

**Canadian Association of Neuropathologists
L'Association Canadienne des Neuropathologistes**

CASE No 7

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This man had a long and tortuous history, but died of a clinically enigmatic illness that began at age 27 and claimed his life by age 31. Born at 36 weeks (2931g), his postnatal period was complicated by RDS requiring ICU care for several weeks. He was discharged after 84 days. Routine lung X-ray led to discovery of a problem affecting the bones, which gave a radiological appearance "as if someone had scooped out the inside of the bones". Developmental delay necessitated special schooling. He underwent correction of genu valgus. Age 11 in Quebec Special Olympics, won a Silver Medal for discus/shot put. Graduated age 18 from vocational school and butchery course, lived independently, did his own banking, got his learner's permit to drive, worked in security and restaurants. Age 27, progressive neurological condition began with intellectual, physical and personality changes, progressive dysarthria, bilateral gaze-evoked nystagmus, intention tremor and poor heel-shin on left (could tandem walk only 3-4 steps). He was aggressive at times. Full scale IQ was 57; verbal IQ 64; performance IQ 56 (WAIS age 16 IQ scores were 70-79). Age 28 profound physical deterioration began with clenching of left hand and dragging left leg; had to pry fingers off objects. Hypertonia and brisk reflexes in all limbs (L>R); dystonic posturing left hand/arm. Power 5/5, sensation normal. Deteriorated to a wheelchair-bound condition with rigidity, weakness, spasticity. Over 30 pneumonias. PEG tube for repeated aspiration, death by age 31.

Material Submitted: 1 H&E slide

Question: Diagnosis?