Correspondence

Journal of Medical Genetics, 1983, 20, 313-315

Does sacral agenesis predispose to spina bifida?

SIR,

Families are presenting for genetic advice in regard to sacral agenesis (SA). We could find no good published studies which defined the genetics of SA or gave a recurrence risk. The relationship of SA to other forms of neural tube defects (NTD) has not been defined. The availability of intrauterine diagnosis for NTD makes it important to determine the strength of the association.

We ascertained 22 families who had had a child treated for SA at our hospital. Of these children, 15 had neurological dysfunction involving bowel, bladder, or limbs and seven had anorectal anomalies. One child was from a diabetic pregnancy and had multiple malformations. One family had two sibs with SA and associated anterior myelomeningocele, both index cases in our study. There were 14 males and nine females.

Birth order and miscarriage frequency did not differ from expectation. There were 40 sibs and 265 first cousins who were unaffected.

Sacral agenesis is a very heterogeneous entity.¹ A number of published reports are consistent with an autosomal dominant pattern of inheritance² and include families in which both sacral agenesis and spina bifida have occurred in different members. Occasional reports would be consistent with autosomal recessive inheritance.³⁻⁴ Maternal diabetes has been implicated in a number of studies⁵⁻⁸ and paternal diabetes has been noted in two reports.^{1,9} A number of reports indicate the concurrence of SA and NTD in the same patient^{1,4,10-13} (table). In addition, Anderson¹² reported SA in nine of 73 patients with occult spinal dysraphism, and Dubowitz¹³ reported SA in six of 12 patients with a lipoma of the spinal cord.

TABLE Reported association of neural tube defects and sacral agenesis.

	Patients with sacral agenesis	Associated myelomeningocele	Associated sacral lipoma
Durham Smith ⁹	26	13	
Andrish <i>et al</i> ¹⁰	17	3	2
Mariani <i>et al</i> ¹¹	11	2	1
Blumel <i>et al</i> ⁵	50	5	
White and Glauber ¹	22	9	

Although our own series of patients was too small to draw any conclusion about the relationship of SA to other NTD, the reports cited suggest that such a relationship exists. Until the strength of this association is defined, it would seem prudent to offer to families who have had a child with SA intrauterine diagnosis for NTD in a subsequent pregnancy with particular emphasis on high quality ultrasound.

It would require a much larger series of patients to establish an empirical recurrence risk but the occurrence of two affected sibs in this small series suggests a risk of recurrence of the order of 1 in 40. Further studies of SA are needed.

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*Dr Ruth Magnus died before the final preparation of this manuscript.