient had a positive family history for a similar disorder. All cases in this study arose sporadically. Patients with isolated macrodactyly were excluded if they had no other systemic manifestations of overgrowth syndromes (12). Eighteen patients were diagnosed with Klippel-Trenaunay-Weber syndrome, and 10 were diagnosed with Pro-

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