Familial syringomyelia

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SYNOPSIS  Four cases of syringomyelia in two separate families are reported.

This paper reports four cases of syringomyelia in two separate families involving two sisters and a brother and sister. Familial syringomyelia is a very rare occurrence and has not been previously documented in the United Kingdom.

CASE 1

Mrs F.H., aged 54 years, is the eldest of four sisters. She was referred by her general practitioner for diagnosis because of radiological changes in her left hand compatible with a degenerative arthritis. The patient gave a 25 year history of inability to detect temperature changes, and pain in the left arm; the arm had become noticeably weaker over the years, and she had experienced difficulty in moving her shoulder fully for several years.

On examination, she had a mild kyphosis with a short neck. There was subcutaneous tissue swelling and thickened skin over the left hand and forearm with old and recent scars from healed burns within this area. There was mild wasting and a detectable weakness of the muscles of the left hand. Forearm and shoulder girdle muscles were detectably weaker than in the right arm. Left upper limb tendon jerks were absent. There was complete inability to detect pain and temperature sensation over the C2 to T6 dermatomes on the left. Light touch was impaired over the left C2 to C4 dermatomes only. Vibration sense was impaired in the left arm and in the left leg. Joint position sense appeared normal, as was her balance on either leg. Apart from an absent triceps jerk neurological examination of the right upper limb was normal. There was no spasticity of the lower limbs and plantar responses were flexor. Cranial nerves were normal and there was no nystagmus.

A left Charcot shoulder joint was present and radiography also confirmed degenerative joint changes in the metacarpophalangeal and interphalangeal joints of the left hand. Skull and cervical spine radiographs were normal. Myelography showed herniation of cerebellar tonsils to the C1 level (Chiari type I malformation) with slight dilatation of the cervical cord. Spinal fluid was normal with a protein content of 30 mg/dl. A diagnosis of syringomyelia was made.

In summary, this patient has a classical 25 year history of unilateral syringomyelia. It is unclear at present whether her condition continues to deteriorate and for this reason surgery has not been advised.

CASE 2

Mrs C.K. is aged 46 years and the younger sister of F.H. (case 1). In 1955 she presented with a two year history of difficulty in gripping with both hands and inability to detect hot and cold water with them. She had experienced several painless burns and injuries. Syringomyelia was diagnosed and she received radiotherapy to the cervical cord in February 1956. The patient had not been seen from 1956 until 1973, she felt that her condition had been fully arrested and denied any symptoms to suggest otherwise.

Recent examination showed that she had a right Horner's syndrome and impairment of the right corneal response with slight loss of pain sensation of the periphery of her right face in front of the right ear as the only cranial nerve abnormalities. There was moderate wasting of the small muscles of both hands and forearms with some clawing of the ulnar fingers, severe weakness of finger flexion and almost total weakness of the interosseus muscles of both hands. All upper limb tendon jerks were absent. There was no abnormality of power or tone in the lower limbs but knee reflexes were abnormally brisk and both plantar responses extensor. Sensory testing showed severe impairment of pain over the right C2 to T1 dermatomes and left C5 to C8 dermatomes, with inability to appreciate temperature changes over a similar area. There was mild inability to appreciate light touch over all fingers. Vibration sense was absent below her rib cage. Proprioceptive sense and balance were good. There was no clinical evidence...
of Charcot arthropathy. Radiographs of the skull and cervical spine were normal, as was a spinal fluid examination with a protein content of 25 mg/dl. A myelogram has not been performed.

In summary, this patient has a 21 year history of syringomyelia. She herself feels that her condition has been arrested after radiotherapy in 1956. Clinical examination would support this.

CASE 3
Mrs M.L., aged 51 years, is one of four siblings and first presented in 1963 with a two year history of weakness in the right hand and burning sensations in the right upper limb. Eleven years previously she had suffered shooting pain in the neck radiating to the forehead and associated with numbness of the left side of her neck and face, all exacerbated by sneezing. These symptoms, together with burning feelings over the left face, disappeared spontaneously at the time. Examination showed subcutaneous thickening of the fingers of the right hand. There was moderate wasting of the small muscles of the right hand with corresponding weakness. Tendon jerks were absent in the upper limbs and increased in the lower limbs, but plantar responses were flexor. There was impaired sensation to pain in the right C7 to T1 and left C3 to C4 dermatomes but other sensations appeared normal. Cranial nerves were normal and there was no nystagmus.

Spinal fluid examination was normal with a protein content of 30 mg/dl. Radiographs of the skull and cervical spine showed no abnormality. Myelography in 1963 was reported as showing 'slight swelling of the cervical cord and abnormal flow of Myodil through the foramen magnum' (G. Thomson). A diagnosis of a cervical cord lesion was made, syringomyelia being the likeliest cause.

Her present condition is much the same. She complains of a burning pain in the right hand present most of the time and which nothing relieves. The right hand still feels weak. She has episodic aching of the right shoulder. On examination, there was no nystagmus and cranial nerves were normal. The right hand showed slight wasting of the first dorsal interosseous muscle, the thenar eminence and the right forearm. There was mild weakness of the right hand movements and of extension and flexion of the wrist and forearm. There was no evidence of weakness or wasting in the left upper limb. Upper limb jerks remained absent but there was no longer any sensory abnormality. Apart from impaired joint position sense in the right toes, the lower limbs were normal.

In summary this patient has a 13 year history which clinically suggests syringomyelia. Normal cervical spine radiographs and typical myelographic findings support this diagnosis. She is not clinically deteriorating and the disappearance of sensory signs suggests minimal improvement.

CASE 4
Mr H.B., the elder brother of Mrs M.L. (case 3) presented in 1955 at the age of 43 years. He complained of numbness of the left side of his face, chest, and left arm of nine years' duration, coming on after wrenching his left arm while lifting a ladder.

On examination, abnormal findings were confined to the nervous system. The pupils were normal and no nystagmus was present. There was absence of pain and temperature sensation over the mandibular and maxillary territory of the left fifth nerve with a diminished corneal reflex on that side. There was a similar spinothalamic sensory loss from C2 to T6 dermatomes on the left and diminution of light touch over the same area. There was no muscle wasting and power was normal in both upper limbs but the left upper limb tendon jerks were absent. The left knee and ankle jerks were exaggerated but the plantar responses were both flexor. A diagnosis of syringomyelia was made clinically. Radiography of the skull and cervical spine were normal. Spinal fluid examination showed no abnormality with a protein content of 10 mg/dl. He was treated with radiotherapy to the cervical spine and over the ensuing 13 years, up to his death, there was minimal improvement in his neurological status. In 1968 he had a thoracotomy for repair of a hiatus hernia and died postoperatively from pneumonia. Unfortunately, at postmortem examination the central nervous system was not examined.

In summary, this patient had a 22 year history of syringomyelia. There was no deterioration in his condition after radiotherapy in 1955.

DISCUSSION
As far as we can determine, for the last 50 years the world literature contains reports of familial syringomyelia only in a brother and sister (Barré and Reys, 1924), in two sisters (van Bogaert, 1934), and once in monozygotic twins (Wild and Behnert, 1964). Syringomyelia has always been recognized as a non-familial condition.

Syringomyelic syndromes have recently been classified into communicating and non-communicating groups (Williams, 1970). The latter group consists of a cystic dilatation of the cord not in communication with the spinal fluid pat-
ways. It has been described after traumatic paraplegia, in association with intramedullary cord tumours, and with spinal arachnoiditis.

In the communicating group CSF pathways communicate with a cyst (syrinx) in the spinal cord. These patients commonly have a persistent communication between the floor of the fourth ventricle and the syrinx by a patent spinal central canal (Gardner et al., 1957; Gardner, 1965). In addition, they are thought to have defective fourth ventricular drainage by the foramen of Magendie. Indeed, Gardner demonstrated this in all of the 74 cases he operated on, and other workers have confirmed this finding (Conway, 1967, Hankinson, 1970). The commonest cause of such an abnormality is a type I Chiari malformation with prolapsed cerebellar tonsils. Less commonly, arachnoiditis, a posterior fossa tumour or cyst is present.

Typically, the Chiari group present as classical syringomyelia with a long history starting in early adult life or before. Symptoms have been aggravated or first noticed after trauma in a very few cases (Foster and Hudgson, 1973). In the majority of cases myelography demonstrates abnormal flow through the foramen magnum, and tonsillar prolapse can be shown. In addition, cervical cord dilatation is often present. In our patients, cases 1 and 3 both had myelograms showing cervical cord swelling. Case 1 showed tonsillar herniation and, although the malformation was not demonstrated in case 3, ‘abnormal flow of Myodil through the foramen magnum’ was reported. Cases 2 and 4 both had classical syringomyelia with long histories and, although myelography was not performed, it seems highly probable that a similar anomaly was present. The fact that these patients had normal cervical spines by no means excludes a Chiari malformation. Foster and Hudgson (1973) had 74 out of 100 syringomyelic patients with normal cervical spines and 47 of these had ectopic cerebellar tonsils on myelography.

The mechanisms producing the syrinx in the communicating group remain in dispute (Gardner, 1965; Williams, 1970). However there can be little doubt that there is a developmental abnormality during embryogenesis in those cases with impaired ventricular drainage due to a Chiari malformation. Evidence that the Chiari type I malformation is familial is difficult to find. However, the type II malformation, the more severe form of the condition (Chiari, 1891), is associated with hydrocephalus and meningomyelocele which have an increased familial incidence. An environmental rather than a genetic factor has been postulated as the cause for this because of the low incidence in monozygotic twins. Arrested hydrocephalus is not uncommon in patients with syringomyelia; Foster and Hudgson (1973) reported seven out of their 100 cases. In addition, syringomyelia occasionally coexists with spina bifida (Gardner, 1973).

In syringomyelia there is an increased incidence of skeletal abnormalities, especially affecting the base of the skull. In particular, Spillane et al. (1957) found basilar impression of the skull in one-third of patients with syringomyelia. This condition appears to be inherited as an irregular dominant, both in patients with and without syringomyelia (Bull et al., 1955), suggesting that there may be a genetic predisposition in some instances of syringomyelia.

In the light of the association of syringomyelia with a number of other familial disorders, as discussed above, the occurrence of familial cases of syringomyelia is perhaps not surprising. It may be much commoner than the literature would suggest. If looked for, further familial cases may well be discovered. This would add further weight to a genetic or environmental element in the aetiology of this condition.

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REFERENCES

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