PIPADOR REVISION

MRCOG Part 1 Revision Notes

Williams Syndrome

CELL BIOLOGY • LECTURE 118

VISUAL EXAMINATION REVISION NOTES

2026 Edition

MRCOG Part 1 Revision Notes

Lecture 118: Williams Syndrome

Cell biology • Visual revision edition — expands Lecture 114

COLOUR CODE USED THROUGHOUT THESE NOTES

ELASTIN / VESSELS	VISUOSPATIAL	FACE / ANXIETY	CALCIUM / CARE	SOCIAL / VERBAL

LEARNING OBJECTIVES — tick off as you go

- ☐ Describe the 7q11.23 deletion and gene–phenotype links.
- ☐ Explain the elastin arteriopathy and anaesthetic risk.
- ☐ List facial, endocrine and other features, including hypercalcaemia.
- ☐ Describe the cognitive and behavioural profile and safeguarding issues.
- ☐ Plan diagnosis, lifelong care and pregnancy in an affected woman.

1 Genetics

IN ONE LINE: *~1.5 Mb deletion at 7q11.23 (NAHR) taking out ELN and ~26 other genes. Elastin loss drives the vessels; LIMK1 and GTF2I families shape the mind.*

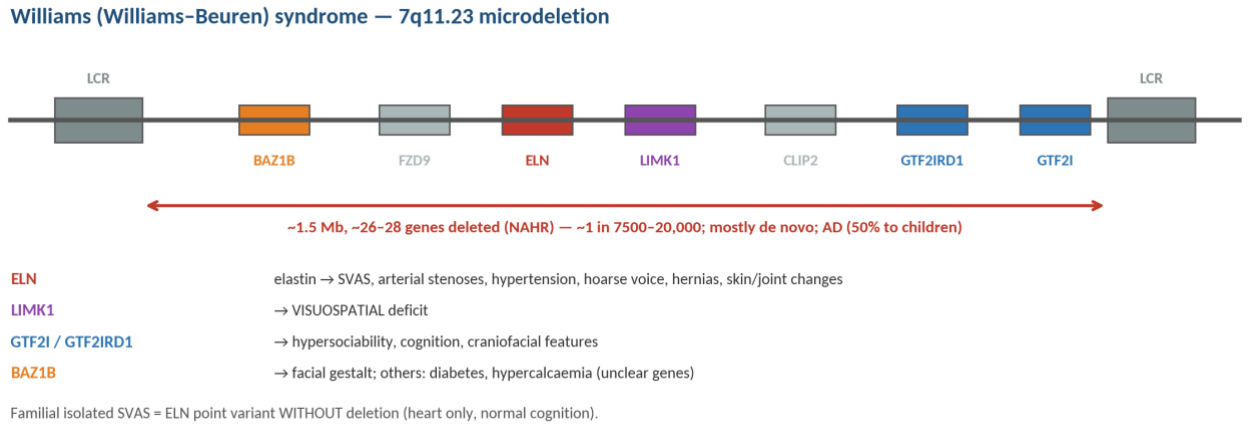


Figure 1. The 7q11.23 region.

2 Heart and Vessels

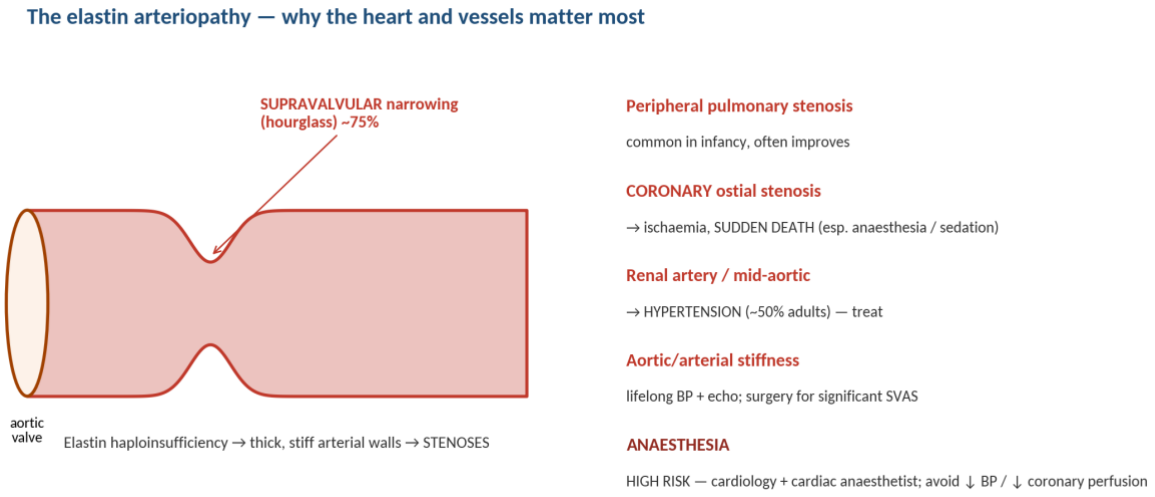


Figure 2. Elastin arteriopathy.

MNEMONIC / MEMORY HOOK

'WILLIAMS = Wide mouth, ID, Lacy iris, Loquacious, Increased Ca, Aortic (supravalvular) stenosis, Music, Sociable'.

3 Features and Profile

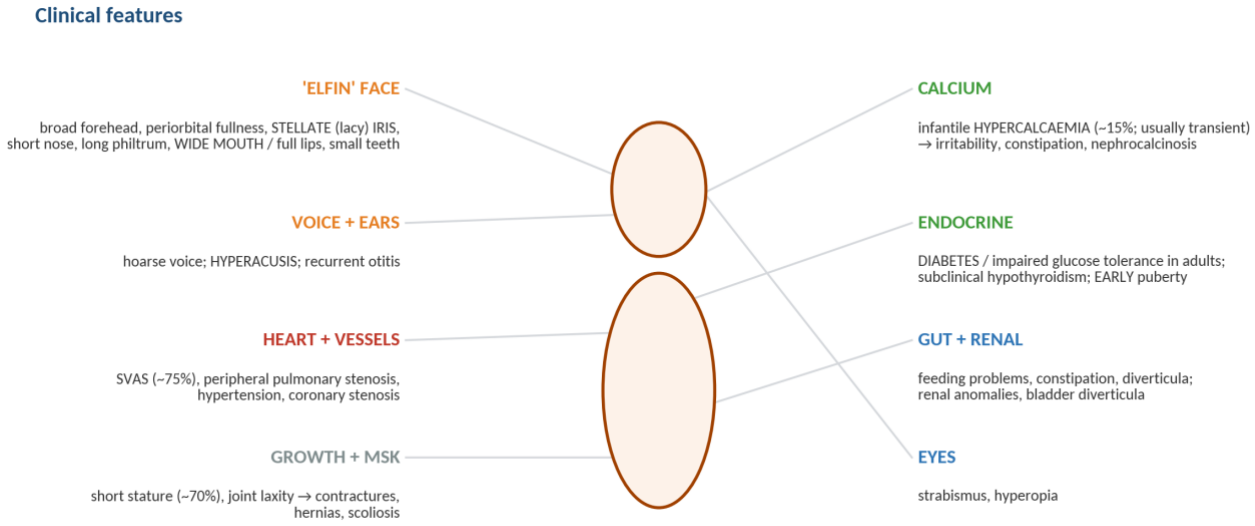


Figure 3. Clinical features.

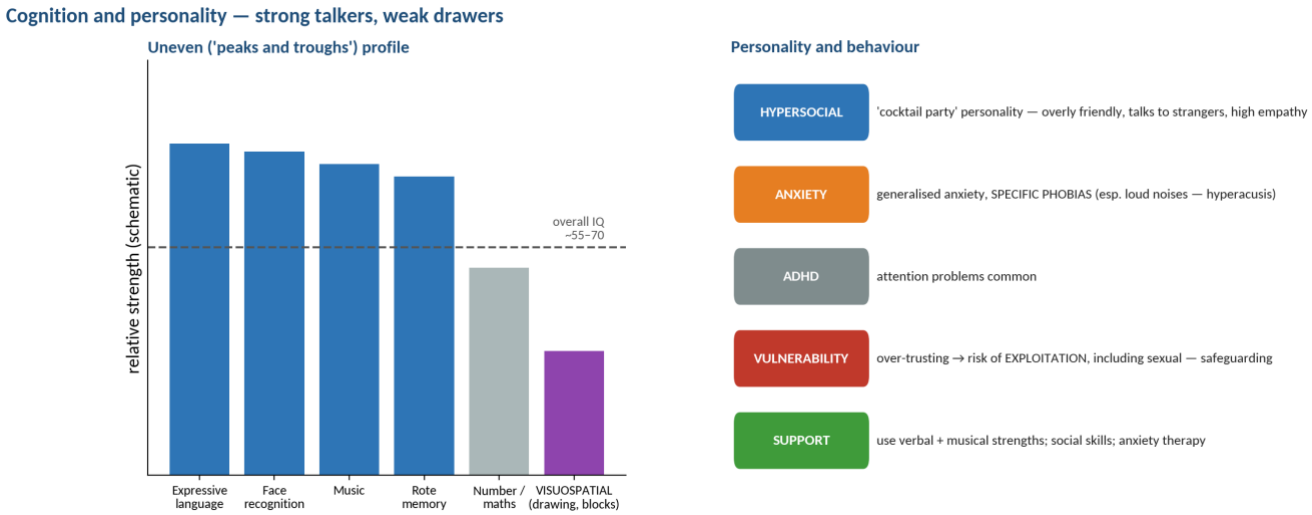


Figure 4. Cognition and personality.

4 Management and Pregnancy

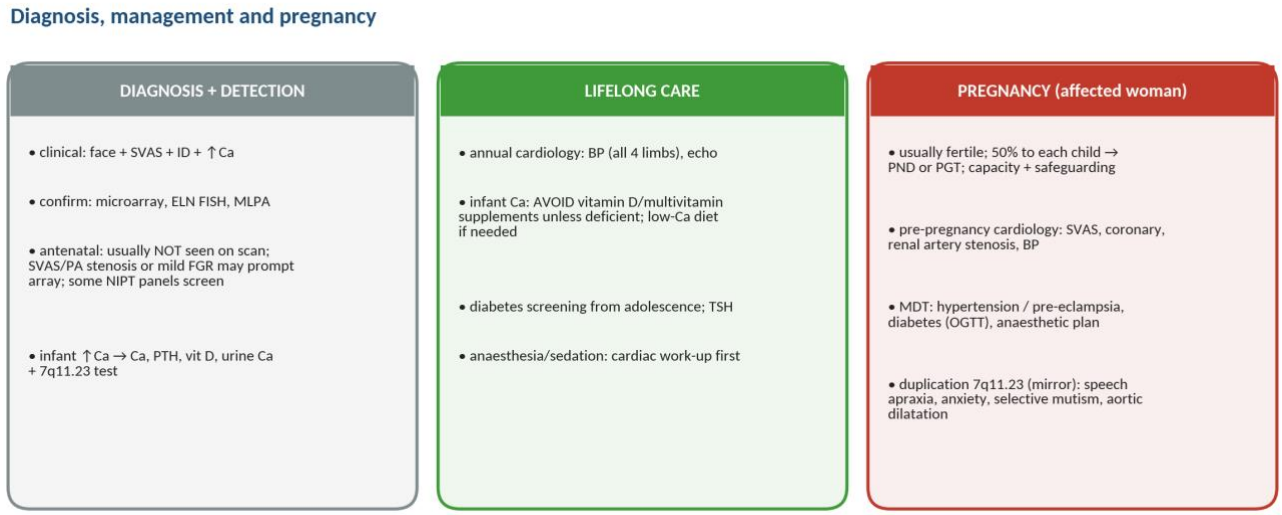


Figure 5. Diagnosis, management, pregnancy.

- **Capacity and consent**: assess decision-making capacity (Mental Capacity Act 2005); support informed choice; be alert to safeguarding concerns given social disinhibition. **Differentials**: familial SVAS (ELN variant), Noonan (pulmonary stenosis), other causes of infantile hypercalcaemia (CYP24A1, excess vitamin D).

> ★ **PEARL**
>
> **Williams = SVAS + HYPERcalcaemia + over-friendly + poor visuospatial skills; 22q11.2 = conotruncal + HYPOcalcaemia.**
>
> **Coronary/arterial stenoses make sedation and anaesthesia dangerous — cardiology review first, including before caesarean or neuraxial block.**

5 MRCOG Exam Pearls — Last-Minute Page

Numbers to know

~1.5 Mb	**~26–28**	**1 in 7500–20,000**	**~75%**
7q11.23 deletion	Genes deleted	Incidence	SVAS

~15%	~70%	55–70	50%
Infant hypercalcaemia	Short stature	Typical IQ range	Risk to children

HIGH-YIELD CHECKLIST

- ☐ 7q11.23 ~1.5 Mb deletion; NAHR; mostly de novo; AD.
- ☐ ELN → SVAS, PPS, coronary/renal stenoses, hypertension.
- ☐ LIMK1 → visuospatial; GTF2I → hypersociability.
- ☐ Elfin face, stellate iris, wide mouth, hoarse voice.
- ☐ Infant hypercalcaemia: avoid vitamin D supplements.
- ☐ Adult diabetes, hypothyroidism, early puberty.
- ☐ Verbal > visuospatial; music; anxiety, phobias, hyperacusis.
- ☐ Exploitation risk — safeguarding, capacity.
- ☐ Anaesthesia high risk; annual cardiology + BP.
- ☐ Pregnancy: cardiology, BP, OGTT, 50%; dup 7q11.23 = mirror.

Self-assessment — cover the right-hand column

Question	Answer
Gene behind the vascular features?	ELN (elastin)
Most characteristic heart lesion?	Supravalvular aortic stenosis
Gene linked to visuospatial deficit?	LIMK1
Calcium problem in infancy?	Hypercalcaemia
Supplement to avoid in infants?	Vitamin D (unless deficient)
Characteristic iris pattern?	Stellate (lacy) iris
Personality type?	Over-friendly 'cocktail party'
Why is anaesthesia risky?	Coronary and arterial stenoses — sudden death
Mirror duplication features?	Speech apraxia, anxiety, selective mutism, aortic dilatation
Risk to children of an affected woman?	50%

6 References

- Kozel BA et al. Williams syndrome. Nat Rev Dis Primers 2021.
- American Academy of Pediatrics. Health care supervision for children with Williams syndrome. Pediatrics 2020.
- Williams Syndrome Foundation (UK) — clinical guidance.