

**OUR MISSION IS TO
OVERCOME THIS INTRACTABLE DISEASE -TGCV**



**THE 2nd INTERNATIONAL SYMPOSIUM on
Triglyceride Deposit Cardiomyovascuopathy
&
Neutral Lipid Storage Disease**

PROGRAM & ABSTRACTS

DATE: April 19-20, 2013

VENUE: Osaka University Nakanoshima Center 10F

PRESIDENT: Ken-ichi Hirano Osaka University



* These photos were kindly provided from Dr. Michio Tanaka.



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PRESIDENT: Ken-ichi Hirano Osaka University

- 17:00-17:12** Chairperson: Daniella Tavian, Catholic University of the Sacred Heart, Italy
- P-1 Triglyceride deposit cardiomyovasculopathy with severe heart failure
Naoko Takada, Tohoku University, Japan
- P-2 A genetic mutation of adipose triglyceride lipase accumulates in a small village in Tohoku District, Japan
Akira Suzuki, Osaka University, Japan
- 17:12-17:30** Chairperson: Ken-ichi Hirano, Osaka University, Japan
- P-3 Triglyceride deposit atherosclerosis with infiltration of less intimal macrophage foam cells in TG deposit cardiomyovasculopathy
Yoshihiko Ikeda, National Cerebral and Cardiovascular Center, Japan
- P-4 Phenotypic plasticity of vascular smooth muscle cells isolated from adipose triglyceride lipase-deficient mice
Yanhui Lin, University of the Ryukyus, Japan
- P-5 Biomarker discovery for triglyceride deposit cardiomyovasculopathy using proteome and transcriptome analysis
Yasuhiro Hara, National Institute of Biomedical Innovation, Japan
- 17:30-17:48** Chairperson: Hitoshi Chiba, Hokkaido University, Japan
- P-6 A pathophysiological model and rationale for possible treatment with medium chain fatty acids for human adipose triglyceride lipase deficiency
Ken-ichi Hirano, Osaka University, Japan
- P-7 Detection of medium-chain triglyceride in fibroblast from adipose triglyceride lipase deficiency by Orbitrap LC/MS
Shu-Ping Hui, Hokkaido University, Japan
- P-8 Detection of medium- and long-chain fatty acids accumulation by triglyceride deposit cardiomyovasculopathy-indicated fibroblasts using fast spectral coherent anti-Stokes Raman scattering microscopy
Harsono Cahyadi, Osaka University, Japan
- 17:48-18:00** Chairperson: Shigetoshi Sano, Kochi University, Japan
- P-9 First large deletion in ABHD5 promoter region causing Chanarin-Dorfman syndrome
Sara Missaglia, Catholic University of the Sacred Heart, Italy
- P-10 Epidermal triglyceride levels are correlated with severity of ichthyosis in Dorfman-Chanarin syndrome
Kimiko Nakajima, Kochi University, Japan
- 18:00-18:18** Chairperson: Yasuyuki Nagasawa, Hyogo College of Medicine, Japan
- P-11 Screening for triglyceride deposit cardiomyovasculopathy in type 2 diabetes mellitus patients
Nobuhiro Zaima, Kinki University, Japan
- P-12 The biochemical characterization of human adipose triglyceride lipase (ATGL) and development of its specific measurement system: A property of his6-ATGL expressed in COS1 cells
Atsuko Takagi, National Cerebral and Cardiovascular Center Research Institute, Japan
- P-13 Adipose triglyceride lipase is downregulated in db/db mouse heart – An aggravating mechanism of diabetic cardiomyopathy
Tomoaki Inoue, Fukuoka City Medical Association Hospital, Japan
- 18:18-18:30** Chairperson: Nobuhiro Zaima, Kinki University, Japan
- P-14 Establishment of the personification-mouse model with the TGCV patient specific iPS cells
Shunmei Chiba, University of Ryukyus, Japan
- P-15 The world smallest Microminipigs: Type 1 diabetes mellitus induced by streptozotocin
Hiroaki Kawaguchi, Kagoshima University, Japan

18:40- Reception (*buffet-style/9F SALON DE L'AMICAL*)

9:00-10:00

O1. Neutral lipid storage disease (NLSD)/triglyceride deposit cardiomyo- vasculopathy (TGCV)

Chairpersons: Matthijs Hesselink, Maastricht University, The Netherlands
Ken-ichi Hirano, Osaka University, Japan

- 1) Genotype-phenotype correlation in neutral lipid storage disease with myopathy
Daniella Tavian, Catholic University of the Sacred Heart, Italy
- 2) Skin abnormality in Dorfman-Chanarin syndrome
Shigetoshi Sano, Kochi University, Japan

10:00-11:00

O2. Possible therapeutic strategies for adipose triglyceride lipase deficiency

Chairpersons: Masatsugu Hori, Osaka, Japan
Elena Pennisi, A.C.O. San Filippo NERI-Rome, Italy

- 1) Effects of bezafibrate treatment in two sisters with mutations in the PNPLA2 gene, causing Neutral Lipid Storage Disease with Myopathy
Norihide Fukushima, Osaka University, Japan
- 2) Effects of bezafibrate treatment in two sisters with mutations in the PNPLA2 gene, causing Neutral Lipid Storage Disease with Myopathy
Matthijs Hesselink, Maastricht University, The Netherlands
- 3) Multi-disciplinary therapeutic approach for Adipose Triglyceride Lipase Deficiency
Ken-ichi Hirano, Japan TGCV Study Group, Japan

(Preparation for web conference)

11:10-12:20

O3. From rare disease to common disease

- Cardiomyovascular TG deposition in diabetes mellitus -

Chairpersons: Kunihisa Kobayashi, Fukuoka University, Japan
Ken-ichi Hirano, Osaka University, Japan

Keynote lecture: Diabetic cardiovascular disease (Web)
Joel D. Schilling, Washington University, USA

- 1) Impact of diabetes mellitus on the fatty heart: learning from autopsy cases
Seiya Kato, University of the Ryukyus, Japan
- 2) Human coronary atherosclerosis with triglyceride-deposit smooth muscle cells
Yoshihiko Ikeda, National Cerebral and Cardiovascular Center, Japan
- 3) Quantitative coronary CT angiography: A proposed system for assessment of lipid accumulation in coronary arterial wall
Hiroaki Naito, National Cerebral and Cardiovascular Center, Japan

12:20-

Closing Remarks: Ken-ichi Hirano, Chairman of the Symposium

15:00-

Seminar for General Public