Summary

A 22-month-old girl with cystinotic rickets was given 1 μ g 1,25-dihydroxycholecalciferol (1,25-DHCC) daily in addition to standard treatment. Her rickets healed and linear growth rate appeared to increase. It is suggested that the effect of 1,25-DHCC and its metabolically active analogues on cystinotic rickets should be further studied.

We are grateful to Dr. R. H. Wilkinson for biochemical help, to Mr. A. J. Bron for the slit-lamp examinations, and to Dr. J. G. G. Ledingham for advice, and also to Leo Laboratories and Roche Products Ltd. for supplies of vitamin D metabolites.

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Short stature with pigmentation

The course of 4 children who grew slowly has been followed for from 2½ to 23 years; they were all below

average in intelligence but not severely subnormal; the skin showed excessive brown pigmentation which was not racial—2 were of southern English descent and 2 Jewish. Pigmentation increased on exposure to light. One was an only child, each of the others had one healthy sib of normal size and colouring; the parents were healthy and not consanguineous. In each case pregnancy, birth, and neonatal state were normal; the babies were not small at birth, their weight being in the range 2950-3450 g. Other features are indicated in the Table.

Table Features of 4 patients with short stature and pigmentation

	Cases			
	1	2	3	4
Sex	F	F	F	M
Adult height (cm)	143	*	123	145
Relatively short limbs	+	?	+	+
Small hands and feet	+	?	+	+
Reduced bone age	+	0	0	+
Sparse scalp hair	+	0	+	+
High forehead	+	+	+	+
Long cranium	±	?	+	+
Small low ears	+	+	+	+
Epicanthus and antimongoloid slant	+	+	+	+
Refractive error	+		+	
Weak lateral rectus	+			+
Systolic murmur	+		+	
Mental dullness	ESN	+	+	+
School	ESN	Normal	Normal	ESN
Menarche (years)	22	?	16	—
Cerebral atrophy on AEG	+	†	+	?
ACTH level normal in blood		+	+	+
MSH		+		+
Growth hormone level after stimulation			+	
Chromosome analysis normal			+	

*Under 3rd centile at the age of 8 years.

†AEG not done but x-rays showed a tubular bony spur arising from the anterior wall of the pituitary fossa.

ESN=school for educationally subnormal; ACTH=adrenocorticotrophic hormone; MSH=melanocyte-stimulating hormone; AEG=lumbar pneumoencephalogram.

In 3 cases vomiting was severe in the early months and also in the fourth case at 3 years; fever was associated with vomiting in 3. Tonsillitis occurred often in 3, leading to tonsillectomy. In each head size was increased or was concordant with age rather than bodily size at some stage. Dentition was normal.

Available information suggests that when grown up these patients are rather hypomelic little people with brownish skins, cheerful, with useful social accomplishments and somewhat low intelligence. As an example, at the age of 26 in Case 1 the weight was of an average 16-year-old, the height of a 10-year-old, with chest circumference suitable for 12 years, and span and upper:lower segment ratio for 6 years.

a

b

c

d

Fig. Faces of patients, showing epicanthus, antimongoloid slant of orbital fissures, with prominent eyes and pronounced nasolabial folds. (a) Case 1, female, at 4 years; (b) Case 2, female, at 5 years; (c) Case 3, female, at 15 years; (d) Case 4, male, at 3 years.

Breasts were flat but nipples were of adult appearance. She had neither pubic nor axillary hair, both of which were present in Case 2 at 23 years. There was some resemblance in facial appearance between the patients.

No explanation of the pathology of this condition is available, but it is notable that in the 2 patients in whom pneumoencephalography was done the brain appeared partially atrophic, while in the third there was a bony abnormality in the pituitary fossa. Lateral rectus muscle weakness in 2 suggests implication of the 6th cranial nerve.

I have found no published record of similar cases.

Summary

Four hypomelic children of abnormally short stature had slight intellectual defect, melanotic skin, and some facial features in common. 3 were followed to the age of 23–26 years, and they remained small and pigmented.

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The following articles will appear in future issues of this journal:

Long-term control of central nervous system leukaemia. *Moir A. Gribbin, R. M. Hardisty, and Judith M. Chessells.*

Yeast opsonization defect and immunoglobulin deficiency in severe infantile dermatitis (Leiner's disease). *D. I. K. Evans, A. Holzel, and H. MacFarlane.*

Early neonatal bacteraemia: comparison of group B streptococcal, other Gram-positive and Gram-negative infections. *Heather Jeffery, Ruth Mitchison, J. S. Wigglesworth, and Pamela A. Davies.*

Increasing breast feeding in a community. *K. S. Sloper, E. Elsdon, and J. D. Baum.*

Giant cell arteritis with gangrene in a child. *G. McEnery.*

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