Key Points

- Numerous genetic causes for connective tissue diseases are now known
- Families of disorders can be recognised from the genes involved
- Not every tall person with arachnodactyly has Marfan syndrome
- Regular echocardiography should be undertaken in individuals suspected of having Marfan syndrome
- Lumbar magnetic resonance imaging may assist the diagnosis of Marfan syndrome

in other organ systems (ie skeleton, eye or dura). Magnetic resonance imaging of the lumbar spine looking for dural ectasia can help to establish a diagnosis of Marfan syndrome where definitive evidence from the classical systems is lacking (Fig 1). It is found in 60% of those with classic Marfan syndrome, but may also be present in individuals with less severe phenotypes. The severity of dural ectasia is highly variable, from modest effacement of the epidural fat through scalloping of the posterior border of the lumbar vertebrae to anterior meningocele. Mild variants of dural ectasia should be interpreted with care.

Management. Regular use of beta-blockers in patients with established Marfan syndrome and exhibiting evidence of aortic dilatation retards the progression of aortic distension, thereby delaying the onset of complications such as aortic dissection and aortic reflux. Serious consideration should be given to prophylactic aortic surgery in all individuals in whom the aortic diameter at the sinus of Valsalva reaches 5.5 cm. The use of beta-blockers and the introduction of elective surgery have probably significantly contributed to the increase in life expectancy that has recently been noted[6].

Screening

A variety of mutations in *FBN1* have been described in patients with Marfan syndrome[7]. These range from mutations causing premature stop codons (effectively null alleles) through mutations causing exons to be spliced out of the RNA transcript, to mutations likely to have profound structural effects on profibrillin (eg tyrosine for cysteine mutations ablating intra-chain disulphide bonds necessary for protein folding). Although genetic screening is practicable in many cases, it has not been our experience that it is widely sought by expectant parents.

Unfortunately, the wide range of mutations in the structural components found in these disorders of the mesenchymal tissues does not translate easily to potential cures.

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CME Septicaemia SAQs

Answers to CME SAQs published in *JRCPL* November/December 2000

Q1	Q2	Q3	Q4	Q5	Q11	Q12	Q13	Q14	Q15
a) T	a) F	a) T	a) F	a) F	a) T	a) T	a) T	a) F	a) T
b) T	b) F	b) F	b) F	b) T	b) F	b) F	b) F	b) F	b) F
c) T	c) T	c) T	c) T	c) F	c) F	c) T	c) T	c) T	c) T
d) F	d) T	d) T	d) F	d) F	d) T	d) F	d) F	d) T	d) F
e) T	e) T	e) F	e) F	e) T	e) F	e) F	e) F	e) F	e) F

Q6	Q7	Q8	Q9	Q10	Q16	Q17	Q18	Q19	Q20
a) F	a) T	a) F	a) F	a) F	a) T	a) T	a) T	a) T	a) T
b) T	b) T	b) T	b) T	b) F	b) T	b) T	b) T	b) F	b) T
c) T	c) F	c) T	c) F	c) T	c) T	c) F	c) T	c) F	c) T
d) F	d) F	d) F	d) T	d) T	d) F	d) F	d) T	d) F	d) F
e) F	e) T	e) T	e) T	e) T	e) T	e) T	e) T	e) T	e) F