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PANCREATIC FUNCTION IN CYSTIC FIBROSIS PATIENTS WITH TWO SEVERE CFTR MUTATIONS DECLINES WITHIN THE FIRST YEAR OF LIFE

J. Walkowiak[1], D. Sands[2], A. Nowakowska[2], R Piotrowski[2], K. Zybert[2], K-H. Herzig[3], A. Milanowski[2]

[1]*Institute of Pediatrics, Karol Marcinkowski University of Medical Sciences Poznan, Poland* [2]*Mother and Child Institute, Department of Pediatrics, Warszawa, Poland*
[3]*A.I. VirtanenInstitute for Molecular Sciences and Department of Surgery, University of Kuopio, Finland*
jarwalk@am.poznan.pl

Recent investigations of the genotype in CF patients have revealed a very close correlation with pancreatic phenotype, allowing long-term prognosis. However, there is very little data concerning the decline of exocrine pancreatic function within the first year of life in relation to the genotype. Therefore, we have assessed the decline of pancreatic function in CF infants carrying two "severe" CFTR mutations who were diagnosed in a neonatal screening program. Materials & methods: Twenty-eight CF were included in the study. In all patients, fecal pancreatic elastase-1 (E1) concentrations and fecal fat excretion were determined at diagnosis and at 12 months of age. Results: In all CF patients, E1 concentrations of the first assay after the diagnosis (3-4 months of age) were lower than the cut-off level of <200µg/g stool. However, the presence of steatorrhea was only found in 71.4% of them. At the age of 12 months, all screened CF subjects had E1 concentrations lower than 100µg/g and all were pancreatic insufficient. In conclusion, in CF patients carrying two typical severe CFTR mutations, pancreatic insufficiency develops in early life. Therefore, these patients demand immediate careful monitoring of pancreatic status from diagnosis onwards. The use of fecal elastase-1 tests in the present study allowed for the earlier delineation of patients developing later steatorrhea.