Thoracic outlet syndrome—a diagnosis mislaid for 46 years

Thoracic outlet syndrome (TOS) is often a nebulous condition of the arms that eludes diagnosis as there are no specific symptoms nor sensitive or specific investigations that confirm its existence. Many doctors are unaware of TOS or how to diagnose it, particularly when the symptoms and signs are vague. In rare cases, however, a patient has such obvious symptoms and signs that the diagnosis cannot be refuted. Lauren is one of these cases. She presented with vague arm pains but clear neurological signs of denervation of the T1 root (both the median nerve innervated abductor pollicis brevis and ulnar nerve supplied hand intrinsic muscles were atrophied). This indicated the problem was at the level of the T1 root or lower trunk brachial plexus rather than at the periphery. The differential diagnosis includes carpal and cubital tunnel syndrome or Charcot-Marie Tooth, although Charcot-Marie Tooth is bilateral, symmetrical, and involves the feet. The diagnosis of TOS is made by clinical examination, with provocation of the pain or symptoms by compression of the supraclavicular fossa being the most reliable sign coupled with the lack of localising signs around the carpal and cubital tunnel.

The cause of TOS is anatomical variations or anomalies that cause compression or irritation of the neurovascular structures in the supraclavicular fossa as they pass over the first rib between the scalenes, then under the clavicle. The most common anomaly is a cervical rib and associated musculotendinous anomalies. All these anomalies are not usually identifiable on magnetic resonance imaging or ultrasonography. And neurophysiology is often not diagnostic but will exclude carpal and cubital tunnel syndrome.

Having had little benefit from a structured TOS therapy programme, Lauren underwent brachial plexus exploration with excision of a portion of her first rib. The pain persisted. A further operation was performed after much delay, with greater success. Unfortunately this was short lived and Lauren’s pain rapidly recurred.

On each occasion, the diagnosis of TOS must be checked and other potential peripheral or central causes of symptoms excluded. Having reconfirmed the diagnosis of TOS and in view of considerable loss of function, the only solution is to carry out further procedures to attempt to liberate the nerves and vessels from scar tissue and compression. Her latest procedures attempted to protect the brachial plexus nerves from scar by wrapping them in subcutaneous fat tissue transferred from the infracavicular chest wall.

This case illustrates the difficulty in diagnosis and treatment of TOS. Although significant neurological injury makes the diagnosis easier, the severity and delay, especially when pain is established, make treatment difficult. TOS should be suspected and treated early for optimal chances of success.

Henk Giele

The following is a short speech by a patient with thoracic outlet syndrome:

I can’t even remember the number—nerve conduction studies with needles and electrodes, vascular scans, magnetic resonance imaging, radiography, etc—and everyone seemed to be confused by the results.

Eventually I was told I had a condition called thoracic outlet syndrome and that it was rare, especially in my case as it affected my veins, arteries, and nerves. I was then told that I needed surgery to remove some ribs and fibrous bands to stop the damage to my hand, as the veins were being squashed and affecting blood flow.

I spoke to my doctor and various specialists and I did a lot of research myself, but there was not much information on thoracic outlet syndrome. I did, however, find a good website called PubMed. I also found a surgeon in America whose book I ordered. This explained everything in depth.

I could not believe it; one minute I was at college and working at my mum’s salon enjoying my life and friends and the next I was being told I had to have major surgery for a rare condition. I made sure I worked hard at college and completed the course. It took my mind off things, but I couldn’t ignore the fact that my arm and hand seemed to be deteriorating, and at an alarming speed. I was in constant pain and was now on strong pain medication. My left hand had become “claw-like”—where the muscles were wasting my fingers were being pulled in. I hated this.

It took a long time before I had my first operation. I was told the surgery would not improve the muscle atrophy as the damage had been done, but that the pain would improve. After surgery I was in agony, with the same pains and more; my left eye was also drooping. I was reassured that after a few months things would settle down.

A CLINICIAN’S PERSPECTIVE

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On each occasion, the diagnosis of TOS must be checked and other potential peripheral or central causes of symptoms excluded. Having reconfirmed the diagnosis of TOS and in view of considerable loss of function, the only solution is to carry out further procedures to attempt to liberate the nerves and vessels from scar tissue and compression. Her latest procedures attempted to protect the brachial plexus nerves from scar by wrapping them in subcutaneous fat tissue transferred from the infracavicular chest wall.

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A PATIENT’S JOURNEY

Thoracic outlet syndrome

Lauren Deane, Henk Giele, Karen Johnson

After undergoing sensory tests for numbness in her left arm, this patient was diagnosed as having thoracic outlet syndrome. Despite seven operations, she is still in pain.

It all started around nine years ago when I was 17; I began to get a strange tingling and a dull ache in my left arm. I noticed I couldn’t carry a bag over my left shoulder, as my arm seemed to go numb and achy. At the time I was training at college to be a beauty therapist. I kept getting strange electric shock sensations and stabbing pains and the feeling of a dead heavy arm, but I remembered putting it down to over-working at college and at work. I ignored it for as long as possible. I mentioned it ber putting it down to over-working at college and at work. I mentioned it to doctors. The BMJ welcomes contributions to the series. Please contact Peter Lapsley (plapsley@bmj.com) for guidance.

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This is one of a series of occasional articles by patients about their experiences that offer lessons to doctors. The BMJ welcomes contributions to the series. Please contact Peter Lapsley (plapsley@bmj.com) for guidance.

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Previous articles in this series

Lessons from patients’ journeys (BMJ 2013;346:f1988)
Klinefelter’s syndrome—a diagnosis mislaid for 46 years (BMJ 2012;345:e6938)
Kallmann syndrome (BMJ 2012;345:e6971)
Non-coeliac gluten sensitivity (BMJ 2012;345:e7982)
Restless legs syndrome (BMJ 2012;345:e7592)

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Around six months later I still had the pains. In addition, symptoms were developing on my right side. I went to see my surgeon but she was quite dismissive. I was worried about the same problem developing on the right side so spoke with my doctor who referred me back to the surgeon. Tests detected similar problems on the right side.

I had to have the same surgery on the right side. My left side still didn’t feel right though, despite my surgeon insisting it was, so I carried on researching for myself and with help from my mum we emailed a specialist in the United States. He thought I should seek a second opinion. At this point my left hand had become a lot worse and my fingers were shrinking. I couldn’t keep on any of my rings. I could barely attend to my own cosmetic needs let alone do it as a career all day every day.

The second specialist said I would have to have more surgery on my left side as soon as possible. I stayed in hospital for a week. It took months to recover and I was in excruciating pain, but after a while things seemed a bit better. Unfortunately my right hand had deteriorated and I underwent a second operation. Although surgery helped the right arm, my left arm did not improve and my specialist wanted me to see a consultant in Oxford.

This specialist too seemed concerned and decided that I needed further surgery as something was definitely compressing my left side. After a fifth operation I remained in a lot of pain. Despite this I continued to try and push forward. My life had been changed and with that went my career in beauty and I eventually enrolled in a counselling course and managed to qualify as a counsellor. During my studies I was told that I needed my left side operated on again as the surgery had not worked. I was devastated by the prospect of more surgery but I did manage to finish my course first.

I have since had my sixth and seventh surgeries. The sixth was on my left side in September 2011 and the seventh on my right side in March 2012. I have now had four major surgeries on my left side and three on my right side. I have had another two ribs out, lots of scar tissue removed, tissue attached to my chest wall, and part of my lung taken away (I also had reconstructive chest surgery and the main nerve from the spine to arm cut and reattached on the left side, and the nerve straightened on the right side). I have been told that for at least two years I will need to wear splints on both hands and arms. I am hoping that the last two procedures will make a difference to my daily pain levels and disability.

Both my hands are clawed. I cannot work any more and I struggle on a daily basis with even the simplest things such as opening a can, cleaning, and even dressing and sleeping. I cannot socialise to the same extent as a typical 26 year old as I get tired from the pain. I rely on my partner, family, and friends to help me each day. The privilege of doing everyday tasks is not realised until it’s taken away. As I am still in so much pain daily, I am on a lot of medication. I save up money every month and pay privately for acupuncture and sports massage.

This condition affects every part of my life. Even though I try to stay positive and active, it is hard. I still hope that I can pursue a career in counselling.

My surgeon and doctor as well as numerous other consultants have told me that this is the worst and most complicated case of thoracic outlet syndrome they have ever seen. Even though the condition has altered my life and nearly everything in it I will not be beaten by it. Thoracic outlet syndrome is a part of my life; I am not a part of its life.

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10-MINUTE CONSULTATION

Hearing loss in adults

Rachel Edmiston,1 Caroline Mitchell2

A 60 year old former steelworker mentions to his general practitioner that his family is complaining that the television is too loud.

What you should cover

Hearing difficulties
• Onset of symptoms: acute, chronic, fluctuating, or recurrent?
• Unilateral v bilateral?
• What impact is there on day to day communication (for example, hearing in groups or one to one)?

Associated ear, nose, and throat (ENT) symptoms
• Vertigo: described as a sensation of dizziness likened to “room spin” associated with or without nausea
• Otorrhoea: is it purulent or clear?
• Tinnitus
• Otalgia: otitis media or externa (is there any associated itching or discharge?)
• Head and neck: localised pain, swelling, lump
• Nasal obstruction
• Epistaxis

Risk factors for otological disease
• Infection (adult or childhood), trauma, or previous surgery
• History of exposure to noise (including occupational)
• Use of ototoxic drugs: permanent damage from aminoglycosides (such as gentamicin) or chemotherapy drugs (particularly platinum based treatments such as cisplatin); reversible damage from

USEFUL RESOURCES FOR PATIENTS AND HEALTH PROFESSIONALS

Patient.co.uk (www.patient.co.uk) medical conditions including thoracic outlet syndrome
NINDS Thoracic Outlet Syndrome Information Page (www.ninds.nih.gov/disorders/thoracic/thoracic.htm)—An American website with information about thoracic outlet syndrome, including recent research and links to other related sites

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This is part of a series of occasional articles on common problems in primary care. The BMJ welcomes contributions from GPs.
Bone conduction is heard better than air.

**Sensorineural deafness:** sound lateralises to the affected ear.

Conductive deafness: sound lateralises to the affected ear.

Air conduction is heard better than bone conduction.

**History of pain or bleeding from the ear**
History of head trauma: exclude temporal bone fracture or ossicular discontinuity

**History of Paget’s disease**
Early treatment of disease can reduce degree of hearing loss.

**Early treatment of disease can reduce degree of hearing loss**

**Referrals**

**Urgent referral to ENT**—Sudden sensorineural hearing loss, middle ear effusion, focal neurology with cholesteatoma, malignant otitis externa

Routine referral to ENT—Unilateral hearing loss, asymmetrical hearing loss, discharging ears, cholesteatoma, disabling Meniere’s, otosclerosis, or persistent perforations.

**Referral to audiology**—Presbycusis or noise induced hearing loss

**Referral for “assess and fit” appointment**—Patients aged 50–80 years with no otological disease if they would consider using hearing aids

**Box 1 | Red flags**

- **Sudden onset or rapidly progressive hearing loss**
  A rapid onset (over a 72 hour period) of a sensation of hearing impairment in one or both ears is a medical emergency and requires urgent referral to exclude acoustic neuroma.

- **Asymmetric hearing loss (with or without tinnitus)**
  Exclude acoustic neuroma

- **History of pain or bleeding from the ear**
  History of head trauma: exclude temporal bone fracture or ossicular discontinuity

- **Referred otalgia:** occurs commonly with head and neck malignancies

- **History of Paget’s disease**
  Early treatment of disease can reduce degree of hearing loss

**Box 2 | How to do an initial hearing assessment**

**Whispered voice test**
- Stand at arm’s length (0.6 m) behind the patient and whisper a combination of numbers and letters (for example, 7 F 9). Ask the patient to repeat these
- If a correct response is given, hearing is considered normal; if incorrect, repeat the test with a different letter and number combination
- The patient is considered to have passed the screening test if they repeat at least three out of a possible six letter and number combinations correctly
- Test each ear individually while gently occluding the contralateral ear. The examiner should exhale completely before testing to ensure as quiet a voice as possible

**Tuning fork tests:** 512 Hz
- **Rinne test**
  - Air conduction is heard better than bone conduction (“Rinne positive”) = sensorineural deafness or normal hearing
  - Bone conduction is heard better than air conduction (“Rinne negative”) = conductive deafness
  - Use masking of untested ear to prevent false negatives
- **Weber test**
  - Conductive deafness: sound lateralises to the affected ear
  - Sensorineural deafness: sound lateralises to the unaffected ear

**Red flags**
- Be aware of the red flag symptoms and signs (box 1)

**What you should do**

**Examination**
Examine for external ear changes, discharge, wax, or anomalies in the tympanic membrane. If wax, discharge, or debris obscure complete visualisation of the membrane, removal of wax or aural toilet will be necessary. For impacted wax, advise the patient to use olive oil drops for one week and arrange ear canal irrigation (provided that there is no perforation of the tympanic membrane or history of mastoid surgery). Reassess after irrigation.

Look out for a retracted tympanic membrane, which often results from repeated infections and may lead to ossicle erosion, perforation, and formation of cholesteatoma. If a retracted tympanic membrane is associated with hearing loss or persistent otorrhoea refer for further assessment.

Assess hearing using the whisper test to help gauge the level of hearing loss, and perform the 512 Hz tuning fork tests (box 2). These tests are not 100% reliable but are helpful to guide diagnosis.

Perform a focused cranial nerve examination to exclude a central cause. A lesion on cranial nerve VIII may also affect cranial nerves V and VII, leading to altered facial sensation, facial muscle weakness, and altered taste sensation.

Examine the head and neck for lumps and lymph nodes.

**USEFUL RESOURCES**

For patients
- [www.earcarecentre.com](http://www.earcarecentre.com)—This website of Rotherham Primary Ear Care and Audiology Services (part of the Rotherham NHS Foundation Trust) provides tips on ear care and use of hearing aids and also gives useful links
- [www.deafnessresearch.org.uk](http://www.deafnessresearch.org.uk)—Deafness Research (now merged with Action on Hearing Loss) gives excellent patient resources on common conditions and treatments
- [www.patient.co.uk/doctor/Deafness-in-Adults.htm](http://www.patient.co.uk/doctor/Deafness-in-Adults.htm)—A useful resource clarifying hearing tests including hearing thresholds and the whisper test.

For professionals
- [www.patient.co.uk/doctor/Deafness-in-Adults.htm](http://www.patient.co.uk/doctor/Deafness-in-Adults.htm)—Offers useful tips for diagnosis, management, and referral guidance
- [www.patient.co.uk/doctor/Hearing-Tests.htm](http://www.patient.co.uk/doctor/Hearing-Tests.htm)—A useful resource clarifying hearing tests including hearing thresholds and the whisper test.

**References**

- [BMJ 2013;346:e5743](http://www.bmj.com/content/346/7899/e5743)
- [BMJ 2012;345:e6824](http://www.bmj.com/content/345/7899/e6824)
- [BMJ 2012;345:e7533](http://www.bmj.com/content/345/7899/e7533)
- [BMJ 2013;346:f985](http://www.bmj.com/content/346/f985)

usually heal spontaneously (keep the ear dry); perforation owing to chronic suppurrative otitis media often requires surgical repair (referral required if cholesteatoma is present (symptom: persistent otorrhoea))

- Middle ear effusion (tympanic membrane dull with a yellow tinge)—Effusion is rare in adults; rule out sinusitis and refer for nasoendoscopy to exclude obstruction of the eustachian tube with a nasopharyngeal tumour

**Conductive loss (bilateral)**

Otosclerosis commonly presents with gradual deafness with or without tinnitus. The tympanic membrane may be normal.

**Sensorineural loss (bilateral and gradual)**

Bilateral and gradual sensorineural hearing loss is often associated with a normal tympanic membrane.

Age related presbycusis is the commonest type of sensorineural hearing loss in older adults (audiogram shows symmetrical bilateral loss at high frequencies). With a history of noise exposure consider noise induced hearing loss, which shows on an audiogram as a classic “notch” at around 4000 Hz.

**Sensorineural loss (unilateral)**

Meniere’s disease presents with fluctuating hearing loss associated with episodic vertigo, tinnitus, and a sensation of pressure in the ear (aural fullness).

Always consider acoustic neuroma with unilateral sensorineural hearing loss. Commonly the hearing loss is progressive with associated tinnitus; rarely acoustic neuromas may be bilateral.

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ANSWERS TO ENDGAMES, p 38 For long answers go to the Education channel on bmj.com

**PICTURE QUIZ**

“It’s just a muscle sprain”

1. The four week history of intermittent pain with night symptoms in the absence of a history of trauma is worrying.
2. Examination of the spine and knee is useful to exclude more proximal and distal disease, respectively. The presence or absence of a mass, joint effusion, swelling, and skin rashes can also help in the diagnosis.
3. Yes. An initial radiograph is important to exclude sinister causes of pain, and radiography of the extremities exposes patients to relatively little radiation. Modern radiation machines expose patients to less than 1.5 days of background radiation on imaging of the extremities, so femoral radiography is a safe, inexpensive, and vital way to excluding serious disease.
4. A mixed lytic and blastic aggressive tumour within and extending out from the distal diaphysis of the femur, with an associated soft tissue mass and distal mineralised skip lesion (figs 3 and 4).
5. Urgent referral to a supraregional bone sarcoma unit, with subsequent prompt and local magnetic resonance imaging of the lesion and entire bone.

**STATISTICAL QUESTION**

**Generalisation and extrapolation of study results**

Statement a would be justified, whereas b and c would not.

**Fig 3** | Anterioposterior radiograph of the distal femur showing new expansile bone formation (white arrow) with overlying soft tissue mass; periosteoal reaction, with the deposition of Codman’s triangles between the undersurface of the periosteum and outer host bony cortex (black arrow); indistinct lesionai border, with a wide zone of transition; and a distal “skip” lesion (double white arrowhead)

**Fig 4** | Lateral radiograph of the midshaft femur showing new expansile bone formation (white arrow) with overlying soft tissue mass; periosteal reaction, with the deposition of Codman’s triangles between the undersurface of the periosteum and outer host bony cortex (black arrow); indistinct lesionai border, with a wide zone of transition; and a distal “skip” lesion (double white arrowhead)