HOKKAIDO UNIVERSITY

Title	Human Genetic Disorders Caused by Mutations in Genes Encoding Biosynthetic Enzymes for Sulfated Glycosaminoglycans
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Shuji MIZUMOTO

1) SHORT BIOGRAPHICAL SUMMARY

I began to learn glycobiology of glycosaminoglycans (GAGs) and proteoglycans from Prof. Kazuyuki Sugahara at Kobe Pharmaceutical University during my thesis, working on sulfotransferases involved in the biosynthesis of chondroitin sulfate (CS) and dermatan sulfate (DS). During my postdoctoral work at Hokkaido University, I was fortunate to find several genetic disorders including Larsen-like syndrome, spondyloepiphyseal dysplasia-Omani type, and Ehlers-Danlos syndrome caused by the mutation in the genes of GAG-biosynthetic enzymes collaborating with medical doctors and geneticists. My resent research interests are human genetic diseases, tumor metastasis, and structure-activity correlation of GAG chains and proteoglycans.

2) CURRENT POSITION

a. title : Postdoctoral fellow

b. department: Laboratory of Proteoglycan Signaling and Therapeutics, Faculty of Advanced Life Science,

c. institution: Hokkaido University, Hokkaido, Japan

3) EDUCATION

a. highest degree granted: Ph.D.

b. degree discipline: Pharmaceutical Sciences

c. year degree was granted: 2006

d. degree-granting institution: Kobe Pharmaceutical University, Kobe, Japan

(4) NONSCIENTIFIC INTERESTS

scuba diving, baseball, music

(5) CURRENT PHOTO

Author Profile

1) SHORT BIOGRAPHICAL SUMMARY Please include how the author became interested in this area of research.

Dr. Shiro IKEGAWA started his professional carrier as an orthopedic surgeon in 1983 after he graduated from Faculty of Medicine, University of Tokyo. He led the special clinic for skeletal dysplasia (monogenic bone and joint diseases) in Department of Orthopedic Surgery, Tokyo University Hospital since 1987. He was a chief surgeon of Department of Orthopedics, National Rehabilitation Center for Disabled Children in 1994 when he abandoned his carrier as a surgeon and become a student of Department of Biochemistry, Cancer Institute to study genome science. It was because he recognized the coming of the genome era. Dr. Ikegawa became Assistant Professor of Institute of Medical Science, University of Tokyo in 1995. He became Laboratory Head (PI) of the current laboratory in RIKEN in 2000. His research interest has been lying consistently on genetic bone and joint diseases since he was a surgeon.

2) CURRENT POSITION

a. title team leader

b. department Lab. of Bone and Joint Diseases

c. institution Center for Genomic Medicine, RIKEN

3) EDUCATION

a. highest degree granted

Doctor (DMSc)

b. degree discipline

Medical Science

c. year degree was granted

1996

d. degree-granting institution

Tokyo University

(4) NONSCIENTIFIC INTERESTS

Rugby football

Kazuyuki Sugahara

I learned about the biochemistry of proteoglycans (PGs) and glycosaminoglycans (GAGs) from Albert Dorfman and Nancy Schwartz at the University of Chicago, while working on the biosynthesis of hyaluronan and chondroitin sulfate (CS). After accepting an instructor position at Kyoto University in 1982 and then an associate professor position at the Kobe Women's College of Pharmacy (now the Kobe Pharmaceutical University) in Japan in 1990, I focused my studies on the structure, biosynthetic mechanism, and biological functions of GAGs, including CS and heparan sulfate (HS). My early postdoctoral work, which motivated me to work on the so-called common GAG-protein linkage region tetrasaccharide (GlcUA-Gal-Gal-Xyl) found in different GAG species, led me to the discovery of a sulfate group on the Gal residue in CS chains in 1988. This sulfate has not been found in HS. This work was the foundation for my interest in the molecular mechanism underlying the biosynthetic differential assembly of CS and HS chains built on the differently modified linkage structure. Molecular cloning of various glycosyltransferases and sulfotransferases in Kobe with Hiroshi Kitagawa and many other colleagues led me to my current interest in the genetic disorders caused by the aberrant synthesis of GAG side chains of PGs.

2) CURRENT POSITION
a. title : Professor Emeritus.
b. department: Laboratory of Proteoglycan Signaling and Therapeutics, Faculty of Advanced Life Science.
c. institution: Hokkaido University, Sapporo, Japan.

3) EDUCATION
a. highest degree granted: Ph.D.
b. degree discipline: Pharmaceutical Sciences.
c. year degree was granted: 1976.
d. degree-granting institution: Kyoto University, Kyoto, Japan.

(4) NONSCIENTIFIC INTERESTS: Singing and dancing a major form of classical (since the 14th century) Japanese musical drama (Noh play).

(5) CURRENT PHOTO: Separately attached.

Summary:

Current position: Professor Emeritus, Laboratory of Proteoglycan Signaling and Therapeutics, Faculty of Advanced Life Science, Hokkaido University, Sapporo, Hokkaido, Japan.

Education: Ph.D. in Pharmaceutical Sciences, 1976, Kyoto University, Kyoto, Japan, where he demonstrated the beta-mannosidic linkage for the innermost mannose in the *N*-linked oligosaccharides of the glycopeptides isolated from ovalbumin.

Nonscientific interests: Singing and dancing a major form of classical (since the 14th century) Japanese musical drama (Noh play).