Familial syringomyelia

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SUMMARY A woman with syringobulbia and syringomyelia and her sister with syringomyelia are described. The diagnosis was confirmed radiologically and surgically in the first sister and no other definite abnormalities of her craniocervical junction were found.

Only five kindreds with true familial syringomyelia are reported in the English language literature to our knowledge.1-6 We describe two sisters, one with syringobulbia and syringomyelia and one with syringomyelia.

Case reports

Case 1: At age 12 yrs, this 33-year-old woman developed dysesthesias and numbness in the left arm, shoulder, and face. Numbness and burning pain subsequently involved the face, head, shoulder, chest, and arms bilaterally. She noted diplopia, oscillopsia on down gaze, difficulty tasting, hoarseness, dysphagia, weakness of both upper extremities, and unsteady gait.

Physical examination revealed a short neck and kyphosis of the lower cervical and upper thoracic spine. She had a right Horner’s syndrome and diplopia on left and up gaze with the right eye image projected laterally. There was clockwise rotatory nystagmus on lateral and down gaze, and vertical nystagmus on down gaze. Pin, touch, and temperature sensation were diminished over the face. The left corneal reflex was absent. Taste was decreased on the anterior two thirds of the left tongue. She was hoarse, the left soft palate did not elevate, and the left gag reflex was absent. The left sternocleidomastoid was weak. The tongue deviated to the left on protrusion. The legs were stronger than the arms; the left side was stronger than the right. The right hand was weakest and intrinsic hand muscles were wasted. Tandem walk was unsteady and tandem Romberg manoeuvre was positive. Tendon reflexes were absent in arms, normal in legs. Babinski’s sign was absent. Position sense was intact. Vibratory sensation was slightly impaired in the left fingers. Pin and temperature sensation were markedly decreased from C2 to T6, more on the left. Touch was felt above T6, but was diminished on the left.

Serum laboratory values, skull and cervical spine radiographs, cranial CT scan, and visual and brainstem auditory evoked responses were normal. CSF protein was 0.57 g/l. Latencies of median somatosensory evoked responses were normal, but the amplitude of the P/N13 wave (upper cervical cord-lower medulla) on left-sided stimulation was one third that on the right. Right peroneal nerve somatosensory evoked responses were normal. N/P37 (scalp) and LP-N/P37 (lower back to scalp) latencies were prolonged on left peroneal nerve stimulation. Sensory nerve conduction studies were normal as were motor nerve conduction velocities. Electromyographic evidence of active denervation and/or reinnervation was present in right C5 to T2 myotomes and left C7 to T1 myotomes.

Cervical metrizamide myelography revealed low-lying cerebellar tonsils that did not project below the foramen magnum and a small diameter cervical spinal cord. A cervical CT scan 6 hours later showed uptake of contrast into the spinal cord from C2 to C6, indicating a syrinx.

At C7-T1 laminectomy, the spinal cord was flattened and atrophic, but it expanded with each positive pressure inspiration. At midline posterior myelotomy a smooth-walled cavity containing clear colourless fluid was entered at a depth of 2.5 mm. A spinal cord stent drain was placed. She tolerated the procedure well and was little changed two months later.

Case 2: This 50-year-old sister of Case 1 noted dysesthesias in left face, arm, and chest in her early teens and subsequently noted weakness and decreased pain and temperature sensation in both upper extremities. On examination she had a short neck and mild kyphosis. Cranial nerves were normal except for slightly decreased pin and temperature sensation in the trigeminal distributions, worst on the left. Both upper extremities were weak, proximally greater than distally, left greater than right. The intrinsic hand muscles were atrophied. Tendon reflexes were absent in the arms, but normal in legs. Babinski’s sign was absent. Position and touch sensation were intact. There was some decreased vibration sensation in the left hand and markedly decreased pin and temperature sensation from C3 to T8 on the left, and from C4 to T6 on the right. She declined further evaluation.

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Other family members
There was no consanguinity and the paternal side of the family had no known neurological complaints. The mother of Cases 1 and 2 did not feel pain or temperature in her hands and could take things out of a hot oven without using potholders, but it was uncertain if her upper extremities had been weak. A brother had right arm weakness and decreased pain and/or temperature sensation in his hands. Three other brothers did not have neurological complaints. A maternal first cousin had a baby with "wide open spine" who died a few months after birth. Case 2's three children and sixteen nieces and nephews had no neurological complaints.

Discussion
Case 1 had confirmed cervical syringomyelia and symptoms and signs consistent with syringobulbia. Case 2 had symptoms and signs of cervical syringomyelia. The mother of these women may have had a similar disorder, suggesting dominant inheritance.

Few well documented kindreds with cervical syringomyelia\(^1\)\(^-\)\(^5\)\(^7\)\(^-\)\(^9\) with either dominant\(^6\) or recessive\(^4\)\(^7\)\(^8\) inheritance have been reported. Unequivocal radiological and surgical confirmation was obtained in only one family\(^3\) and pathological confirmation was obtained in another.\(^8\) Myelo- graphic enlargement of the cervical spinal cord was found in one member of each of three other kindreds, but intramedullary cavities were not demonstrated.\(^3\)\(^4\) Reports of other families said to have cervical syringomyelia (reviewed in\(^2\)\(^4\)\(^10\); see\(^11\)\(^-\)\(^13\)) contain insufficient confirmatory data.

Many members of kindreds with syringomyelia have occipital dysplasia, basilar impression, and/or the Arnold-Chiari Type I malformation.\(^3\)\(^5\)\(^9\)\(^14\)\(^15\) Syringomyelia, when present, could be secondary to altered cerebrospinal fluid flow due to the other anomalies at the craniocervical junction.\(^16\) The findings in Case 1 may argue against this hypothesis as no other definite anomalies of her craniocervical junction were found. However, her craniocervical junction was not explored at surgery, she did not receive a magnetic resonance imaging scan,\(^17\)\(^-\)\(^19\) and such abnormalities could have been present at an earlier age.

Familial syringomyelia may occasionally arise from shared environmental factors since dizygotic twins that both had syringomyelia have been reported,\(^1\) some monozygotic twins are not concordant for syringomyelia,\(^1\) and there is increased prevalence of syringomyelia in certain areas.\(^1\)\(^20\)\(^21\)

Familial syringomyelia may result from abnormalities in genes directing spinal cord development.\(^22\) Molecular genetic techniques potentially enable these genes to be identified.\(^23\) Syringomyelia and myotonic dystrophy were simultaneously inherited in two families,\(^24\)\(^-\)\(^26\) and the genes may be linked in these kindreds. Marker loci for the myotonic dystrophy gene on chromosome 19\(^23\)\(^27\)\(^28\) might also serve as markers for some forms of familial syringomyelia.

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