59th ANNUAL DIAGNOSTIC SLIDE SESSION 2018.

CASE 2018 #10

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Clinical History:

An 11-year-old male was previously healthy until approximately the year prior, when the family noticed that he was walking on the lateral aspect of his right foot, resulting in callus formation and closed pressure ulcer. During physical examination, it was noted that the patient had a high arch of the right foot as well as a 3 cm. leg length discrepancy. Several subtle café-au-lait spots were also identified. Subsequent radiographs demonstrated multiple stress fractures, which prompted MRI of the spine and lower extremities demonstrating enlargement of the right lumbosacral plexus and right sciatic nerve extending distally down the thigh, progressing to include the tibial nerve and common peroneal nerve to the bifurcation with continued involvement of the deep and superficial peroneal nerve branches to the foot. At the time of surgical intervention for orthopedic corrections, a biopsy of the tibial nerve was performed.

Material submitted: Virtual H&E image of tibial nerve biopsy

Points for discussion:

- 1-Differential Diagnosis
- 2-Pathogenesis
- 3-Role of molecular genetics