

Seckel Syndrome

Sir,

Although the literature on the 'Seckel syndrome' is very sparse, I was not surprised to see that Sauk *et al* (1973) missed my report (Szalay, 1964) of a patient with the Seckel syndrome. In 1963 and 1964 (Szalay, 1963; 1964), I used the feature of craniofacial disproportion to separate the Seckel and Russell dwarfs from other forms of intrauterine growth retardation (IUGR); I still feel (Szalay, 1972a and b; 1973) that this feature is a useful clinical point in differentiating these dwarfs (Table).

My report (Szalay, 1964) was the second to reveal normal chromosome studies in the Seckel dwarf. However, cytogenetic studies (especially the new banding techniques) are essential in view of the increasing numbers of chromosomal anomalies being found (Table). More recently, acute myeloid leukemia has been reported in a patient with IUGR, microcephalic dwarfism, and a ring 1 chromosome (Bobrow *et al*, 1973).

I last saw my patient in 1963. A recent inquiry revealed that she was employed in a menial position in a protected environment for some time and has remained mentally retarded, microcephalic, and dwarfed. At the present time, she cannot be located but follow-up

attempts are continuing (personal communication, Professor H. Medovy, Winnipeg, Canada).

Yours, etc,

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REFERENCES

- Bobrow, M., Emerson, P. M., Spriggs, A. I., and Ellis, H. L. (1973). Ring-1 chromosome, microcephalic dwarfism, and acute myeloid leukemia. *American Journal of Diseases of Children*, **126**, 257–260.
- Sauk, J. J., Litt, R., Espiritu, C. E., and Delaney, J. R. (1973). Familial bird-headed dwarfism (Seckel's syndrome). *Journal of Medical Genetics*, **10**, 196–198.
- Szalay, G. C. (1963). Pseudohydrocephalus in dwarfs: the Russell dwarf. *Journal of Pediatrics*, **63**, 622–633.
- Szalay, G. C. (1964). Intrauterine growth retardation versus Silver's syndrome. *Journal of Pediatrics*, **64**, 234–240.
- Szalay, G. C. (1972a). Russell dwarf versus Silver syndrome. (Letter.) *Journal of Pediatrics*, **80**, 1066–1068.
- Szalay, G. C. (1972b). Facial profile of the Russell dwarf. (Letter.) *Journal of Pediatrics*, **81**, 1035.
- Szalay, G. C. (1973). Definition of the Russell-Silver syndrome. (Letter.) *Pediatrics*, **52**, 309–310.

TABLE*

Form of intrauterine dwarfism	Craniofacial disproportion	Reported chromosomal anomalies†	Clinical features
Seckel	Yes	Ring 1 chromosome	IUGR; microcephaly
Silver	No	Diploid-triploid mosaicism	IUGR; congenital asymmetry
Russell	Yes	47,XY,+18/46,XY mosaic 45,X/46,XY mosaic	IUGR; pseudohydrocephalus

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† See references in Szalay (1973).

Cellular Metachromasia with Toluidine Blue O in Cultured White Cells of Cystic Fibrosis Heterozygotes

Sir,

Studies on cultured leucocytes of cystic fibrosis (CF) patients and heterozygotes (Danes and Bearn, 1969; Danes *et al*, 1969) have confirmed the occurrence in

these cells of Toluidine Blue O metachromasia similar to that previously observed in cultured fibroblasts (Danes and Bearn, 1968). In some CF patients, however, the cultured white cells remained ameta-chromatic and a similar absence of metachromasia was noted in leucocytes from the parents of these individuals. Because of our interest in CF heterozygote detection we examined white cell cultures from 45 parents of CF patients (ie,