

Case 3

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Clinical Abstract: A 36 year old woman was admitted to hospital in June of 1986 after a fall at home. Her neurologic problems first appeared at age 13 when she was admitted to a hospital because of quadriplegia. Myelogram discloses a syrinx from the medulla to C₆. At C₁₋₄ laminectomy the surgeon noted that the ligamentum flavum and dura were thickened, and the arachnoid was "milky." Function improved post-operatively.

Myelogram in March at age 22 disclosed incomplete block at C₃₋₄. Skull x-rays showed bony densities in both parieto-temporal regions. Chest x-ray disclosed developmental abnormalities of the clavicles, anterior ribs, humeral heads and vertebral bodies. An ophthalmologist diagnosed hyperopic astigmatism and endothelial corneal dystrophy. A C₄₋₇ laminectomy was done in June .

She was stable until age 27 when she developed a mild quadriplegia and headaches. She had mild optic disc swelling and spasticity in the lower extremities. CT scan of the head revealed moderate enlargement of the lateral and fourth ventricles. A ventriculo-peritoneal shunt led to improvement of symptoms.

Cervical spine x-rays at age 33 showed narrowing of the vertebral bodies and widening of the prevertebral soft tissue space.

Examination showed a short woman with coarse facial features, acne and a prominent forehead. Both corneas were cloudy. The interphalangeal joints of the hands and feet were visibly enlarged. A niece had a similar appearance.

She had a suboccipital craniectomy and C₇ laminectomy.

Material Submitted: Soft tissue removed at surgery.

Points for Discussion:

Diagnosis