The cystic fibrosis gene and resting energy expenditure

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To determine whether the increase in resting energy expenditure in cystic fibrosis is associated with the primary genetic defect (ΔF_{508}) or with declining pulmonary function, or both, we tested resting every energy expenditure prospectively in 32 male subjects (aged 7 to 39 years) who were normally nourished and had good pulmonary function. They were categorized into three genotype groups on the basis of the presence or absence of ΔF_{508} and pancreatic function. Mean resting energy expenditure was 104% of the predicted value and was not associated with genotype. When 29 subjects with normal nutritional status but variable lung function were added to the group, there was a strong correlation between declining pulmonary function and increased resting energy expenditure. We conclude that increased resting energy expenditure in normally nourished boys and men with cystic fibrosis appears to be more closely associated with declining pulmonary function than with genotype. (J PEDIATR 1991;119:913-6)

Several investigators have shown evidence of increased energy expenditure in people with cystic fibrosis,[1-5] a factor that may contribute to undernutrition and growth retardation and thus to an unfavorable prognosis.[6, 7] Some authors have proposed that the increase results from a primary defect in the CF gene product[3, 4]; others have suggested that deteriorating pulmonary function is associated with increased resting energy expenditure.[1, 5] We previously demonstrated, in an unselected group of subjects with CF, elevated REE (119% ± 13%) that appeared to be related to both pulmonary and nutritional status.[1] More recently, we showed that a relationship exists between genotype and phenotype,[3] and that pancreatic sufficiency is a marker of a genotype resulting in less severe disease. We took advantage of the availability of genotype data on our CF clinic population to examine retrospectively the relationship between genotype and REE in the population whose REE we previously reported.[1] Patients (29 male and 26 female) were categorized as either homozygous or heterozygous on the basis of the presence or absence of the major CF mutation (ΔF_{508}).[9-11] The REE in the homozygous Δ_{508} group was 120% ± 23% of that predicted, and in the heterozygous group 111% ± 24% of that predicted. These data are similar to those reported recently by O'Rawe et al.[4]

CF	Cystic fibrosis
FEV_1	Forced expiratory volume in 1 second
FFBM	Fat-free body mass
REE	Resting energy expenditure
WFH	Weight as a percentage of ideal weight for height

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