Acta orthop. scand. *49*, 475–480, 1978

SACROCOCCYGEAL AGENESIS

A Report of Four New Cases

E. DOUNIS

Southmead Hospital, Bristol, England

A total of 188 cases of sacral agenesis reported in the English literature are reviewed, and four new cases added. In 112 cases where the sex was given 60 were males, (53 per cent) and 52 were females (47 per cent). In 163 cases where clinical details were available, 47 children (26 per cent) had unilateral or bilateral dislocation of the hips, 77 (49 per cent) had unilateral or bilateral club-feet and 23 children (17 per cent) had spina bifida deformities. Comments are made on the aetiology, the clinical picture and the management.

Key words: agenesis; agenesia; lumbosacral; sacrococcygeal; sacrum

Accepted 14.xi.77

Sacral agenesis was first described by Hohl in 1852. Further cases have been reported sporadically in the English literature, many describing accompanying musculoskeletal and visceral abnormalities.

The aim of this paper is to add a further four cases and to analyse the incidence of accompanying malformations. Comments are made on the relation of the condition to maternal diabetes, the clinical picture and the aims of management. Table 1 records chronologically the previously reported cases, the sex and the accompanying congenital malformations.

CASE REPORTS

Case 1. A baby girl (T.D.) was born on 7.5.73 to healthy parents, there being no family history of diabetes.

She was noted to have a barrel-shaped chest, atrophic calves and bilateral club-feet (Figure 1). Radiographs (Figure 2) demonstrated absence of the fourth and fifth lumbar vertebrae, sacral agenesis and congenital dislocation of the right hip.

Her club-feet were treated by strapping for 6 weeks and at the age of 4 years they are normal. The congenital dislocation of the right hip was treated initially by an Aberdeen splint, but it did not reduce. Open reduction and de-rotation osteotomy were performed at the age of 18 months followed by a Salter's osteotomy at the age of $3\frac{1}{2}$ years. At the age of 4 years she walks with a slight limp. She is continent of faeces, but still suffers from precipitancy of micturition. She attends a normal school.

Case 2. A baby girl (S.M.) was born on 28.5.73 to healthy parents; the mother had no previous miscarriages. There were no virus infections during the pregnancy and there was no history of diabetes in the family.

She was noted to have bilateral dislocation of the hips and bilateral club-feet with marked atrophy of both calves.

Radiographs (Figure 3) demonstrated sacral agenesis, absent of the fifth lumbar vertebra and spina bifida of the fourth lumbar vertebra. The lumbar spine was interposed between the two iliac bones to replace the absent sacrum. An intravenous pylogram (IVP) examination showed a single pelvic kidney.

Treatment of the club-feet and congenital dis-

Table 1. Cases of sacral agenesis reported, the sex and the accompanying congenital malformations

Year	Author	No. of cases	Sex	C.D.H. bilat. or unilat.	T.E.V. bilat. or unilat.	Spina bifida	Visceral abnormalities
1852	Hohl	1					
1859	Wertheim	1					
1884	Litzman	1	F				
1889	Albert	1					
1910	Mally	1		1	1		
1910	Friedel	1			1		
1910	Fitch	1					
1911	White	1			1	1	
1913	Charlier & Sauti	1					
1913	Rendy	1		1	1		
1921	Elmslie	1	M	1	1		
1923	Bradburn	1					
1924	Stewart	1			1		
1924	Desfosses & Mouchet	1			1		
1924	Foix & Hilleman	1					
1925	Leri & Linossier	1	F		1		
1927	Drehmann	1			1		
1929	Brack	1					
1931	Feller & Srwenbern	1			1	1	
1935	Hamsa	2	M	1	1		
			M	1			
1935	Girard	1	F		1		
1935	Barros Lima	1					
1935	Keinbock & Zimer	1					
1936	Arauzo	1					
1936	Lamoot & Graux	1	F				
1936	Muller	1	M	1	1		
1937	Hilgenreiner	1		1	1		
1938	Pouzet	2	M				
			M	1	1		
1940	Zeligs	2	?				
			F		1		
1941	Feller & Sternberg	1			1		
1945	Berman	1	F				
1947	Litcher	1	F		1	1	
1950	Freeman	1	M				
1951	Del Duca et al.	2	F				
			M	1	1		
1953	Katz	1	M		1		
1953	Soothill	1	M		1		
1957	Williams & Nixon	51		1	5		43
1957	Pirkey & Purcell	2	M		1		
			M		1		
1959	Blumel et al.	50	M 27	18	27	14	8
			F 23				
1961	Dassel	1	F		1		

Table 1—cont.

Year	Author	No. of cases	Sex	C.D.H. bilat. or unilat.	T.E.V. bilat. or unilat.	Spina bifida	Visceral abnormalities
1963	Rüssell & Aitken	5	F 1 M 4				
1967	Frantz & Aitken	3			3		
1969	Banta & Nichols	7	F 4 M 3	4	4	1	
1971	Rosenfelder	24	F 12 M 12	8	12	6	
1976	Abraham	1	M	1	1		
1977	Dounis	4	F 3 M 1	3	4	2	2

location of the hips was abandoned after 2 months because of severe mental retardation. The patient remains in a hospital for mentally subnormal children.

Case 3. A boy (A.E.) was born on 14.4.68 to healthy parents. The mother did not suffer from any virus infections during the pregnancy and there was no history of diabetes in the family. The child was born with severe kyphoscoliosis, bilateral club-feet and flail legs. Radiographs demonstrated sacral agenesis, multiple hemivertebrae and subluxation at the left hip. The exact level of the agenesis was impossible to determine because of the kyphoscoliosis and pelvic tilt. An IVP demonstrated fused kidneys.

All attempts at corrective treatment were abandoned after a short time because of the flail legs. Although incontinent of urine and faeces, he attends normal school and manages using a wheelchair and orthotic devices.

Case 4. A baby girl (J.M.) was born on 3.8.71 to healthy parents with no family history of diabetes. She attended an orthopaedic clinic at the age of 5 years, having suffered progressive cavovarus deformity of both feet over a period of 2 years. She was noted to have atrophy of both calves and radiographs demonstrated sacral agenesis below the level of the second sacral vertebra (Figure 4). At the present time she is undergoing surgery for correction of foot deformities.

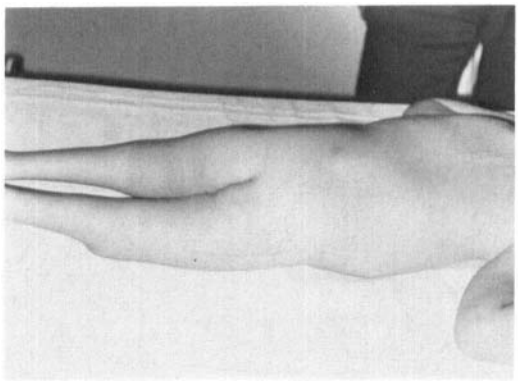

Figure 1. Barrel shape chest and atrophic calves (Case 1).

DISCUSSION

There have been a number of theories concerning the aetiology of malformations and absence of the caudal part of the spine. A family history has been reported in only one case, a father and son (Pouzet 1938). Maternal diabetes may also be a significant aetiological factor. In the 192 cases described in the literature, 16 diabetic mothers have been reported (8.33 per cent), and this incidence may be even higher as many did not record the health of the parents.

It has further been suggested that insulin injections may have a teratogenic effect on the embryo. This view has been supported by

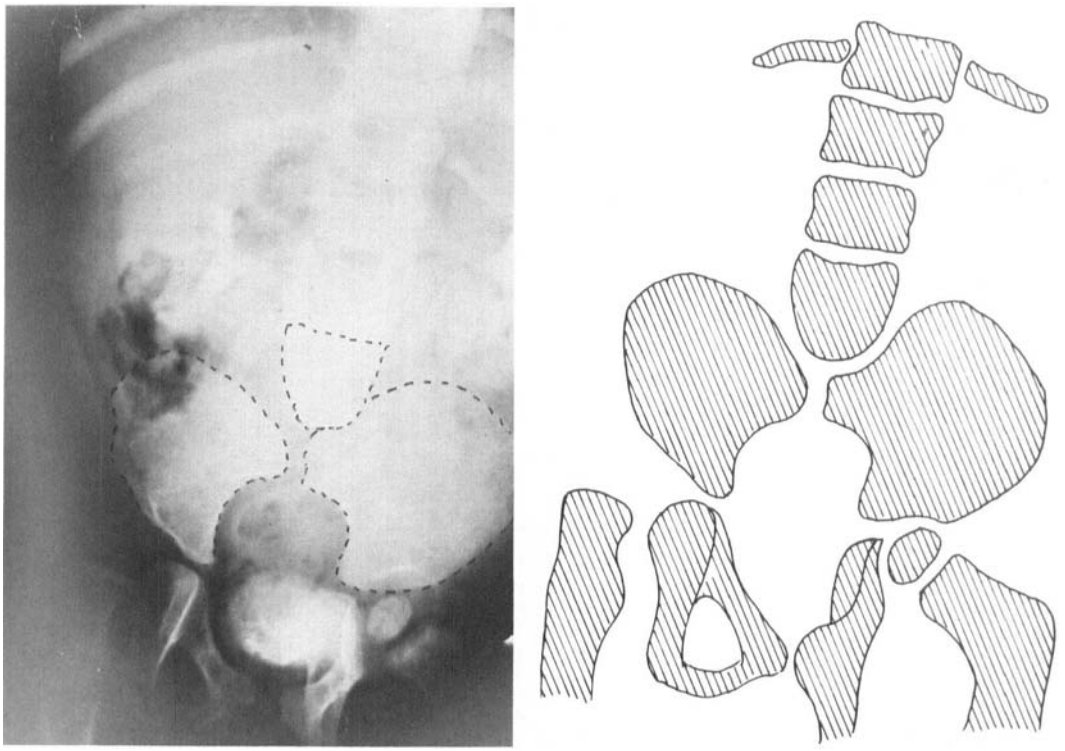

Figure 2. Absence of the lower two lumbar vertebrae and sacrum with apposition of the iliac bones (Case 1).

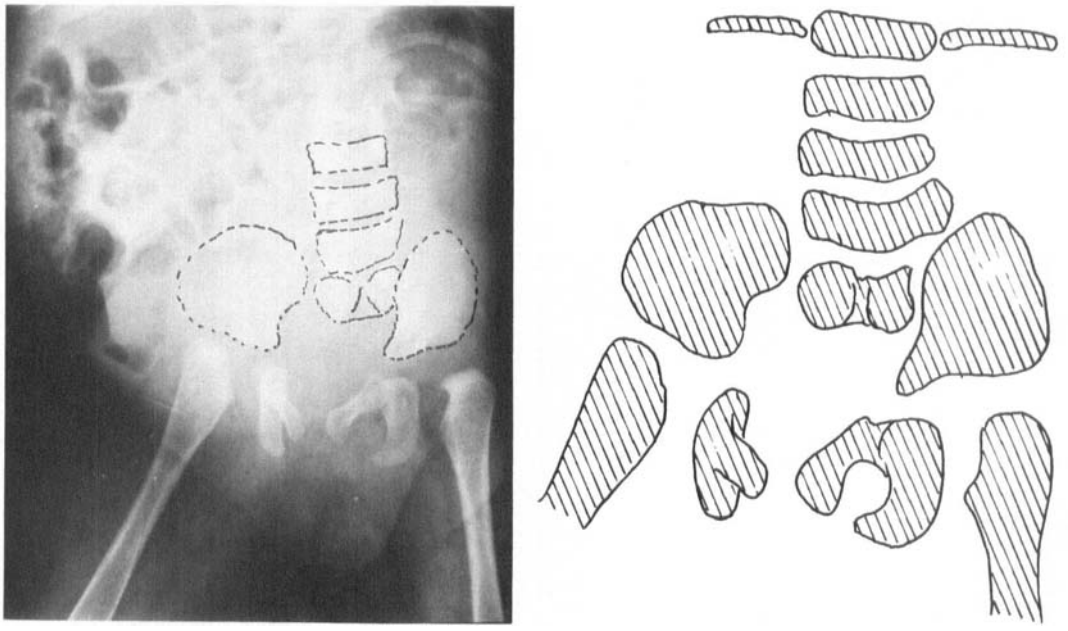

Figure 3. Spina bifida of the fourth lumbar vertebra and absence of the sacrum and fifth lumbar vertebra. The lumbar spine is interposed between the iliac bones (Case 2).

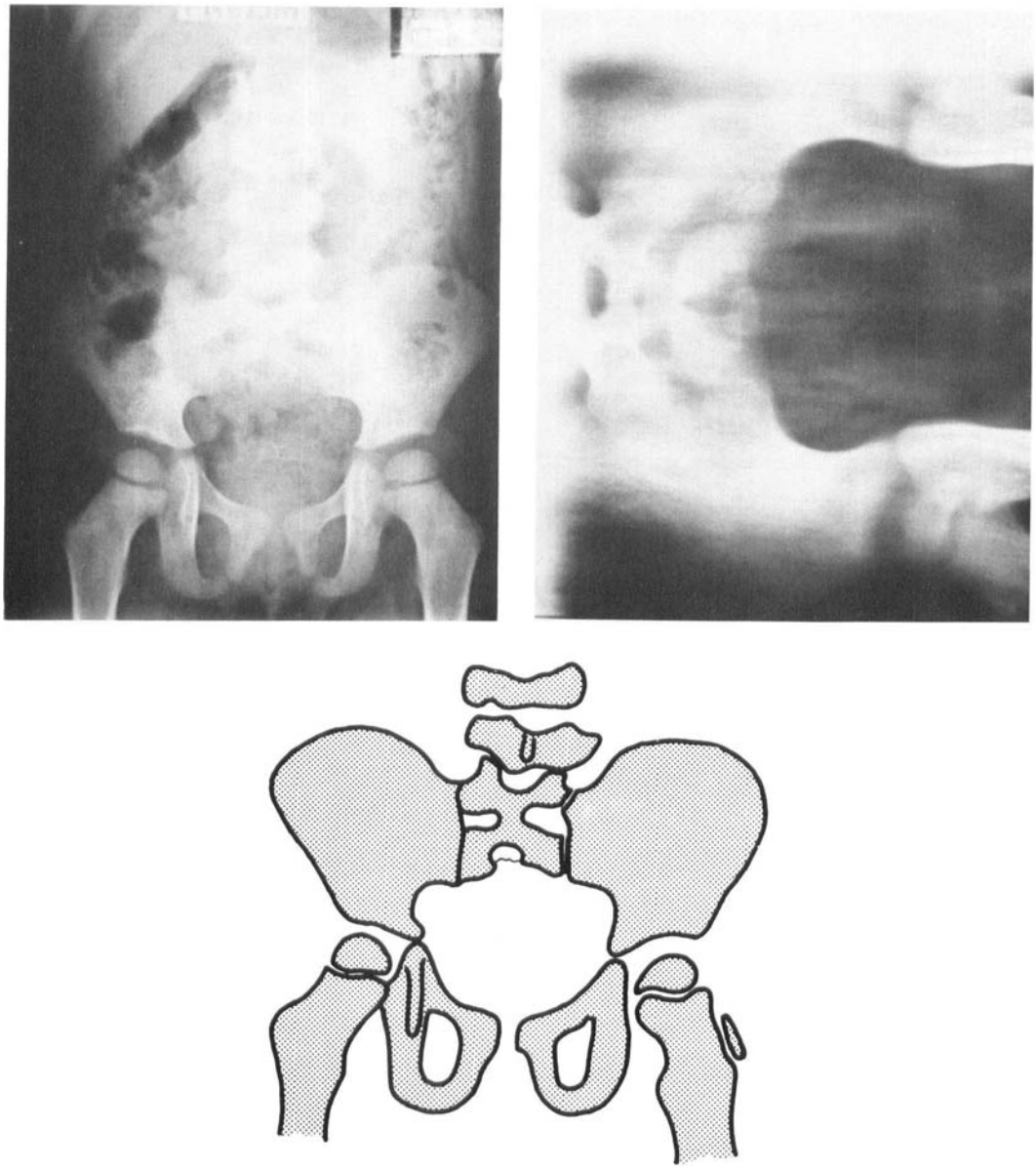

Figure 4. X-ray and tomography show partial absence of the sacrum (Case 4).

Duraiswami (1950) who has experimentally produced caudal depression in chickens by injecting insulin into the eggs. He excluded trauma as a cause, since inactivate insulin or normal saline injections produced no deformity.

The typical clinical picture is of a child with a barrel-shaped chest contrasting with narrow and flattened buttocks and shortened intergluteal cleft. The lumbar lordosis is normally absent, although there may be prominence of the lower lumbar spine (Figure 1). The legs are cone-shaped due to atrophic or absent calf muscles. The pelvic ring is completed with either direct apposition of the iliac bones or with interposition of the lum-

bar spine replacing the absent sacrum (Figures 2, 3). Scoliosis occasionally occurs and is, in some cases, paralytic or due to hemivertebrae. Visceral abnormalities such as anal atrasia, fused kidneys and congenital heart malformations are not uncommon.

The management of these cases is symptomatic for each system, and the treatment should start as soon as possible.

ACKNOWLEDGEMENTS

I wish to thank Mr H. E. D. Griffiths for allowing me to report three of his cases and for reading the manuscript, and Mr. P. J. Witherow for allowing me to report one of his cases. I am also grateful to Mr. W. G. J. Hampson for his encouragement in the writing of this paper and for reading and correcting it.

REFERENCES

Abraham, E. (1976) Lumbosacral coccygeal agenesis. *J. Bone Jt Surg.* **58**-A, 1169–1170.

Addison, J. (1955) Sacro-coccygeal agenesis. *Proc. roy. Soc. Med.* **48,** 163–164.

Blumel, J., Burke Evans, E. & Eggers, G. W. N. (1959) Partial and complete agenesis or malformation of the sacrum and associated anomalies. *J. Bone Jt Surg.* **41**-A, 497–517.

Danforth, C. H. (1932–33) Artificial and hereditary suppression of sacral vertebrae in the fowl. *Proc. Soc. exp. Biol. Med.* **30,** 143–145.

Dassel, P. M. (1961) Agenesis of the sacrum and coccyx. *Amer. J. Roentgenol.* **85,** 697–700.

Del Duca, V., Vernon Davis, E. & Barroway, J. N. (1951) Congenital absence of the sacrum and coccyx. *J. Bone Jt Surg.* **33**-A, 248–253.

Duraiswami, P. K. (1950) Insulin induced skeletal abnormalities in chickens. *Brit. med. J.* **2,** 384–390.

Elmslie, R. C. (1921) Case of congenital absence of sacrum with associated dislocation of one hip and talipes calcaneo-valgus. *Proc. roy. Soc. Med.* **15,** 8.

Frantz, C. H. & Aitken, G. T. (1967) Complete absence of the lumbar spine and sacrum. *J. Bone Jt Surg.* **49**-A, 1531–1540.

Freeman, B. (1950) Congenital absence of the sacrum and coccyx. *Brit. J. Surg.* **37,** 299–303.

Girard, P. M. (1935) Congenital absence of sacrum. *J. Bone Jt Surg.* **17,** 1062–1064.

Hamsa, N. R. (1935) Congenital absence of sacrum. *Arch. Surg.* **30,** 647–666.

Hohl, A. F. (1852) *Zur Pathologie des Beekens. I. Das schrag-ovale Beeken.* p. 61. Wilhelm Engelmann, Leipzig.

Katz, J. F. (1953) Congenital absence of the sacrum and coccyx. *J. Bone Jt Surg.* **35**-A, 398–402.

Pirkey, E. L. & Purcell, J. H. (1957) Agenesis of lumbosacral vertebrae. *Radiology* **69,** 726–729.

Pouzet, F. (1938) Les anomalies de development due sacrum. *Lyon chir.* **35,** 371–373.

Rosenfelder, R. (1971) Vertebral agenesis. *Phys. Ther.* **48,** 203–217.

Rüssell, W. E. & Aitken, G. T. (1963) Congenital absence of the sacrum and lumbar vertebrae with prosthetic management. *J. Bone Jt Surg.* **45**-A, 501–507.

Soothill, J. F. (1953–54) Congenital absence of sacrum and coccyx. *Guy's Hosp. Rep.* **102–103,** 87–91.

Williams, D. J. & Nixon, H. H. (1957) Agenesis of the sacrum. *Surg. Gynec. Obstet.* **105,** 84–88.

Correspondence to: Dr. E. Dounis, Filis 178, Athens, Greece