The 6th Ann Mtg (2003) – International Symposium – Chromosomal Aberrations and Childhood Epilepsies

Date March 15-16, 2003

Venue Lecture Hall,
Tokyo Women's Medical University, Tokyo, Japan
8-1, Kawada-cho, Shinjuku-ku,
Tokyo 162-8666, Japan

Day 1 **Saturday, March 15** ☛ Day 2

08:30	**REGISTRATION**
09:00	**Opening Address** ○ **Yukio FUKUYAMA**

【Overview】

Chair: Yukio FUKUYAMA

09:05 **1**	**CHROMOSOMAL ABNORMALITIES AND EPILEPSY: CLUES TO FINDING EPILEPSY GENES ?** ○ **Samuel F BERKOVIC** **Epilepsy Research Institute, University of Melbourne, Austin & Repatriation Medical Centre, Victoria, Australia**

【GENERAL】

Chairpersons : John BP STEPHENSON
Makiko OSAWA

09:45 **2**	**ELECTRO-CLINICAL PHENOTYPES OF RARE CHROMOSOME DISORDERS ASSOCIATED WITH EPILEPSY - MANY CHILDREN ARE NOT DYSMORPHIC -** ○ **Sameer M ZUBERI①, Stewart MACLEOD①, Arup MALIK②, Mary E O'REGAN①, Paul EUNSON③, Ailsa MCLELLAN③** **①Fraser of Allander Neurosciences Unit, Royal Hospital for Sick Children, Glasgow, UK ②Department. of Clinical Neurophysiology, Southern General Hospital, Glasgow, UK ③Paediatric Neurosciences, Royal Hospital for Sick Children, Edinburgh, UK**
10:05 **3**	**A HOSPITAL-BASED STUDY OF EPILEPSY IN PATIENTS WITH CHROMOSOMAL ABNORMALITIES** ○ **Yasuhisa TORIBE①, Kana SANTO①, Hitoshi UEDA①, Nobuhiko OKAMOTO②, Yasuhiro SUZUKI ①** **①Division of Pediatric Neurology, and ②Department of Planning and Research, Osaka Medical Center and Research Institute for Maternal and Child Health, Izumi, Osaka, Japan**
10:20 **4**	**AN ANALYSIS OF CHROMOSOMAL ABERRATIONS AND EPILEPSIES IN DOKKYO UNIVERSITY SCHOOL OF MEDICINE** ○ **Hideo YAMANOUCHI, George IMATAKA, Eiji NAKAGAWA, Naomitsu SUZUKI, Akihisa NITTA, Jun-ichi HIRAO, Hiroshi SUZUMURA, *Hiroshi WATANABE, Mitsuoki EGUCHI** **Department of Pediatrics and *Obstetrics, Dokkyo University School of Medicine Tochigi, Japan**

10:35 **5**	**MULTI-INSTITUTIONAL STUDY ON THE CORRELATION BETWEEN CHROMOSOME ABERRATIONS AND EPILEPSY** ○ **Tomohiro KUMADA①②, Masatoshi ITO①②, Tomoko MIYAJIMA①②, Tatsuya FUJII①②, Takehiko OKUNO①②, Tohshin GO②, Haruo HATTORI②, Mieko YOSHIOKA②, Kenichiro KOBAYASHI②, Osamu KANAZAWA②, Jun TOHYAMA②, Noriyuki AKASAKA②, Takanori KAMIMURA②, Mutsuo SASAGAWA②, Hiroshi TANAKA②, Hideki AMAGANE②, Kozo MUTOH②, Yuriko YAMORI②, Toyoko KANDA②, Naoko YOSHIDA②, Haruyo HIROTA②, Rieko TANAKA②, Yasushi HAMADA②** **①Department of Pediatrics, Shiga Medical Center for Children, Shiga, Japan** **②Kyoto Multi-institutional Study Group of Pediatric Neurology, Kyoto, Japan**

【CHROMOSOME 1p DELETION】

Chairpersons : Agatino BATTAGLIA
Takao TAKAHASHI

10:50 **6**	**REFINEMENT OF A 1 MB CRITICAL REGION FOR 1P36 DELETION SYNDROME** ○ **Kenji KUROSAWA** **Division of Medical Genetics, Kanagawa Children's Medical Center, Yokohama, Japan**

【CHROMOSOME 4p DELETION】

11:10 **7**	**WOLF-HIRSCHHORN (4p-) SYNDROME** ○ **Agatino BATTAGLIA** **Stella Maris Clinical Research Institute for Child and Adolescent Neurology and Psychiatry, Calambrone, Pisa, Italy**
11:30 **8**	**Chairpersons : L.A.E.M. LAAN** **Kenji SUGAI** **EPILEPSY IN WOLF-HIRSCHHORN SYNDROME (4P- SYNDROME)** ○ **Kuriko SHIMONO①, Katsumi IMAI①, Kazumasa OTANI②, Noriko KAMIO①, Takeshi OKINAGA①, Yasuhisa TORIBE③, Yasuhiro SUZUKI③, Keiichi OZONO①** **①Department of Pediatrics, Osaka University Graduate School of Medicine, Osaka, Japan** **②Otani Clinic, Gobo, Wakayama, Japan** **③Division of Pediatric Neurology, Osaka Medical Center and Research Institute for Maternal and Child Health, Izumi, Osaka, Japan**
11:45 **9**	**AN ADULT CASE OF 4p - SYNDROME PRESENTING HEMI-CONVULSION ―HEMIPLEGIA―EPILEPSY SYNDROME** ○ **Yasushi ITO①②, Satoko KUMADA①, Akira UCHIYAMA①, Hirohumi OHASHI③, Masaharu HAYASHI④, Kiyoko KURATA①, Kimiko TAMAGAWA⑤, Makiko OSAWA②** **①Department of Pediatrics, Metropolitan Fuchu Medical Center for Severe Motor and Intellectual Disabilities, Tokyo, Japan** **②Department of Pediatrics, Tokyo Women's Medical University, School of Medicine, Tokyo, Japan** **③Division of Medical Genetics, Saitama Children's Medical Center, Saitama, Japan** **④Department of Clinical Neuropathology, Tokyo Metropolitan Institute for Neuroscience, Tokyo, Japan** **⑤Department of Neuropediatrics, Tokyo Metropolitan Neurological Hospital, Tokyo, Japan**

12:00 **10**	**CLONIC STATUS EPILEPTICUS IN THREE CASES WITH CHROMOSOMAL OR GENE ABNORMALITIES: VIDEO-EEG DEMONSTRATION** ○ **Osamu KANAZAWA, Jun TOHYAMA, Noriyuki AKASAKA, Takanori KAMIMURA** **Department of Pediatrics, Epilepsy Center, National Nishi-Niigata Central Hospital, Niigata, Japan**
12:00-13:30	**LUNCH**
【ANGELMAN SYNDROME I】 **Chairperson : Shunsuke OHTAHARA**	
13:30 **11**	**ANGELMAN SYNDROME: CLINICAL PRESENTATIONS, DIAGNOSTIC CRITERIA AND NEUROGENETIC ASPECTS** ○ **Charles A. WILLIAMS** **Division of Genetics, Department of Pediatrics, University of Florida, Gainesville, U.S.A.**
【ANGELMAN SYNDROME II】 **Chairpersons : Charles A. WILLIAMS Shinji SAITOH**	
14:10 **12**	**ANGELMAN SYNDROME PATIENTS WITH UNI-PARENTAL DISOMY OR IMPRINTING DEFECTS IN JAPAN** ○ **Shinji SAITOH①, Takahito WADA②, Maki OKAJIMA①, Kyoko TAKANO①, Akira SUDO①, Norio NIIKAWA③** **①Department of Pediatrics, Hokkaido University, Sapporo, Japan ②Department of Preventive Medicine, Shinshu University, Matsumoto, Japan ③Department of Human Genetics, Nagasaki University, Nagasaki, Japan**

14:30 **13**	**NEUROLOGICAL ASPECTS OF ANGELMAN SYNDROME IN CHILDHOOD** ○ **Laura A.E.M. LAAN** **Department of Neurology, Leiden University Medical Center, Leiden, The Netherlands**
14:50 **14**	**Asian Research Award Lecture --- ELECTROCLINICAL CHARACTERISTICS OF SEIZURES ： COMPARING PRADER-WILLI SYNDROME WITH ANGELMAN SYNDROME** ○ **Pen-Jung WANG①②, Wang-Tso LEE②, Whey-Chen SUE③, Jia-Woei HOU②④** **①Department of Pediatrics, Tzu Chi Medical Center, Hualein, Taiwan** **②Department of Pediatrics, National Taiwan University Hospital, Taipei, Taiwan** **③Department of Pediatrics, Taipei Municipal Women and Children Hospital, Taipei, Taiwan** **④Department of Medical Genetics, Chang Gung Children's Hospital, Taipei, Taiwan**
【CONSENSUS DEVELOPMENT SEMINAR ON "ANGELMAN EEG"】 **Chairperson : Yukio FUKUYAMA**	
15:10 **15**	**WHAT IS A TYPICAL " ANGELMAN EEG" ?** **HOW FAR IS IT SPECIFIC FOR AS?** **Main speaker: Laura A.E.M. LAAN** **Department of Neurology, Leiden University Medical Center, Leiden, The Netherlands** **Designated discussant: Hirokazu OGUNI** **Department of Pediatrics, Tokyo Women's Medical University, Tokyo, Japan**
15:10-16:10	**INTERMISSION**

【ANGELMAN SYNDROME III】

Chairpersons : Samuel F.BARKOVIC
Eiji OKA

16:10 **16**	**THE RELATIONSHIP BETWEEN SEVERITY OF EPILEPSY AND DEVELOPMENTAL OUTCOME IN ANGELMAN SYNDROME** ○ **Yoko OHTSUKA, Katsuhiro KOBAYASHI, Tatsuya OGINO, Harumi YOSHINAGA, Eiji OKA** **Department of Child Neurology, Okayama University Graduate School, Okayama, Japan**
16:25 **17**	**THE EPILEPSIES OF ANGELMAN SYNDROME - A LONG-TERM FOLLOW UP -** ○ **Naoko UEMURA①, Akiko MATSUMOTO②, Toshiyuki KUMAGAI③, Kiyokuni MIURA③, Takashi OHKI③, Seiji MIZUNO①, Miho NAKAMURA④, Kazuyoshi WATANABE⑤, Tamiko NEGORO⑤, Akihisa OKUMURA⑤, Kohzaburo ASO⑥, Fumio HAYAKAWA⑦, Yoko KONDO⑧** **①Department of Pediatrics, Aichi Human Service Centre, Kasugai, Aichi, Japan** **②Kobato Gakuen , Aichi Human Service Centre, Kasugai, Aichi, Japan** **③Department of Pediatric Neurology, Aichi Human Service Centre, Kasugai, Aichi, Japan** **④Institute for Developmental Research, Aichi Human Service Centre, Kasugai, Aichi, Japan** **⑤Department of Pediatrics, Nagoya University Graduate School of Medicine, Nagoya, Aichi, Japan** **⑥Department of Pediatrics, Aichi Prefecture Medical Welfare Center of Aoitori Gakuen, Nagoya, Aichi, Japan** **⑦Department of Pediatrics, Okazaki City Hospital, Okazaki, Aichi, Japan** **⑧Department of Pediatrics,Gifu Social Insurance Hospital, Kani, Gifu, Japan**

16:40 **18**	**MYOCLONIC ABSENCE STATUS EPILEPTICUS IN TWO PATIENTS WITH ANGELMAN SYNDROME** ○ **Tatsuro IZUMI, Shin-ichi UCHIYAMA, Makoto KOIZUMI, Kazuhide IMAI, Aya NYU, Masayoshi SUZUKI** **Division of Pediatrics and Child Neurology, Department of Brain and Nerve Science, Oita Medical University School of Medicine, Oita, Japan**
16:55 **19**	**NEURORADIOLOGICAL FINDINGS OF ANGELMAN SYNDROME** ○ **Shigeru HANAOKA, Kenji SUGAI** **Department of Child Neurology, National Center Hospital for Mental, Nervous and Muscular Disorders, National Center of Neurology and Psychiatry, Kodaira, Tokyo, Japan**

【INVERSION DUPLICATION 15 SYNDROME】

Chairpersons : Peng-Jung WANG
Hirokazu OGUNI

17:10 **20**	**THE 15q13 TETRASOMY SYNDROME ASSOCIATED WITH EHLERS-DANLOS SYNDROME.** ○ **Reiko MUTO, Shigeru YANAGAKI, Kitami HAYASHI, Makoto FUNATSUKA, Masako SAKAUCHI, Seigo SHIRAKAWA, Emiko TACHIKAWA, Hirofumi OHASHI*, Makiko OSAWA** **Department of Pediatrics, Tokyo Women's Medical University, School of Medicine, Tokyo, Japan** ***Division of Medical Genetics Saitama Children's Medical Center, Iwatsuki, Saitama, Japan**

21	**THE INV DUP(15) SYNDROME** ○ **Agatino BATTAGLIA** **Stella Maris Clinical Research Institute for Child and Adolescent Neurology and Psychiatry, Calambrone, Pisa, Italy**
【CORTICAL MIGRATIONAL DISORDERS】 **Chairperson : Kazuyoshi WATANABE**	
17:45 **22**	**CHROMOSOMAL ABNORMALITIES AND DISORDERS OF CORTICAL MIGRATION AND ORGANIZATION** ○ **Eva ANDERMANN** **Neurogenetics Unit, Montreal Neurological Institute & Hospital, McGill University, Montreal, Canada**
18:20	**To be closed**
18:30	**SOCIAL PARTY at "ALFARO"**

Day 2 **Sunday, March 16**

【WEST SYNDROME】

Chairpersons : Pratibha SINGHI
Kazuie IINUMA

09:00
23

Asian Research Award Lecture
-- PROFILE OF WEST SYNDROME IN NORTH INDIA

○ **Pratibha SINGHI, Munni RAY, Gaurva GUPTA**

Department of Pediatrics, Postgraduate Institute of Medical Education and Research, Chandigarh, India.

09:15
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MUTATIONS OF ARX CAUSE SYMPTOMATIC AND NON-SYMPTOMATIC OR CRYPTOGENIC WEST SYNDROME

○ **Mitsuhiro KATO①④, Ying PENG①, Soma DAS①, Kristin PETRAS①, Yukio SAWAISHI⑤, William B. DOBYNS①-③**

Departments of ①Human Genetics, ②Neurology and ③Pediatrics, The University of Chicago, Chicago, IL, USA;
④Department of Pediatrics, Yamagata University School of Medicine, Yamagata, Japan;
⑤Department of Pediatrics, Akita University School of Medicine, Akita, Japan

【CHROMOSOME 18q DELETION】

09:35
25

AUTONOMIC SEIZURES IN 18q- SYNDROME

○ **John BP STEPHENSON**

Fraser of Allander Neurosciences Unit, Royal Hospital for Sick Children, Glasgow, UK

【RING CHROMOSOME 20】

Chairpersons : Sameer M ZUBERI
Tateki FUJIWARA

09:50 **26**	**EPILEPSY IN PATIENTS WITH RING CHROMOSOME 20** ○ **Yushi INOUE** **National Epilepsy Center, Shizuoka Medical Institute of Neurological Disorders, Shizuoka, Japan**
10:15 **27**	**PRESENTATION, CLINICAL EVOLUTION AND OUTCOME IN RING CHROMOSOME 20 SYNDROME** ○ **Sameer M.ZUBERI①, Arnaud J.BIRABEN②,** **On behalf of the International Ring Chromosome 20 Collaboration** **①Fraser of Allander Neurosciences Unit, Royal Hospital for Sick Children, Glasgow, UK** **②Unité d'Épileptologie, CHU de Rennes, Rennes, France**
	Chairpersons : Arnaud J. ABIRABEN **Tunekazu YAMANO**
10:40 **28**	**STRIATAL DOPAMINE METABOLISM IS DECREASED IN PATIENTS WITH EPILEPTIC SEIZURES ASSOCIATED WITH RING CHROMOSOME 20 SYNDROME. RESULTS OF A PET STUDY IN 14 PATIENTS** ○ **Arnaud J.BIRABEN①, Franck SEMAH②** **①Unité d'Épileptologie, Centre Hospitalier Universitaire de Rennes, Rennes, France** **②Service Hospitalier Frédérique Joliot-Curie, Centre de L'Energie Antomique, Orsay, France**
11:05 **29**	**A 5-YEAR-OLD GIRL WITH RING 20 CHROMOSOME SYNDROME PRESENTING CHARACTERISTIC FRONTAL LOBE EPILEPSY** ○ **Kaori SASAKI, Hirokazu OGUNI, Kaoru ETO, Tomoko USUGI, Makoto FUNATSUKA, Makiko OSAWA** **Department of Pediatrics, Tokyo Women's Medical University, Tokyo, Japan**

【COFFIN-LOWRY SYNDROME】

Chairpersons : John STEPHENSON
Hideo YAMANOUCHI

11:20
30

THE MOVEMENT DISORDERS OF COFFIN-LOWRY SYNDROME

○ **John STEPHENSON①, Mary HOFFMAN②, Richard BEACH③, Jane FALCONER④, Robert McWILLIAM①, Aline RUSSELL⑤, Sameer ZUBERI①**

①Fraser of Allander Neurosciences Unit, Royal Hospital for Sick Children, Glasgow, UK
②Coffin-Lowry Syndrome Foundation, Sammamish, WA, USA
③Paediatric Unit, Norfolk and Norwich Hospital, Norfolk, UK
④Shebburn Surgery, New Abbey, Dumfries, UK
⑤Neurophysiology Department, Southern General Hospital, Glasgow, UK

11:35
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DROP EPISODES IN COFFIN-LOWRY SYNDROME: EXAGGERATED STARTLE RESPONSES TREATED WITH CLONAZEPAM

○ **Miki NAKAMURA①, Takanori YAMAGATA①, Mariko MOMOI①, Toyo YAMAZAKI②**

①Department of Pediatrics, Jichi Medical School, Tochigi, Japan
②Yamazaki Pediatric Clinic, Tochigi, Japan

【KABUKI SYNDROME, 22q11.2 DELETION SYNDROME】

Chairpersons : Eva ANDERMANN
Shinichi NIIJIMA

11:50 **32**	**FAVORABLE SEIZURE OUTCOME IN KABUKI MAKE-UP SYNDROME ASSOCIATED WITH EPILEPSY** ○ **Atsushi OGAWA①, Sawa YASUMOTO①, Yasuko TOMODA①, Masaharu OHFU①, Akihisa MITSUDOME①, Yoshikazu KUROKI②** **①Department of Pediatrics, School of Medicine, Fukuoka University, Fukuoka, Japan** **②Division of Medical Genetics, Kanagawa Children's Medical Center, Kanagawa, Japan**
12:05 **33**	**SEIZURE PROBLEMS IN THE 22q11.2 DELETION SYNDROME** ○ **Ayako MUTO①, Hirokazu OGUNI①, Makoto FUNATSUKA①, Kitami HAYASHI①, Makiko OSAWA①, Rumiko MATSUOKA②,** **①Department of Pediatrics, Tokyo Women's Medical University, Tokyo, Japan** **②Department of Pediatric Cardiology , Tokyo Women's Medical University, Tokyo, Japan**
12:20	**CLOSING REMARKS & ADJOURN** ○ **Makiko OSAWA**
12:30-14:00	**BUSINESS MEETING OF THE COUNCILLORS**