Congenital anomalies occurring with OA/TOF

Amber, an adult born with OA/TOF

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lifelong support for those born unable to swallow

This booklet is produced by TOFS, the charity that provides information to increase knowledge, encourage self-advocacy, promote wellness and improve health outcomes.

Some infants born with Oesophageal Atresia/Tracheo-Oesophageal Fistula (OA/TOF) may have additional congenital anomalies. Units treating babies with suspected VACTERL should use a screening tool at birth, so all defects are checked for and nothing is missed.

These may be classified within the VACTERL Association or be recorded as stand-alone malformations. In medical terms, an 'association' is a group of symptoms that appear together more often than would be expected by chance.[4] Many of the conditions within the VACTERL Association will be dealt with within the first year of life.

This booklet:

- Provides information for those born with these rare and complex conditions.
- Seeks to assist healthcare providers understand the additional malformations and how they may impact each other.
- Recognises the need for a high-quality transition from children's services to adult services.
- Identifies the lifelong nature of some congenital anomalies and how individuals are not always fixed at the initial surgery.
- Endorses the need for multi-disciplinary, collaborative care pathways to improve health outcomes and quality of life.

VACTERL Association:

Occurs in around 1: 10,000 - 1: 40,000 of the population;[1] it is an acronym for a collection of anatomical differences which have occurred in the very early stages of the fetal development.

People diagnosed with VACTERL association typically have at least three of these characteristic features.[2]

More people with these birth defects are surviving into adulthood; no two individuals are likely to be affected in the same way. Some will have health problems, including new diagnoses as a result of their congenital malformations.[3]

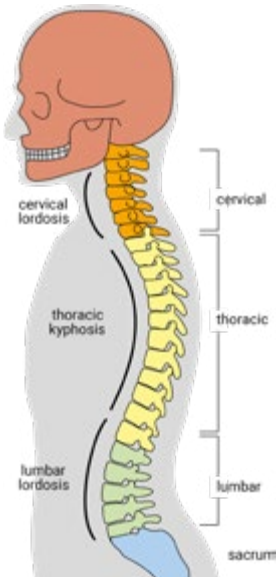
It is important to recognise that some adults with VACTERL Association are at a high surgical risk due to airway, vertebral, cardiac and other organ anomalies. They need careful assessment by anaesthetists, obstetricians and other healthcare teams prior to investigations, treatments and surgery to ensure optimum health outcomes.

VERTEBRAL (V)

Vertebrae are flat bones which are stacked on top of each other to form the spine. Vertebral abnormalities are defects of the spinal column. Between 60-80% of babies diagnosed with VACTERL Association are, in varying degrees, affected by spinal problems.[4]

The vertebrae may have a different shape than normal; hemivertebrae, butterfly shaped.

Fused vertebrae, missing vertebrae or additional vertebrae may be present. These defects can exist alongside missing or extra ribs, curvature of the spine (scoliosis). Deformities of the lower back can cause nerve fibres to be “pinched”, which can result in problems with mobility, bladder and bowel function.



Signs and symptoms:	Possible causes:
Pain: Shoulder, elbow, hip.	Bursitis - an inflammatory condition. Osteoarthritis.
Tendon stiffness, tightness and pain around a joint.	Tendonitis - inflammation of a tendon.
New or worsening back pain.	Scoliosis may cause discomfort/pain.
Rib cage misaligned, shoulder blade prominence.	Scoliosis - when the spine curves to the side. (Early splinting and physiotherapy may delay the onset and severity of scoliosis).
Weakness and/or numbness in legs. Difficulties controlling bladder and/or bowel.	Tethered cord - a congenital anomaly where there is abnormal attachment of the spinal cord to the spinal canal. This is rarely seen in adults and would require urgent attention from a neurosurgeon.

Suggested referrals:
Specialist spinal surgeon for correction of the scoliosis should be considered.
Neurosurgeon: Tethering of the cord causing urinary retention or incontinence requires urgent attention to avoid long term damage.
Orthopaedic Specialist; Physiotherapist, Orthotist, Pain Management Team, Continence Advisor.

ANORECTAL (A)

Anorectal malformation is an umbrella term used to describe birth defects which affect the lower end of the digestive tract. Abnormalities may include imperforate anus, anal stenosis, a fistula (connection) between the rectum to the urinary tract or the reproductive system. This affects between 60-90% of infants born with VACTERL Association.[4]

These anomalies can affect bowel, bladder, kidneys, reproductive systems, sacrum and spinal cord. Anorectal malformation varies from minor anomalies (imperforate anus) to major malformations such as recto-urinary fistula in males to cloaca anomaly in females.

Anorectal malformations can have a profound impact on pelvic floor nerve and muscle functioning with lifelong consequences. Incontinence can lead to psychological challenges which can impact on all aspects of life.

Signs and symptoms:	Possible causes:
Abdominal pain, cramps, diarrhoea.	Constipation - faecal incontinence.[5]
Reduced appetite.	Fear of faecal urgency/incontinence.
Rectal bleeding.	Rectal prolapse - when the rectum slides out through the anus.
Recurrent urinary infections.	Kidney problems linked with anorectal malformation.
Concerns re: sexual health and fertility.	Sexual function problems.[6] Low sperm count. Painful intercourse.[7]
Anxiety, depression.	Faecal and or urinary incontinence can result in psychological challenges affecting the child, the adult and the family.

Suggested referrals:
Specialists: Colorectal, Uro-gynaecology, Andrology.
Long term problems such as faecal incontinence or intractable constipation will require a bowel management programme involving medications and bowel washout.

CARDIAC (C)

Between 40-80% of babies diagnosed with VACTERL have a heart condition, (Congenital Heart Disease - CHD).[4]

The most common heart defect occurring in VACTERL Association is a Ventricular Septal Defect (VSD); this is a hole between the lower chambers of the heart. Sometimes this can close spontaneously, at other times, an operation is required.

Additional heart anomalies range from easily correctable defects such as PDA, ASD and VSD to major anomalies such as hypoplastic heart syndrome, transposition of the great vessels, valvular defects and Tetralogy of Fallot.

Adult patients may present with significant cardiac issues which impact on their quality of life.

Signs and symptoms:	Possible causes:
Shortness of breath, lack of energy. Reduced exercise tolerance.	Heart is working unduly hard.
Palpitations or heart beating rapidly.	Irregular or abnormal heartbeats.
Swollen feet (oedema).	Heart struggles to pump blood around the body as well as it should.

Suggested referrals:
Referral to Cardiology Specialist, Heart investigations: ECG, Chest X-Ray. Blood tests for kidney and liver function.

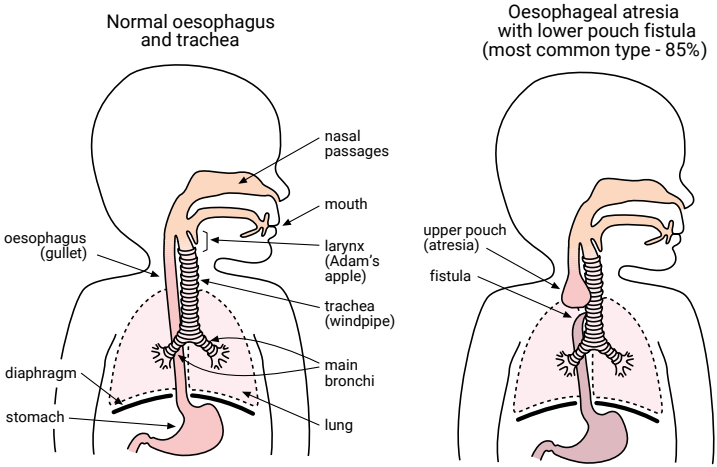
Tracheo-Oesophageal Fistula (T)

Tracheo-oesophageal Fistula and/or Oesophageal Atresia (OA/TOF) occur when there is an abnormal connection between the trachea and the oesophagus and there is a gap in the length of the oesophagus: this is the most common malformation in VACTERL Association.

There are five classifications or types of OA/TOF, each detailing where the trachea connects with the oesophagus and where the oesophagus narrows or ends in a blind pouch. Surgery to repair the fistula and bring together both ends of the oesophagus is needed to prevent chest infections, pneumonia and to enable the infant to thrive. Some babies require staged surgery, this depends on the length of the oesophageal gap. (This means the repair requires more than one operation.) Many of the health issues faced in childhood and later in life result from the interplay of the respiratory and digestive tract defects.[8]

Signs and symptoms:	Possible causes:
Dysphagia, a feeling of food getting stuck, regurgitation.	Oesophageal narrowing also known as stricture can occur at the site of anastomosis. Dysmotility - foods and liquid do not pass easily down the oesophagus.
Respiratory and ENT symptoms. Shortness of breath, cough.	GORD (Gastro Oesophageal Reflux Disease) - where acid from the stomach leaks backwards - into the oesophagus. Recurrent fistula. Restrictive, obstructive or mixed lung problem.
Chest/neck pain.	Oesophageal spasm - this is a sharp clenching pain which can make swallowing difficult.
Worsening of reflux symptoms.	Oesophageal stricture, oesophagitis. Barrett's Oesophagus: in some adults cells in the lining of the oesophagus have started to change.

Signs and symptoms:	Possible causes:
Dizziness, diarrhoea, increased perspiration.	Dumping Syndrome (fluctuating blood glucose levels).
Iron deficiency anaemia.	Proton Pump Inhibitors (PPIs) used in the treatment of GORD may interfere with the absorption of vitamin B12 which can lead to iron deficiency anaemia. [9]



Suggested referrals:

Respiratory and Gastroenterology Specialists. Proton pump inhibitors (PPIs) may be prescribed.
Annual influenza vaccination recommended.
Antibiotic threshold should be lower if there are worsening symptoms.
Consider preventative antibiotics and chest/airway clearance physiotherapy.
Iron infusions may be necessary in those who cannot tolerate or absorb iron orally.
Healthy eating and smoking cessation should be encouraged.
Monitoring Peak Flow - knowing personal "norm" encouraged.
Devices to help inhaled medications reach the lungs, for example an AeroChamber®

For further information on Tracheo-Oesophageal Fistula visit tofs.org.uk or scan the QR code:

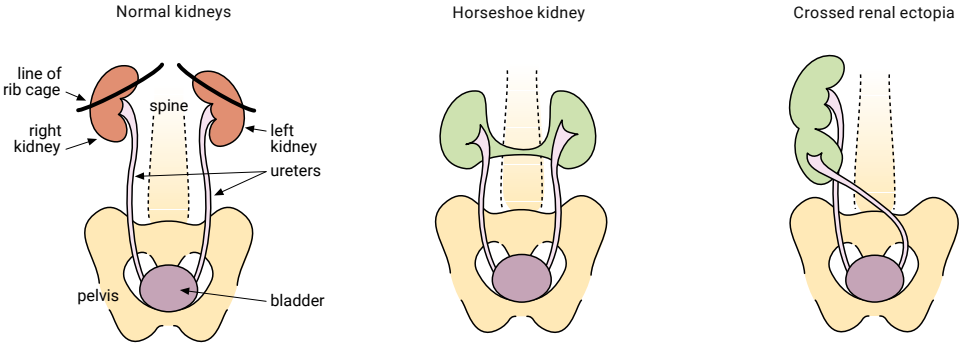
RENAL (R)

Around 60% of babies diagnosed with VACTERL association have some form of renal impairment.[10] Not all anomalies will cause problems immediately after birth.

A wide variety of abnormalities may be present and may affect the kidneys and urinary tract. These abnormalities can vary in severity and include absent kidney(s), polycystic kidneys, vesicoureteral reflux, neuropathic bladder and more. Treatment options will vary depending on the type and degree of abnormality diagnosed. Investigations and monitoring is essential to preserve renal function.[11]

Signs and symptoms:	Possible causes:
Recurrent urinary tract infections.	Vesicoureteral reflux (VUR) where urine flows backward from the bladder to one or both ureters and sometimes to the kidneys.
Kidney problems.	Polycystic kidney(s) - Clusters of cysts within the kidney. Horseshoe Kidney - when the kidneys are attached at the base.
High blood pressure.	Kidney disease.

Suggested referrals:
Urologist/Nephrology Specialist. Early reporting of urinary symptoms encouraged, persistent and/or recurrent infections referral for further investigations. Health surveillance - monitoring blood pressure and renal function.



LIMB (L)

Up to 70% of infants diagnosed with VACTERL also have limb abnormalities.[12]

These may include fusion of the two forearm bones, under development or absence of the radius, under development or absence of thumb(s), the presence of an extra bone in a thumb, extra digits or webbing of the digits.

Surgery may be necessary to improve limb function and appearance. This is complex surgery and requires a multidisciplinary approach from highly specialised surgeons, physiotherapists and occupational therapists.

Signs and symptoms:	Possible causes:
Chronic pain - Compensatory use of other joints.[13]	Osteoarthritis.
Reduced function.	Injury to affected limb.
Anxiety/depression.	Mental health challenges related to disability.

Suggested referrals:
Orthopaedic Specialist team including Occupational Therapist, Physiotherapist.
Advice re: chronic issues related to limb abnormality, pain management, employment and challenges to daily activities and quality of life.

Dental care

- Severe health issues in early life can disrupt the formation of tooth enamel with problems most evident in the molars.
- Progressive loss of enamel can occur due to erosion associated with gastroesophageal reflux disease; this can lead to cosmetic changes, chipping, sensitivity and, in some cases, death of the nerve and possible dental infections.[8]
- Oral sensitivity and fear of dental care may stem from medical interventions in early childhood - tube feeding, oral suction, intubation, etc.
- Association with traumatic experiences can lead to avoiding the dentist, which can result in poor mouth and teeth care, reduced opportunity for surveillance and preventative treatments.
- Good dental hygiene is important; including minimising acid damage to the teeth.
- Avoid tooth brushing for up to 30 minutes after an acidic drink.
- Avoid scrubbing with the toothbrush and use a fluoride toothpaste. Maintaining good oral hygiene is particularly important for those with cardiac problems.[14]

Pregnancy

- Pre-conceptual care should be available; referral to a specialised genetic counselling service, if appropriate (visit tofs.org.uk/genetics for more info).
- Maternal anxiety may be heightened; mental health may be fragile.
- Additional reassurance and support may be necessary, with access to GPs, midwives, obstetricians to allay concerns.
- Detailed scans to check infant growth and development.
- Indigestion and reflux may be worsened due to hormonal changes.
- Epidural advice should be sought (in patients with Vertebral issues, for example Scoliosis). [15]
- Obstetric management to preserve pelvic floor in women born with anorectal malformation. Predelivery evaluations and consider C-Section delivery.[16] Caesarean delivery is usually recommended in mothers who were treated for anorectal malformation in the newborn period.
- Appropriate aids to support the mother's limbs during labour (in patients with limb issues).
- Increased monitoring of maternal cardiac and renal function (in patients with cardiac issues).

Enhancing well-being

There are a number of interventions and therapies which may be beneficial to adults who face long term complications of OA/TOF VACTERL.

A Care-Coordinator can be invaluable, ensuring the person's changing health needs are identified and, wherever possible, met.

Some therapies are available through the NHS:

Physiotherapy, Occupational Therapy, Speech and Language Therapy, Counselling or Psychology (known as conventional therapies).

Complementary therapies are likely to incur a cost. Complementary therapies may be used alongside conventional medicines and treatments.

It is recommended that you speak to your GP before choosing a complementary therapy. Some therapies may be helpful for some people but managing symptoms with complementary therapies may delay the treatment of significant illness and, as such, can be harmful.

Physiotherapy Assessment:

Physiotherapists often specialise in different clinical areas of practice. A musculoskeletal physiotherapist may help with pain or mobility issues, whereas a cardiac rehabilitation or respiratory physiotherapist would assist with heart and breathing difficulties.

A physiotherapy assessment may include patient education and advice to restore movement and function:

- Therapeutic manipulation - soft tissue massage.
- Ultrasound may improve circulation and reduce pain.[17]
- Breathing exercises or hands-on therapy for respiratory problems to help with clearing lungs.
- Advice and education on exercises to help with stamina and reduce stiffness and pain.
- Provision of walking aids and other mobility equipment to improve physical fitness and enhance well-being.

Enhancing well-being (cont.)

Occupational Therapy

For adults living with the long-term effects of OA/TOF and VACTERL, occupational therapists may be able to help with a range of things including:

- **Managing fatigue:**

  Energy conservation techniques and advice on pacing daily activities.

- **Vocational advice and support:**

  Educational courses/work experience opportunities. Accessing college/ workplace vocational guidance to delay retirement.

- **Environmental adaptations:**

  Advice on (and sometimes provision of) equipment to help with carrying out activities around the house.

- **Liaison with employers**:

  Workstation assessments and adjustments to working practices.

- **Mental Health:**

  Wellbeing support including stress and anxiety management.

- **Sensory impacts due to developmental trauma:**

  Some Occupational Therapists specialise in helping people who struggle with daily activities because of sensory processing difficulties. These can manifest as a range of problems, which are often accepted as just 'normal for me'. These can include feeling overwhelmed and panicky when going into crowded places, difficulty with planning ahead or staying focused on a task, problems going into lifts or on escalators, sensitivity to touch, noise or to smells and tastes that other people don't seem to notice.

Enhancing well-being (cont.)

Emotional Support[18]

- Helping a person to adapt to living with health difficulties and improve mental health.
- Transition to adult services requires targeted, holistic support to address educational, vocational, and psychosocial needs. [19/20]
- Whilst many adjust well to living with OA/TOF, others experience emotional distress, anxiety and depression.
- Risk factors for developing post-traumatic stress, anxiety, depression and impaired quality of life include exposure to medical trauma; complex or complicated OA/TOF; swallowing and eating difficulties; respiratory symptoms; and dissatisfaction with surgical scars.
- Promoting resilience and recovery - Self-help tools can be helpful in promoting psychological well-being and coping with anxiety during medical procedures or eating in public.
- Psychologists can provide assessment and treatment for anxiety around health difficulties; depression and adjustment problems; eating related problems; and symptoms of post-traumatic stress.

Clinical Pilates and Yoga[21]

- Promoting strength, flexibility, muscle strength, control, balance and pelvic floor strengthening.
- Encouraging intercostal breathing - improved exchange of oxygen and carbon dioxide.

Mindfulness Meditation

- Taking notice of your thoughts, feelings and body sensations.[22]

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About TOFS

This booklet is produced by TOFS.

It has been peer-reviewed by the charity's medical patrons and members of our Medical Advisory Group. TOFS would like to thank them for their guidance and contributions.

TOFS is the charity dedicated to offering lifelong support to those born with Oesophageal Atresia (OA), Tracheo-Oesophageal Fistula (TOF) and associated conditions.

Our vision is for anyone born with OA/TOF to be able to live life unlimited.

Our position paper entitled: "Towards a Holistic Model for the treatment of those born with Oesophageal Atresia (OA) from diagnosis to transition and adult care" is written from a patient perspective, reflecting on patients as "experts by experience". The charity endorses the author's proposal of the provision of centralised services to enable children and adults to achieve the highest standard of care. so available is our "Adult OA/TOF Management Handbook", a reference guide for health professionals treating

Find out more

Scan to read our position paper in full.

Scan to read the Adult OA/TOF Management Handbook.

Scan for more information about OA/TOF

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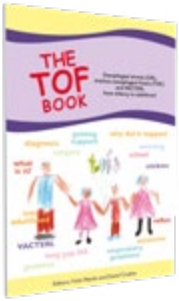
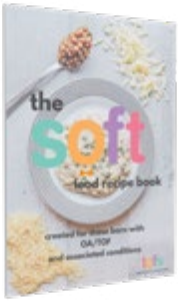
tofs.org.uk/oa-tof-information/resources/vacterl





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For more information about OA/TOF or to become a member of TOFS, please visit tofs.org.uk

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