

SWEAT ELECTROLYTES AS A CLINICAL AND GENETIC TEST IN MUCOVISCIDOSIS

Study of Variability and Genetic Conditioning

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SUMMARY

Aiming at a better clinical and genetic utilization of the sweat test in mucoviscidosis, the authors have carried out an evaluation of the variability and the genetic conditioning of the level of Na⁺ and Cl⁻ electrolytes in human sweat.

The modifications of the human sweat levels of Na⁺ and Cl⁻ electrolytes play a relevant role in the diagnosis of malabsorption syndromes, such as mucoviscidosis. Moreover, similar modifications have been shown by clinically healthy relatives of patients of mucoviscidosis.

On the other hand, values have been shown to vary, according to sex, age, and season of the year (Lobeck and Huebner 1962).

It has therefore been considered worthwhile (1) to standardize the method, and (2) to find out the extent of the hereditary conditioning of Na⁺ and Cl⁻ sweat levels. The HTLI Cystic Fibrosis Analyzer technique has been followed.

STANDARDIZATION OF THE METHOD

(1) *Variability of the method*: the measure of the same sample, performed 10 times by the same operator, has yielded an average value of 47.0 ± 1.7 .

(2) *Individual variability*: the measure of 10 different samples, drawn from the same clinically healthy subject at different times, has yielded an average value of 42.2 ± 5.0 .

(3) *Sex and age variability*: four groups of 10 subjects each (10 males vs. 10 females aged 4-9 years, and 10 males vs. 10 females aged 14-20 years) have been tested, with the following results:

Age group	Males (N = 20)	Females (N = 20)	Total
4-9 years (N = 20)	30.1 ± 11.6	31.8 ± 10.9	30.9 ± 10.6
14-20 years (N = 20)	39.0 ± 10.8	39.6 ± 8.9	39.3 ± 9.6
Total	34.5 ± 11.8	35.7 ± 10.1	35.1 ± 10.8

The difference between males and females appears to be nonsignificant ($t = 0.34$, 38 *DF*, $P = 0.60$), whereas the difference between the two age groups appears to be significant at a 5% level ($t = 2.56$, 38 *DF*, $0.05 \leq P \leq 0.01$).

HEREDITARY CONDITIONING

The sweat test has then been performed on a total of 40 twin pairs, distributed in eight subgroups of five each, according to sex, age group, and zygosity (5 MZ and 5 DZ male and 5 MZ and 5 DZ female twin pairs aged 4-9 years, vs. 5 MZ and 5 DZ male and 5 MZ and 5 DZ female twin pairs aged 14-20 years). Determination of zygosity was based on serological and other criteria, considering as DZ serologically discordant pairs, and as MZ serologically concordant ones with a probability of dizygosity lower than 0.05 (cf. Smith et al. 1961), besides being concordant with respect to other, nonquantifiable parameters.

Correlation coefficients (r) and estimates of hereditary conditioning (H) of the trait are shown in the following table:

Age group	Males		Females		Total		
	MZ	DZ	MZ	DZ	MZ	DZ	
4-9 years	r	0.705	0.277	0.969	0.108	0.806	0.172
	H	0.59		0.96		0.76	
14-20 years	r	0.068	— 0.138	0.634	— 0.174	0.298	— 0.219
	H	0.18		0.68		0.42	
Total	r	0.508	0.091	0.601	0.053	0.585	0.099
	H	0.46		0.58		0.54	

Probable error of H , $PE_H = \pm 0.094$.

CONCLUSIONS

The findings of an individual variability and of an age variability indicating that environmental factors must play a role in the modifications of Na^+ and Cl^- levels in human sweat, are thus confirmed by the H estimates, showing a hereditary conditioning of approximately 50%. Moreover, when age groups are considered, environmental influence appears to increase with age, the H estimate being higher for the first age group (76%) and lower for the second (42%).

These findings may help to safely apply the sweat test to the clinical and genetic study of mucoviscidosis.

REFERENCES

- Lobeck C.C., Huebner D. 1962. Effect of age, sex, and cystic fibrosis on the sodium and potassium contact of human sweat. *Pediatrics*, 30: 172-177.
- Maynard Smith S., Penrose L.S., Smith C.A.B. 1961. *Mathematical Tables for Research Workers in Human Genetics*. London: J. and E. Churchill.

RIASSUNTO

Ai fini di una migliore utilizzazione clinica e genetica del test del sudore nella mucoviscidosi, gli autori hanno condotto una valutazione della variabilità e del condizionamento genetico del livello degli elettroliti Na⁺ e Cl⁻ nel sudore nell'uomo.

RÉSUMÉ

Afin de mieux utiliser le test de la sueur dans la clinique et la génétique de la mucoviscidose, les auteurs ont évalué la variabilité et le conditionnement héréditaire du niveau des électrolytes Na⁺ et Cl⁻ dans la sueur chez l'homme.

ZUSAMMENFASSUNG

Zur besseren klinischen und genetischen Auswertung des Schweißtests bei Mukoviszidosis errechneten Verf. Variabilität und Erbbedingtheit des Na⁺ und Cl⁻ Elektrolytengehalts im menschlichen Schweiß.

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