Kao Hair Science Symposium

Afternoon of the16th, June, 2010

You are cordially invited to a Hair Science Symposium sponsored by Kao Corporation, the leading Hair Care Company in Japan, to be held prior to the opening of the 6th World Conference for Hair Research in Cairns, Australia. In this symposium, the invited special lecturers, Professor George E. Rogers and Dr. Akihiro Fujimoto will present new information about the hair surface structure and hair genes.

Lecturers:

Professor George E. Rogers is Emeritus Professor of the University of Adelaide. Over a period of nearly 50 years he has investigated the structure and biochemistry of the keratin proteins of hair and wool including their genes and aspects of gene expression in hair and wool growth. He has published over 170 papers.

****Dr. Akihiro Fujimoto*** is a researcher of Riken Next Generation Super-Computer Project in Japan. He studied population genetics and bioinformatics. He worked with Prof. Tokunaga in Tokyo University to identify genes related to human adaptations. He performed data analyses and molecular biological experiments, and found genes that are associated with human hair thickness.

Topic 1: The hair cuticle and cornified membranes: Some knowns and unknowns

Professor George E. Rogers, Adelaide University, Adelaide, Australia*

Multidisciplinary research over decades has brought significant understanding of hair and its components from the cellular and macromolecular structure to the hair genes. The cuticle of the hair has important influences on the surface properties of hair, be it care of human hair or textile applications of wool. Research has elicited some of the structural features of cuticle cells but major gaps remain. The basis of resistance of cuticle cells to chemical and physical degradation has been ascribed mainly to two proteins, KAP5 and KAP10, and to disulphide bonds, as well as the surface epicuticle and isopeptide bonding. The epicuticle, about 10nm thick, is a layer isolated from the outermost surface of the cuticle. The KAP 5 and 10 proteins have been localised to the exocuticle by immunoelectron microscopy and by mass spectrometric identification in cuticle cell membranes isolated from wool. Keratin IFs have been localised to the presumptive cuticle although filaments *per se* have not been observed in cuticle cells by electron microscopy (TEM). The endocuticle contains proteins that are not compacted and not chemically resistant like those of the the exocuticle; a protein known as S 100 has been identified in the endocuticle. The basis of hydrophobicity on the hair surface is due to the presence of mainly long-chain fatty acids containining an unusual branched chain moiety known as anteiso 18-methyleiocosanoic acid (MEA, present as approx 50% on dry weight basis). The fatty acids are , covalently bonded (thioester link) to a protein or proteins near the the outermost surface. TEM studies of aberrant human hair from subjects with the genetic defect known as maple syrup urine disease (MSUD) have played a major role in locating MEA exclusively to the the upper surface of all cuticle cells in human hair.. The protein(s) to which the fatty acids are attached are currently unknown, however surveys of proteins in membranous residues isolated from hair suggest that the ultrahigh-sulphur proteins (KAPs 5, and 10) are likely candidates. Claims that involucrin or loricrin could be involved have not been substantiated. The new approaches that retain the protein-lipid linkages have currently been used to investigate some of these issues.

Topic 2: A scan for genetic determinants of human hair morphology with genome-wide SNP database: Association of a nonsynonymous SNP in *EDAR* with Asian hair thickness and its evolution

*Dr. Akihiro Fujimoto**, RIKEN, Tokyo, Japan*

Hair morphology is one of the most differentiated traits among human populations. Genetic backgrounds of hair morphological differences among populations, however, have not yet been clarified. In addition, little is known about the evolutionary forces that have affected hair morphology. To identify hair morphology determining genes, the levels of local genetic differentiation in 170 genes that are related to hair morphogenesis were evaluated by using data from the International HapMap project. Among highly differentiated genes ectodysplasin A receptor (*EDAR*), harboring an Asian-specific non-synonymous single nucleotide polymorphism (1540T/C, 370Val/Ala), was identified as a strong candidate. To examine the association between hair morphology and these candidate genes, we gathered hair and DNA samples in Thailand (Thai-Mai ethnic group), Indonesia and Japanese populations, and established that the Asian-specific 1540C allele is strongly associated with increase in hair thickness (multiple regression analysis considering age, sex and population; P-value = 3.8×10-10). Reporter gene assays suggested that 1540T/C affects the activity of the downstream transcription factor NF-kB. It was inferred from geographic distribution of 1540T/C and the long-range haplotype test that 1540C arose after the divergence of Asians and Europeans and its frequency has rapidly increased in East Asian populations. These findings lead us to conclude that *EDAR* is a major genetic determinant of Asian hair thickness and the 1540C allele spread through Asian populations due to positive selection. Recent studies revealed that 1540T/C is also associated with Asian teeth shape and that enhancement of Edar activity causes change in hair and several glands morphology in mice. Although the selective pressure that has acted on 1540C is still not clear, the polymorphism in *EDAR* may play a key roll in the adaptation of Asian ancestors to their environments.