Always shine brightly. Don't let anyone dim your light

This is the second of a four-part Patient Safety Group series looking at patient perspectives of surgical care

I was born in 1957, at a time when neonatology, diagnostic tools, and long-term surgical follow-up were still in their infancy.

Fortunately, an obstetrician recognised that my difficulties needed urgent attention and transferred me to Birmingham Children's Hospital. There I was diagnosed with a tracheoesophageal fistula (TOF), oesophageal atresia (OA), and an anorectal malformation (ARM).

My first surgical procedure took place within hours of birth. However, at five days old I developed acute intestinal obstruction and underwent a second operation. A necrotic volvulus was discovered, and bowel resection and formation of a colostomy were undertaken.

My stoma was reversed at 2 years at the same time my extra thumb was removed.

Ruled by Quiet Fears

Childhood was marked by years of constipation and bowel washouts. Medications often caused cramps and overflow, severe impactions required manual evacuations under anaesthesia; physically traumatic and emotionally scarring.

My repaired TOF left me with swallowing and breathing difficulties. Food became lodged in my oesophagus, my TOF cough drew unwanted attention. Doctors said I was "fixed," when problems arose, the blame fell on me:

"Chew properly," they said when food became stuck;

I worried, would I soil myself? Did I smell?

"Stop being naughty," when I soiled myself.

My complex conditions have challenged me throughout life: suture abscesses appearing years after surgery.

In my teens discussions about a second colostomy and later decisions to avoid further urogenital reconstruction for fear of harming my fragile but functional anatomy.

Gynaecological investigations revealed further abnormalities which provided the cause for my infertility.

Hospital admission for a suspected ovarian cyst revealed two litres of fluid in my fallopian tube, along with a tendency toward urinary retention requiring catheterisation.

More recently rectal prolapse prompted my first defecating proctogram MRI.

The radiographers were surprised by my anatomy and frustrated when I couldn't hold the contrast in my rectum. Fortunately the imaging revealed my abnormalities and guided the colorectal team's care.

Invisible Burden of Rare Conditions

Adult survivors of OA/TOF face a broad range of ongoing risks that are frequently unrecognised, as shown below.

Respiratory

- Persistent "TOF cough"
- Silent aspiration
- Recurrent chest and sinus infections
- Tracheomalacia and bronchomalacia
- Bronchiectasis
- Vocal cord dysfunction, hoarseness
- Asthma-like symptoms
- Exercise intolerance due to compromised airways

Oesophageal and Digestive

- Food bolus obstruction
- Oesophageal strictures requiring repeated dilatations
- Poor or absent oesophageal motility
- Chronic gastro-oesophageal reflux disease
- Barrett's oesophagus
- Dumping syndrome
- Eosinophilic oesophagitis
- Oesophageal spasm
- Anastomotic complications
- Gastroparesis
- Dysphagia

Nutrition

- Difficulty maintaining weight or absorbing adequate nutrition
- Aversion to textures or limited diet variety
- Requirement for fundoplication or redo procedures
- Long-term enteral feeding (PEG/jejunal tube)
- Parenteral nutrition for some adults

ENT and Upper Airway

- Chronic rhinitis and sinusitis
- Middle-ear infections; reflux reaching the nasopharynx
- Laryngeal clefts

Musculoskeletal

- Chest-wall deformities
- Thoracic nerve or musculoskeletal pain
- Scoliosis related to thoracotomy in childhood

Quality-of-Life

- Anxiety around eating and fear of choking
- Food avoidance and social withdrawal
- Psychological trauma from repeated medical interventions
- Body-image issues related to scars or feeding tubes
- Chronic pain and fatigue impacting daily life

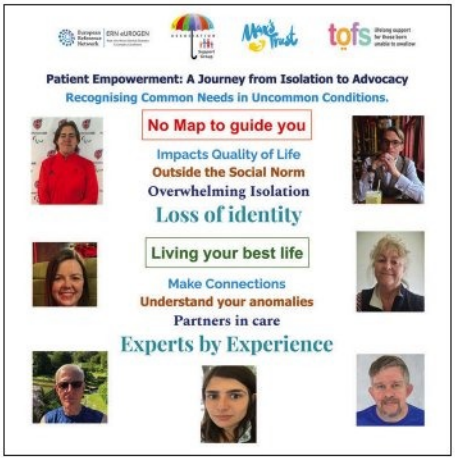
Life on the Edge. Managing the Unpredicatable

Living with an ARM means managing unpredictable bowel issues, constipation, leakage, and sudden evacuations. These affect both comfort and emotional wellbeing.

Strategies including diet, medication, irrigation, or surgery can help.

With preparation, resilience, and self-compassion, adults can regain control, reduce accidents and improve quality of life.

Kate Tyler
Adult born with VACTERL Association; General and Paediatric Nurse (retired)



Urological and Reproductive Care

Lifelong urological care is essential. Children born with an ARM have higher rates of kidney and urinary issues, and the specific type of malformation greatly influences their long-term risk.

- **Males:** may experience epididymo-orchitis, erectile dysfunction, ejaculatory disorders, decreased libido, painful ejaculation and fertility issues.
- **Females:** may have urinary and reproductive anomalies; ectopic ureters, duplicated or septate vaginas, absent or underdeveloped uterus, or a shortened urethra, affecting menstruation, fertility, pregnancy, and labour. Sexual dysfunction and psychological challenges are common.

Spinal Anomalies

Some children with an ARM also have spinal abnormalities, such as tethered cord syndrome or sacral agenesis, which may affect mobility and continence. Routine pediatric screening is now standard, but some adults discover spinal anomalies later in life.

Gaps in Care

Many adults face years of delayed care as clinicians fail to link symptoms to their history. While survival has improved, adult services often lag, and patients are discharged from paediatric care without structured follow-up.

Adults need care that addresses long-term physical risks, provides coordinated multidisciplinary support, and acknowledges the emotional and psychological impact of childhood experiences.

Bridging the Gap

Transition must ensure continuity, safety, and dignity.

Structured, patient-centred approaches, including transition clinics, help prevent adults from being "lost" in the system. Multidisciplinary teams provide coordinated, comprehensive care addressing both physical and functional needs.

VACTERL Association

VACTERL Association is a recognised pattern of congenital anomalies involving multiple organ systems.

It is typically diagnosed when at least three of the following component defects are present: vertebral abnormalities, ARMs, cardiac anomalies, TOF with OA, renal abnormalities, and limb differences.

Adults living with VACTERL, like those with other rare conditions, benefit from shared experience, community support, and advocacy.

Survival to Advocacy

Listening to lived experience is essential for safe, effective lifelong care.

The emotional impact of repeated childhood interventions is often underestimated, and patient insights are too frequently dismissed, making self-advocacy vital.

Connecting with others has brought healing, belonging, and empowerment, showing how collective patient voices can foster safer, more compassionate care. These experiences have led me to advocacy, including volunteering with TOFS and Max's Trust, and serving on the ERN eUROGEN European Patient Advisory Group.

Patient voices must shape lifelong care for rare and complex conditions; survival is just the beginning.

TOFS – Live Life Unlimited

tofs lifelong support for those born unable to swallow

Our vision is for anyone born with OA/TOF to be able to live life unlimited. We are working towards a world in which those born with OA/TOF live long and healthy lives, unconstrained by the impact of being born with these conditions.

https://tofs.org.uk

Max's Trust – A Lifeline for the Anorectal Malformation Community

Max's Trust

A safe, welcoming space where people can share experiences, find encouragement and know they are never alone. It's not just about managing a condition it's about reclaiming confidence, dignity, and a sense of belonging.

https://maxtrust.org

ERN eUROGEN – Improving outcomes for those with rare and complex uro-recto-genital conditions

European Reference Network | ERN eUROGEN

Patients are at the heart of ERN eUROGEN. Through our European Patient Advocacy Group (ePAG), people with lived experience and patient organisation representatives work as equal partners alongside clinicians and researchers.

https://eurogen-ern.eu

(Left) A poster demonstrating the challenges of living with rare and complex conditions and the importance of coming together to share experiences. Listening, valuing and gaining support from others walking a similar path.

(Right) VACTERL Visionaries take to the stage to present"Patient Empowerment: a Journey from Isolation to Advocacy."

Useful Links