



CASE REPORT

Familial testicular torsion in three consecutive generations of first-degree relatives

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Abstract We report the first and largest family with testicular torsion in three consecutive generations affecting four first-degree relatives. The incidence of familial testicular torsion is under reported in the literature. We recommend eliciting family history in evaluation of acute scrotum, as a useful adjunct for clinical decision making. In families with a strong predisposition to testicular torsion, management should include family counseling about the significant risk of occurrence of this condition.

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Introduction

Familial occurrence of testicular torsion has been infrequently reported. We report here on the largest number of first-degree relatives affected by testicular torsion in three consecutive generations of one family.

Case report

A 6-month-old male presented to the emergency room with scrotal redness and swelling, initially noticed by the

parents 2 days prior to presentation. He vomited three times and was afebrile.

Physical examination revealed a swollen red tender left testicle and a normal contralateral testicle. Scrotal ultrasound revealed left heterogeneous testicle with peripheral flow only. Patient's medical history was found to be significant for multiple congenital defects, including hypotonia, gastro-esophageal reflux, failure to thrive, growth retardation, cleft palate, mitral valve prolapse, and velocardio-facial syndrome.

Family history, elicited from the father and affected uncle, was found to be significant for mitral valve prolapse and testicular torsion in childhood in the father. Father's brother has hypotonia and history of childhood testicular torsion. Paternal grandfather had testicular torsion in adolescence. Proband's mother and older sister are healthy. Younger brother who was born subsequently underwent elective bilateral orchiopexy at 10 months of age for undescended testes.

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Table 1 Summary of reported cases of familial testicular torsion.

Case #	Index family	Ages of affected index cases	Presentation	Diagnosis	Delay to diagnosis	Treatment	Summary, comments, predisposing factors
1	A [4]	15 years	Severe pain and swelling of left scrotum for 8 days; fever, nausea, vomiting for 1 day; difficulty walking. PE: tender scrotum, sterile urine	Counterclockwise torsion of 720°; unsalvageable testicle	8 days	Left orchiectomy	3 out of 5 male siblings presented with testicular torsion. All six male members of the immediate family (father, 5 brothers) had hypermobility of the testicle on PE. No history of torsion in father/other generations
2		14 years	Pain, swelling of the right scrotum for 7 days; nausea/vomiting for 48 h. PE: 101.2; tender, indurated testicle 3 times the normal size	Clockwise torsion of 360°; absence of gubernaculum, long mesorchium with a high reflection	7 days	Right orchiectomy	
3		21 years	Pain in the right scrotum for 4 days; difficulty walking. PE: tender, indurated right scrotum	Counterclockwise torsion 900°; gubernaculum absent; reperfusion after detorsion	4 days	Right orchiopexy	
4	B [5]	24 h/1st exam	Finding of enlarged scrotum on first exam 24 h after birth. PE: enlarged, indurated, non-tender right testicle; enlargement of right scrotum over 60 h of life	Supravaginal torsion of the right testicle with ischemia	60 h	Orchiopexy of right testicle	Both non-twin brothers were born as full-term infants; no relevant family history; healthy father with normal genitalia; no other siblings; no other cases of torsion in the family
5		2 h after birth	Incidental finding on routine first physical examination at 2 h after birth. PE: swollen scrotum with bluish discoloration; enlarged and indurated right and left testis	Bilateral infarction of the testicular tissue; no twisting of the spermatic cords	2 h after birth	Orchiopexy of both testicles	
6	C [6]	14 years	48-h history of pain and swelling of the left testicle. PE: febrile; red, edematous, tender left scrotum, 3 times the normal size	Gangrenous testicle with 360° torsion	48 h	Orchiectomy of left testicle; right orchiopexy	Homozygous twins with testicular torsion 3 months apart. Both twins were examined when the first twin presented with torsion; the other twin was found to have a normal physical exam; he presented 5 months later with testicular torsion
7		14–15 years	Left testicular pain 5 months after testicular torsion in the homozygous twin brother. PE: tender testicle	Testicular torsion 180° clockwise	Few hours	Bilateral orchiopexy	

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