

Table 2 Number of pancreatin capsules* taken and fat malabsorption (%) while taking usual and modified pancreatin doses

Case No	Usual pancreatin dose		Modified pancreatin dose	
	No of capsules	% Malabsorption†	No of capsules	% Malabsorption†
1	39	5	18	6
2	37	16	18	18
3	51	8	20	39
4	45	22	27	43
5	50	7	28	5
6	44	7	19	10
7	46	5	19	6

*Capsule numbers refer to an average daily intake.

†% Malabsorption refers to the percentage of orally ingested fat malabsorbed, as determined by a three day fat balance study.

reductions in their enzyme intake without appreciable alteration in their absorption. Both these patients achieved normal fat absorption with the reduced enzyme dose.

Discussion

This study has shown that some patients with cystic fibrosis take inappropriately high doses of pancreatic enzymes and that reduction of this high input may be achieved without deterioration in fat absorption. However, two of the patients did have an increase in fat malabsorption when their enzyme dose was decreased. This argues against a policy of setting an arbitrary limit to the enzyme dose, as is frequently suggested at cystic fibrosis clinical meetings. All patients in this study had reached their current dose of enzymes in response to symptoms perceived as due to residual malabsorption. This study highlights the risks of assuming that abdominal symptoms in patients with cystic fibrosis are invariably due to pancreatic enzyme insufficiency and therefore will respond to an increase in enzyme treatment; five of the seven subjects had normal absorption despite persisting symptoms.

The major implication of the results of the present study is that objective assessment of nutrient absorption should be routine in patients with cystic fibrosis, particularly those with abdominal symptoms. The best of the

currently available tests is the three day faecal fat balance. This procedure is time consuming, prone to large errors if not performed correctly, and rarely welcomed by patients. The results of the present study, however, illustrate the importance of assessing changes in enzyme treatment by direct measurements of nutrient absorption.

In conclusion the present study has shown that some children with cystic fibrosis may be taking inappropriately high doses of pancreatic enzymes because of abdominal symptoms not directly related to exocrine pancreatic insufficiency. It is important to determine whether symptoms are due to fat malabsorption and that increases in enzyme replacement result in improved absorption. Recognition of other potential causes of abdominal symptoms in cystic fibrosis should lead to a more logical approach to the patient with residual symptoms on standard enzyme treatment.

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Dexamethasone treatment for congenital adrenal hyperplasia

M C Young, I A Hughes

Abstract

Ten patients with congenital adrenal hyperplasia (three males, seven females; aged 12-29 years) had their usual glucocorticoid treatment changed to dexamethasone in three crossover dosage regimens. A starting dose of 5 µg/kg/day is suggested but as no one dose regimen resulted in adequate control the timing of the dose must be decided for each patient.

The optimum glucocorticoid preparation, total daily dose, and dose schedule for the treatment of congenital adrenal hyperplasia remain

controversial.¹ Hydrocortisone is probably the preparation of choice in infancy and childhood, but in adolescents near completion of growth and adults dexamethasone may be a convenient alternative.¹⁻³ The potency of dexamethasone in relation to both pituitary-adrenal suppression and side effects is much greater.

Patients

Ten patients (three males and seven females) aged 12-29 years with 21-hydroxylase deficiency (classic salt losing type) congenital adrenal hyperplasia took part in the study.

Department of
Child Health,
University of Wales
College of Medicine,
Cardiff
M C Young
I A Hughes

Correspondence to:
Professor IA Hughes,
University Department
of Paediatrics,
Addenbrooke's Hospital,
Level E8, Hills Road,
Cambridge CB2 2QQ.

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Sialic acid storage disease

P D Cameron, V Dubowitz, G T N Besley, A H Fensom

Abstract

A baby girl with coarse facial features, hepatosplenomegaly, and developmental delay had raised free sialic acid concentrations in her urine and cultured fibroblasts. She died aged 13 months. Sialic acid is an important constituent of many glycoproteins and glycolipids; impaired release from the lysosome may be the underlying biochemical defect.

Disorders of sialic acid metabolism fall into four categories. Raised concentrations of bound sialic acid are found in conditions associated with neuraminidase deficiency.¹ No enzyme deficiency has been shown in three other categories where raised concentrations of free sialic acid are found: Salla disease, sialuria, and severe infantile sialic acid storage disease. Salla disease, named after an area in northern Finland, comprises progressive psychomotor retardation with lysosomal storage in patients reaching adult life.² Two cases of sialuria have been described where only sialic acid in the urine was raised. These patients had coarse facies, hepatosplenomegaly, a mild clinical course, and no lysosomal storage.³ This case report concerns the fourth category: severe infantile sialic acid storage disease.

Case report

The baby girl was the second child of healthy unrelated white parents; her brother, aged 2 years, was normal. Pregnancy was complicated by premature labour at 32 weeks' gestation; breech presentation necessitated a caesarean section. Birth weight and head circumference were both below the third centile. Resuscitation with intubation was required for five minutes. Initial problems included transient tachypnoea and episodes of cardiac failure with dusky spells and hepatomegaly. Chest radiographs showed cardiomegaly and pulmonary plethora. Transfusion was required for anaemia.

Dysmorphic features comprised hypertelorism, prominent epicanthic folds, strikingly fluffy eyebrows, a long philtrum, and a high arched palate (figure). Facial features were generally coarse. She had short stubby fingers and a square set thumb. Her skin was pale with wispy orange hair.

Diuretics and fluid restriction controlled her



Dysmorphic features showing coarse facies, wispy hair and eyebrows, and telangiectasias.

cardiac failure and she was discharged home at 8 weeks. She was readmitted at 11, 13, and 16 weeks with chest infections and cardiac failure. Hepatosplenomegaly and telangiectasias had become prominent. She developed a need for supplemental oxygen, and weight gain remained poor. By 5 months she had rarely smiled, she remained hypotonic, visual following was poor, and auditory response was absent.

Laboratory investigations at 5 months included measurement of blood concentrations of urea, electrolytes, and haemoglobin, liver function, blood and urine amino acids, urine mucopolysaccharides, and lactate. Chromosome analysis, iron studies, thyroid function tests, and lysosomal enzyme activities were also performed. All studies gave normal results. Skeletal survey, cranial ultrasound, and nitrogen washout tests were also normal. Chest radiography showed fine hazy shadowing in the lung fields and cardiomegaly. Ophthalmic examination showed an intermittent divergent strabismus.

Hammersmith Hospital,
London

P D Cameron
V Dubowitz

Royal Hospital for
Sick Children,
Edinburgh
G T N Besley

Regional Genetic
Centre, Guy's Hospital,
London
A H Fensom

Correspondence to:
Dr P D Cameron,
Department of Paediatrics,
University Hospital of Wales,
Heath Park,
Cardiff CF4 4XW.

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M C Young and I A Hughes

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