THERAPEUTIC TRIAL IN WOLMAN DISEASE

To the Editor:

I am pleased to learn that Dr. Wolman is still very much active regarding the disease that bears his name[1].

I have so far examined six infants with Wolman's disease; the first patient at the Boston Children's Hospital in 1962 and five others at the Hacettepe Children's Hospital. All the patients had the same compaints, that is, protuberant abdomen, marked hepatosplenomegaly (spleen exceeding liver size), diarrhea and growth retardation commencing shortly after birth. The parents of the children diagnosed at the Hacettepe Children's Hospital were related and three families had a history of another child with similar complaints who died before reaching the age of six months. All these observations were compatible with the recessive inheritance pattern of the disease, in which deficiency of acid esterase-lipase is essential.

As a result of this enzyme deficiency, degradation of triglycerides and cholesterol esters can not take place. Therefore, storage of neutral lipids and cholesterol esters in the hepatocytes and in the adventitial cells of almost all organs occurs which causes the symptomatology[2,3]. Punctate calcifications of enlarged adrenal glands in infants with the above symptomatology are very helpful in diagnosis.

To my knowledge, all children with the infantile form of the disease have died in early infancy, and the replacement of acid esterase has not been tried in this disease, as has been accomplished in Gaucher disease[4,5]. Therefore, any suggestion in treating these children should be welcomed such as Dr. Wolman's, which had previously been proposed differently[6]. Even though the proposed treatment dose not seem to solve the problem, because with the exception of diarrhea most of the symptoms are already present at birth, it is a start.

Dr. Wolman's approach might be criticized from the ethical point of view that "the risk of treatment is not negligeable"[7]. But, I agree with the concept in the editorial published in The Lancet[8], that a new form of threatment should be tried when we are faced with a life-treatening disorder such as in Wolman disease. However, I believe our efforts should be mobilized towards prenatal diagnosis of this disease, since acid esterase can be determined in amniotic fluid[9] and in fibroblasts[6]. Bone marrow transplantation seems to provide hope in Gaucher disease[10,11]. Why not try it in Wolman disease ?

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