Maffucci Syndrome

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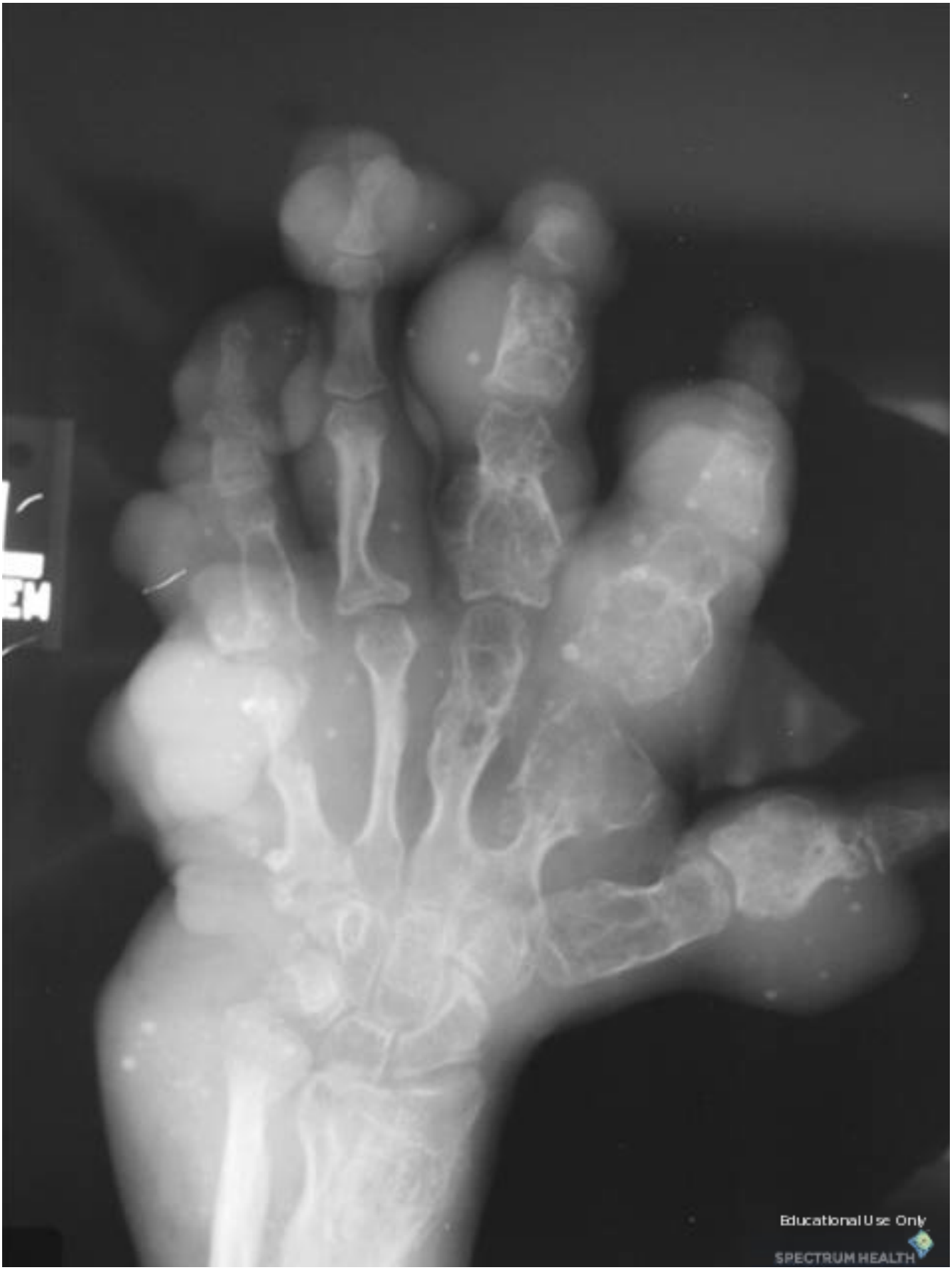
History
Teenager with significant left hand disability.

Diagnosis
Maffucci Syndrome

Discussion
The mutant receptor PTH/PTHrP type 1 is found in the enchondromatous tissue activating the Hedgehog signaling and enchodromatous growth. Maffucci syndrome is enchondromatosis with angiomatosis.
Malignant degeneration occurs in approximately 30%, usually in adulthood, and is more common in Maffucci syndrome. Additionally, there is a higher incidence of extraosseous tumors (usually brain and ovary).
Maffucci syndrome is often confused with Ollier disease. Both have enchondromatosis. Ollier disease has been used to describe unilateral enchondromatosis whereas Maffucci syndrome is enchondromatosis with angiomata.

Findings
CR-Multiple ovoid and pyramidal lucent osseous defects some with expansion and cortical thinning, numerous lobulated soft tissue tumefactions, and numerous phleboliths.

Reference
Spranger JW, Brill PW, Poznanski AK. Bone Dysplasia: An Atlas of Genetic Disorders of Skeletal Development, 2nd Ed. Oxford University Press 2002.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