Anthrop. Közl. *26;* 57—58. 1982.

SINGLE PRIMARY TUBULOPATHIES CAUSING GROWTH IMPAIRMENT

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Abstract: About 25 primary tubulopathies of the kidney are known in man. At least ten of them may cause disturbed growth.

In renal vastage of water, sodium, potassium, calcium, magnesium or inorganic phosphate the mechanism of growth failure is quite obvious, repletion therapy is an important factor in treatment.

Deficient excretion of hydrogen ion invariably leads to stunted growth by secondary mechanism.

Some tubulopathies (dibasic aminoaciduria, Hartnup disease) may cause growth retardation by accumulation of toxic compounds or deficiency of essential substances.

Congenitally disturbed endocrine function of the renal tubule (1-hydroxylation of 25-hydroxycholecalciferol) has also been described.

Primary tubulopathies often manifest themselves by growth failure preventable by early diagnosis or screening.

Key words: tubulopathies, growth impairment.

The machinery of the renal tubule fulfills a large variety of excretory and reabsorptive tasks. Organic compounds like glucose, aminoacids, uric acid are nearly completely reabsorbed in the proximal segment of the tubule while inorganic compounds (sodium, potassium, chloride, bicarbonate, hydrogen, other ions and water) are regulatively reabsorbed, rejected or excreted all along the renal tubule. All these processes need intricate collaboration of transport systems and enzymes. Inherited defects of single transport functions may be innocuous, knowledge of them is still important since they have to be distinguished from severe disorders, e.g. renal glucosuria from diabetes mellitus. Most primary tubulopathies follow a clear-cut mode of inheritance; some tubular defects are shared by the intestinal mucosa. As expected, increasing insight into the subtilities of the underlying biochemical defect has helped to subdivide these disorders into genetically distinct varieties.

Most renal tubulopathies causing symptoms at all interfere to some degree with normal growth during childhood. As a rule, renal vastage or pathological retention of inorganic compounds invariably leads to stunted growth as can be seen in the Table 1 while loss of organic compounds like glucose or dibasic aminoacids is usually compatible with normal growth or lead only quite indirectly to growth impairment.

One of the best indicators of effectiveness of treatment is restitution of normal growth or achievement of catch-up growth. For example, in renal tubular acidosis alkali treatment properly adjusted to the patient's needs

Table 1

Primary disorders of single tubular functions

Name of disorder	Compound(s) involved	Stunted growth	Treatment
1. Bartter syndrome	K^+	+++	++
2. Disabled K^+-excretion	K^+	++	+
3. Idiopathic hypercalciuria	Ca^{++}	++	+
4. Idiopathic hypermagnesiuria	Mg^{++}	++	?
5. Familial hypophosphatoemic rickets	PO_4	++	+
6. Renal tubular acidosis			
Type I(distal)	H^+	++	++
Type II(proximal)	HCO_2^-	++	++
Dista RTA+VIII. nerve	H^+	++	?
7. Renal diabetes insipidus	H_2O	++	+
8. Familial azotaemia	urea	— not necessary	
9. Hypouricaemia	uric acid	— (?) not necessary	
10. Renal glucosuria, Types A, B	glucose	— not necessary	
11. Hyperglycinuria	GLY	(+) ?	
12. Iminoglycinuria, Types I—IV	GLY, PRO, HYPRO	— not necessary	
13. Cystinuria, Types I—III	CYS, LYS, ARG, ORN	—	++
14. Hypercystinuria	CYS	—	++
15. Dibasic hyperaminoaciduria			
Type I, protein intolerance	LYS, ARG, ORN	++	+
Type II	LYS, ARG, ORN	(?)	?
16. Hyperlysinuria	LYS	(+)	?
17. Hartnup disease, Types I, II	neutral aminoacids	(+)	++

results not only in correction of acidosis and concomitant hypercalciuria but also induces acceleration in delayed growth.

Severely impaired growth may be caused by a very long list of disorders, many of which are quite obvious, only a small proportion of children with growth problems have primary tubulopathy; still, it is imperative to look after this group of disorders in every case of stunted growth since early introduction of treatment may lead to complete correction of symptoms.

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