[APSAVD] Sponsored Symposium 1
Familial Hypercholesterolemia (FH) -High Risk-

2016年7月14日(木) 8:30-10:30 APSAVD Lecture Room | 南館 4階 錦
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英語

英語セッション

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How can we dig out FH patients in Japan?

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It is an urgent need to prevent early deaths in Familial hypercholesterolemia (FH) families. FH is one of most common autosomal-dominant diseases, and 1 in 200-500 individuals is heterozygous FH. However, it has been reported that just less than 1% of FH patients has been diagnosed in Japan. Our Group has diagnosed more than 2,000 FH patients in Hokuriku district, and this implies 1/3 of FH is diagnosed. Diagnosis is critically important for the early diagnosis of their untreated family members. We have performed genetic test in LDLR gene routinely, but 80% of FH patients are diagnosed without genetic test. This underlines the considerable importance of the effort for clinical diagnosis. Campaign for enlightening on FH has been continued repeatedly over 40 years in this area, and medical staffs are encoura ged to recall FH in all the patients with hypercholesterolemia; we must examine their tendons and ask familial histories. Universal screening for lipid levels should be potentially powerful, but has not been worked well in Japan even with workplace screening presumably because of low-awareness of FH. Cascade screening should be most cost-effective method in the clinics, but it has been noted that this requires significant number of in dex patients to achieve reasonable level of FH detection rate in the population. Combination of several approaches should be organized for the solution. Now EXPLORE-J project has been launched. Consecutive cases with acute coronary syndrome (ACS) in representative cardiovascular centers all over Japan are enrolled.
Achilles' tendon thickness will be examined with X-ray, and also genetic test in cases with written consent. This study will provide how we can identify FH in patients with ACS. There may be no easy way, but awareness raising should be the basis of all possible approaches for the prevention of early deaths in FH.