History
1 year old male with enlarged 1st and 2nd digits.

Diagnosis
Macrodactyly

Discussion
Focal tissue hypertrophy is often associated with peripheral nerve pathology. The nervous system probably exerts some controlling action on the process of growth, such that impaired nerves result in uninhibited growth. Therefore, the differential diagnosis for this case includes lipomatoses of the nerve (macrodystrophia lipomatosa), neurofibromatosis, and Proteus syndrome. Lipomatoses of nerve presents early in life. While there is no known genetic defect, hypertrophy of mature fat and fibroblasts in the epineurium is thought to result in the morphologic changes. The upper extremity is affected in most of cases, particularly the median nerve. The most frequent clinical manifestation is a slowly growing mass at the wrist, carpal tunnel symptoms and macrodactyly. There is a male predilection in cases not associated with macrodactyly, whereas a female predominance has been noted in cases with digit overgrowth.

The classic peripheral manifestations of neurofibromatosis include limb hemihypertrophy, pseudarthrosis, peripheral nerve neurofibromas, and subcutaneous and plexiform neurofibromas. In the hand cutaneous neurofibromas can cause macrodactyly, plexiform neurofibromas can cause osseous erosions and hypothalamic disease can result in accelerate maturation. Proteus syndrome causes an overgrowth of skin, bones, muscles, fatty tissues, and blood and lymphatic vessels. Patients with Proteus syndrome are usually normal at birth but deformities develop and progressive. The severity and locations of these various asymmetrical growths vary greatly but typically the skull and/or limbs will be affected. Macrodactyly is usually associated with enlargement of the metacarpals and hypertrophy of subcutaneous soft tissues.

Findings
CR-Soft-tissue hypertrophy of the 1st and 2nd digits.
MR-Sagittal and coronal T1 confirms soft tissue hypertrophy. Axial T2 shows marked hypertrophy of the median nerve; fascicles are clearly evident although no overt perineural fat.

Reference
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