The pedigree. The father of the patient had been particularly interested in genealogy and assembled the pedigree (Fig. 4). In four of the eight generations preceding the propositus there were seven common ancestors. From the nearest couple, who were great-grandparents of the father and great-great-grandparents of the mother, the parents were related to each other as second cousins, once removed. Proceeding to the more remote generations, the parents were related to each other from two common ancestors in generation III as third cousins, once removed, and from one common ancestor in generation II as fourth cousins, once removed. Finally, from two common ancestors in generation I their relationship was between that of fourth cousins, once removed, and fifth cousins as a result of the several different paths. Altogether, the coefficient of inbreeding, $F$, was 0.0106 for the propositus, which meant that his parents were related as more than second cousins, once removed ($F = 0.0078$) but less than full second cousins ($F = 0.0156$).

The feminine ancestor in generation I was an aunt of the third President of the United States, Thomas Jefferson; she was the sister of his father. We are unaware that this disease has ever been reported among his descendants.

Discussion

Hyalinosis cutis et mucosae is probably determined by a single gene transmitted as autosomal recessive (McKusick, 1971), though formal genetic analysis of a reasonable number of cases has not been published presumably because of the rarity of the disorder. In South Africa, Heyl (1970) has traced the disorder through generations spanning 300 years and found it to be autosomal recessive (Heyl, 1963). As a matter of fact, the disorder has been reported from South Africa more commonly than from anywhere else (Gordon, Gordon, and Botha, 1969). The mode of inheritance has appeared apparent from the lack of direct transmission, the frequency of affected sibs, and the commonness of parental consanguinity. There is as yet no evidence of genetic heterogeneity in spite of the report by Rosenthal and Duke (1967) of direct lineal transmission in two generations. In the family they reported, the parents, one of whom was affected, were related as full second cousins, and the a priori probability of the other being a carrier was $\frac{1}{16}$ or 0.0625. That four of their five progeny were affected was somewhat unexpected, given that among five child families, only 0.015 would be expected to have precisely four affected on the hypothesis of an autosomal recessive gene. These four affected progeny had a total of 10 children, not one of whom was affected, an event expected as infrequently as 0.001 on a dominant gene hypothesis ($\frac{1}{4}$).

The parents of our patient were consanguineous which adds to the evidence for autosomal recessive determination of the disorder. That they were related as somewhat more than second cousins, once removed, but less than full second cousins was largely determined by the two common ancestors from whom they were descended as second cousins, once removed. The occurrence of several more remote common ancestors, albeit interesting in the construction of the pedigree, contributed just over one-fourth to the total coefficient of inbreeding.

The disfigurement and at least the laryngeal disability of our patient have created a social problem for him. This suggests the likelihood of selection against the gene in others who are perhaps similarly affected. Knowledge of the inheritance of the disorder affecting their son played no part in the decision of his parents not to bear other children, because the exact diagnosis was not made until the mother was menopausal and even then its heritability was not recognized.

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Neonatal testicular torsion in two brothers

Summary. Two brothers presenting neonatal testicular torsion are reported. The findings suggest an autosomal or X-linked recessive pattern of inheritance for the anatomical underlying anomaly.
Only one case of familial recurrence of testicular torsion in adults has previously been reported. This is, to our knowledge, the first report of neonatal testicular torsion in two brothers.

Case reports

Case 1 was born on 8 June 1971, after a normal spontaneous delivery. His mother’s full-term pregnancy was uncomplicated. Birth weight was 3500 g. Swelling of the scrotum was found at 24 hours of age when the first physical examination was performed. The right testicle was enlarged, indurated, and not tender to palpation. The spermatic cord was free and normal. At the age of 60 hours enlargement of the right testis was considered to have increased and the spermatic cord had become thicker. Surgery was then indicated. Supravaginal torsion of the right testicle with ischaemia of the testicular tissue was observed. Detorsion and orchidectomy of the right testicle was performed.

Case 2 was born on 25 April 1973 after a normal delivery. The pregnancy was full term and uneventful. Birth weight was 3600 g. Physical examination at 2 hours after birth was negative except for the scrotum which was swollen with a bluish discoloration. Both testicles were enlarged and indurated. The spermatic cords were not engorged. Serosanguineous fluid was obtained by needle aspiration of the left side of the scrotum. Immediate surgery was performed under the presumptive diagnosis of acute testicular torsion. Bilateral infarction of the testicular tissue specially marked in the left testicle was found. Both spermatic cords were not twisted. Orchidectomy of both testicles was performed.

The family. There are no other sibs. Cases 1 and 2 were born at a maternal age of 26 and 28 and paternal age of 24 and 26 years, respectively. The parents are both healthy and not closely related. There is no indication of remote consanguinity by last names or places of birth of the four grandparents. They all are from white, roman catholic, latin European ancestry. The genitalia of the father were examined and found to be normal in all respects. No other cases of testicular torsion were known in this family.

Discussion

The only report of familial recurrence of acute testicular torsion found in the literature (Cunningham, 1960) concerned three brothers aged 14, 15, and 21 years old at the time they underwent torsion of the spermatic cord, apparently of the intravaginal type. Hypermobility of the testes without a history of torsion was diagnosed in the patients’ father as well as in the remaining two brothers of a sibship with five males. No details are given about how testicular mobility was tested, and no ages are stated for the unaffected father and two sibs.

Based on this sole report McKusick (1971) has considered a suggested mode of inheritance for the anatomical anomaly underlying testicular torsion, this being compatible either with the Y-male-limited autosomal dominant or Y-linked types. The family reported here differs from the one cited in several aspects; age of occurrence, anatomical type of torsion, and suggested genealogical pattern.

Acute torsion of the spermatic cord in the neonatal period is rare and it usually affects the suprapenile portion. Spontaneous detorsion of the cord is not unlikely to occur in this suprapenile type where torsion seldom accomplishes a complete revolution. In these cases, only testicular tissue necrosis will be found at surgery (Rhyne, Mantz, and Patton, 1955). This is interpreted to be the situation for case 2. The occurrence of the same type of torsion at the same age group in two sibs is not likely to be just a chance repetition. Under the assumption of a single genetic predisposition for testicular torsion—disregarding anatomical type—the failure to disclose testicular hypermobility in the father of cases 1 and 2 may reflect our inability to elicit a given clinical sign, overdiagnosis in the case of the family previously reported, or the presence of a variable expressivity for this trait. Otherwise, genetic heterogeneity has to be considered, which is likely to be related to the different well known anatomical types of torsion of the spermatic cord and of the testicular embryonic remnants. If this is the case, the family reported here suggests either an autosomal or X-linked recessive mode of inheritance for at least some of the instances of suprapenile torsion of the spermatic cord.

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Received 18 February 1974.
Neonatal testicular torsion in two brothers.

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*J Med Genet* 1975 12: 112-113
doi: 10.1136/jmg.12.1.112

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