Syringomyelia and Syringobulbia

Syringomyelia is a rare condition in which there is fluid-filled tubular cyst (syrinx) within the central, usually cervical, spinal cord. The syrinx can elongate, enlarge and expand into the grey and white matter and, as it does so, it compresses the nervous tissue of the corticospinal and spinothalamic tracts and the anterior horn cells. This leads to various neurological symptoms and signs, including pain, paralysis, stiffness and weakness in the back, shoulders and extremities. Syringomyelia may also cause loss of extreme temperature sensation, particularly in the hands, and a cape-like loss of pain and temperature sensation along the back and arms. Symptoms typically vary depending on the extent and location of the syrinx. 50% of patients (when all types of syrinxes are considered) experience no or only mild disability.

If the syrinx extends into the brainstem, syringobulbia results. This may affect one or more cranial nerves, resulting in facial palsies. Sensory and motor nerve pathways may be affected by interruption and/or compression of nerves.

The term 'syrinx' derives from Greek mythology: Syrinx was a nymph transformed into hollow reeds which were later used to make panpipes.

Aetiology

A blockage in the circulation of cerebrospinal fluid (CSF) is the most common cause of syringomyelia. The most common reason for this blockage is the presence of a Chiari malformation. This is a developmental abnormality occurring in the fetus, causing the cerebellum to protrude from its normal position through the foramen magnum and into the cervical portion of the spinal canal. There are two main types of Chiari malformation - type I and type II. Type I has no other associated cause, whereas type II is associated with spina bifida.

- Causes of disruption/blockage of CSF circulation include:
  - Arachnoiditis following infection, inflammation, irradiation or bleeding.
  - Meningeal carcinomatosis.
  - Space-occupying lesions.

- Other common causes include:
  - An association with spina bifida even without Chiari malformation.
  - Post-traumatic syringomyelia - secondary to spinal cord injury: this can occur secondary to trauma, radiation, haemorrhage, ischaemic injury or infection.
  - Spinal cord tumours.
  - Arachnoiditis.
  - Idiopathic syringomyelia.
  - Spinal dysraphism: this is incomplete closure of the neural tube (neural tube defects: it includes spina bifida). Identification and treatment of the dysraphism can arrest progression of syringomyelia.

- Cases of autosomal recessive familial syringomyelia have been reported with an estimated incidence of 2% of all syringomyelia cases.

Associations

- Scoliosis
- Spina bifida
- Klippel-Feil syndrome
Epidemiology

- There is a paucity of data on the prevalence of the condition in the West. The widely quoted prevalence of 8.4 per 100,000 derives from a small study of one UK city, published in 1966. More recent work in the USA aimed at quantifying prevalence suggests a prevalence of 7-70 per 100,000. The authors believe that their upper figure is almost certainly too high and their lower figure too low. In Japan the prevalence is quoted at only 1.9 per 100,000.\(^1\)\(^,\)\(^2\)
- Syringomyelia is more common in men than in women.
- Most syrinxes (44\%) in the Japanese survey were cervico-thoracic, with cervical only being the second most common.\(^1\)
- Chiari I accounts for fewer than half of the cases.
- Trauma accounted for 7.5\%.
- 16\% had no known cause.

Presentation

- Most present in the 20s and 30s but can present in childhood or later life.
- In the Japanese survey, 22\% were asymptomatic, and half of cases were associated with Chiari malformation.\(^1\)
- Among the symptomatic patients, sensory disturbances were the most common, affecting nearly three quarters of the group.
- Motor and autonomic nervous dysfunction were also commonly reported.
- Among patients treated surgically, the average age of symptom onset was around 30 years.
- The primary lesion may have been present since birth but the condition progresses very slowly. Progression of symptoms and deterioration occur over many years.
- As a syrinx widens it compresses and injures further nerve fibres.
- Damage to the spinal cord often leads to progressive weakness in the arms and legs, stiffness in the back, shoulders, arms, or legs, and chronic, severe pain. These can be present at presentation.
- Other symptoms may include headaches, a loss of the ability to feel extremes of hot or cold (especially in the hands), and loss of bladder and other functions.
- Each individual experiences a different combination of symptoms depending on where in the spinal cord the syrinx forms and how far it extends.
- Signs of the disorder tend to develop slowly, although sudden onset may occur with coughing or straining.
- Sudden exacerbations can occur and are thought to be caused by rupture of the syrinx because of raised venous pressure, as seen in sneezing or violent coughing.\(^3\)

Sensory features

- Pain and temperature sensation are lost due to spinothalamic tract damage.
- One side may be affected more than the other.
- Classically, the sensation loss is experienced in a shawl-like distribution over the arms, shoulders and upper body.
- Dysaesthesia (pain experienced when the skin is touched) is common.
- Light touch, vibration and position senses in the feet are affected as the syrinx enlarges into the dorsal columns.

Motor features

- These begin to occur as the syrinx extends and damages the lower motor neurons of the anterior horn cells.
- Muscle wasting and weakness begins in the hands and then affects the forearms and shoulders.
- Tendon reflexes are lost.
- Claw hand may be present.
- There may be respiratory muscle involvement.

Autonomic features

- Bladder, bowel and sexual dysfunction can occur.
- Horner's syndrome may be present.
Syringobulbia
This occurs if the syrinx extends into the medulla of the brainstem. The cranial nerves become affected:
- Facial sensory loss can occur as the trigeminal nerve becomes involved.
- Vestibulocochlear nerve involvement causes vertigo and nystagmus.
- Facial, palatal and laryngeal nerve palsy can occur as the VIIth, IXth, Xth and XIth cranial nerves become involved.
- Weakness and atrophy of the tongue is caused by XIIth nerve involvement.

Other possible features
- Charcot joints: shoulder, elbow or wrist are most commonly affected.
- Scoliosis: presentation can be with scoliosis. It has been suggested that patients due for surgery for correction of scoliosis should have an MRI scan first to exclude syringomyelia.
- Painless ulcers on the hands.
- Extension of the syrinx into the lumbar region and involvement of the legs.
- Respiratory insufficiency can occur: there seems to be a multifactorial aetiology involving muscle weakness, central control and chemoreceptors.

Investigations
- MRI is now primarily used for diagnosis and has significantly increased early detection. MRI will show the syrinx in the spine and may demonstrate a causative condition, such as Chiari malformation or the presence of a tumour. Images taken in rapid succession can be used for 'dynamic imaging' to observe the CSF flow around the cord and within the syrinx.
- CT scanning may reveal the presence of tumours and other abnormalities such as hydrocephalus. CT scanning is better at showing abnormalities of bony spinal canal, whilst MRI scanning is better at showing soft tissue.
- Myelography is now rarely necessary to demonstrate syrinx.
- Plain X-rays may show a widened cervical canal.
- Lumbar puncture is best avoided because of risk of herniation.
- It is important to identify the cause of the syrinx formation, if there is one.

Management
General measures
- Physiotherapy and rehabilitation can help to preserve neurological function.
- The patient must be taught to avoid damage which may result from absence of pain.
- No drugs can retard or reverse the condition.
- Analgesics may be required for pain.

Surgery
- Surgery is usually recommended where there are symptoms.
- In the absence of symptoms, or where there is advanced age and only minimal progression, watching and waiting are usually recommended.
- There are a number of neurosurgical approaches that can be used to retard or halt the progress of the condition. They include insertion of a shunt, laminectomy and syringotomy (drainage of the syrinx). The procedure used will depend on the nature of the lesion.
- In Chiari malformation, surgical decompression at the foramen magnum is performed to promote the free flow of CSF, to provide more space at the base of the skull and neck and to minimise the size of the syrinx. Results are generally good.
- In one study, the majority of participants who underwent posterior fossa surgery for a Chiari type I malformation reported significant improvement in their quality of life afterwards.
- Some treatments aim to drain the syrinx using a syringo-peritoneal shunt. This usually halts symptom progression and relieves pain, headaches and tiredness.
- Where tumour is the cause, treatment usually centres on removing the tumour.
- In trauma-related syringomyelia, the preferred surgical approach is to operate at the level of the injury to expand the space around the cord and decrease fluid volume.
Radiation has little use except where used in the treatment of a tumour, when it may help to prevent syrinx expansion.
Delay in surgical treatment may result in irreversible spinal cord injury.
It may be necessary to repeat surgery.

Complications

As myelopathy progresses, the patient can become wheelchair-bound or confined to bed.
Complications can occur due to the immobility caused by the myelopathy, including recurrent pneumonia, pressure sores and pulmonary embolism.
Paraplegia, quadriplegia and bowel or bladder dysfunction can result from the myelopathy itself.
Respiratory failure can occur and is thought to be secondary to extension of the syrinx into the medulla.\[^3\]

Prognosis

This depends on where the syrinx is, how far it extends, its underlying cause and the resulting neurological disability it causes.
Those with moderate or severe neurological features do worse.
Very rarely, a syrinx may spontaneously regress. \[^7, 8\]
Evidence suggests that early surgical intervention in symptomatic patients improves the outcome. \[^9\]

Further reading & references

2. MSpeer; The Prevalence of SM in the U.S., American Syringomyelia & Chiari Alliance Project, 2009

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