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Familial hypercholesterolemia: From experiences of lipid clinic to the international network

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At present, FH is primarily (but not exclusively) considered to be a receptor disease. It is a monogenic disease transmitted through autosomal dominant inheritance and stems from either an LDL-R defect or FDB in cases of PCSK9 mutations. The lipid clinic founded in 1959 was the very first specialist center for FH in the former Czechoslovakia with patients commuting from the all area of 15 million of CZ population. These patients comprised an enormous sample for that time (196 individuals). Currently, the Center of Preventive Cardiology cares for a total of 835 patients with FH (763 adults and 72 children) and it is nowadays the national center of the MedPed project (more than 6000 patients from 90 centers). We are comparing data from the original group (published 1971) with the data from recently studied group. The original study group included a total of 196 people comprised of 94 men and 102 women with a mean age of 38.9 years. It is remarkable that the sample consisted of 103 members of 20 families affected with FH and only 39 individual patients with no relation. The presentation is based on a narrowed sample of 558 adults for whom we have the most complete data. The sample comprises 163 men and 395 women with a mean age of 59.5 ± 18.4 years. The mean age of women is higher at 62.2 ± 17.9 years and the mean age of men is 53 ± 18.0 years. The sample includes 467 probands and 91 family members. Treatment of FH is based on aggressive treatment with lipid lowering drugs, high dose statins and statin/ezetimibe combination. The most frequently used treatment regimen was rosuva 40 mg+ezetimibe and atorva 80mg+ezetimibe. Despite intensive treatment only 38% of patients reached LDL-C goal and 50% LDL-C lowering was reached in 69% of the FH population. LDL apheresis was not used regularly in majority of our FH group. The long term care of FH patients led to development of the Czech Med-Ped (the network of more than 90 lipid clinics), which screened more than 6000 patients with FH (14 homozygotes included). Recently we established the new international project "ScreenPro FH Project" for the screening of FH in the region of Central, Eastern and Southern Europe. This project covers the population of 500 millions of people with the probability of 1-2 millions of patients with FH. We are able to describe the situation in different countries.

Biography

Richard Ceska was graduated at Faculty of Medicine, Charles University in Prague in 1982 and started his medical career at the General University Hospital and one year in Army Hospital in Prague. He is specialized in Internal Medicine and received the first and second degree in Internal Medicine in 1985 and 1989. In 1992 he has received CSc (Czech equivalent of PhD). He has worked as a Fullbright Scholar and Visiting Scientist at The Gladstone Foundation Laboratories for Cardiovascular Disease, San Francisco from 1989-1990. He was named a Professor of Internal Medicine at the Faculty of Medicine, Charles University in Prague in 2006 and is currently the Head of the Center of Preventive Cardiology, Vice-Director of Department of Internal Medicine, Endocrinology and Metabolism. He holds the position of the President of the Czech Society of Internal Medicine and is a Founder and Honorary President of the Czech Society for Atherosclerosis. He is a Secretary of the International Atherosclerosis Society (IAS) and a Member of Administrative Council of the European Federation of Internal Medicine (EFIM). His main scientific interests are internal medicine, preventive cardiology, clinical lipidology and metabolism. He is the first author of 9 monographs and has over 200 scientific publications.

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