

## Invited Plenary Lecture, JSPE International Prize

October, 30(Sat.) 15:00 ~ 16:10 Room 1

Chair Keiichi Ozono (Department of Pediatrics, Osaka University Graduate School of Medicine)

### **Making Diagnoses in Short Stature of Undefined Cause**

Peter E Clayton<sup>1,2</sup>

<sup>1</sup>Division of Developmental Biology & Medicine, Faculty of Biology, Medicine & Health, University of Manchester

<sup>2</sup>Royal Manchester Children's Hospital, Manchester University Hospitals NHS Foundation Trust, UK

## KSPE-JSPE Plenary Lecture

October, 28(Thu.) 10:50 ~ 11:40 Room 1

Chair Tomonobu Hasegawa (Department of Pediatrics, Keio University School of Medicine)

### **Applications of genomics in pediatric endocrine diseases**

Jin-Ho Choi, MD

Department of Pediatrics, Asan Medical Center, University of Ulsan College of Medicine, Seoul, Korea

## English Oral session 1

October, 29(Fri.) 9:40 ~ 10:30 Room 4

Chairs Akie Nakamura (Department of Pediatrics, Hokkaido University School of Medicine)

Kosei Hasegawa (Department of Pediatrics, Okayama University Hospital)

### **EO1-1 Functional analysis of PAX8 variants identified in patients with congenital hypothyroidism**

Khishigjargal Batjargal<sup>1,3</sup>, Toshihiro Tajima<sup>1</sup>, Eriko Jimbo<sup>1</sup>, Takeshi Yamaguchi<sup>2</sup>, Akie Nakamura<sup>2</sup>, Takanori Yamagata<sup>1</sup>

<sup>1</sup>Dept. of Pediatr., Jichi Medical University, Tochigi, Japan,

<sup>2</sup>Dept. of Pediatr., Hokkaido University Graduate School of Medicine, Sapporo 060-8638, Japan.,

<sup>3</sup>Dept. of Pediatr., Mongolian National University of Medical Sciences, Ulaanbaatar, Mongolia.

### **EO1-2 Measurement of reverse T3 level and the T3 to reverse T3 ratio in dried blood spot samples at birth may facilitate early detection of monocarboxylate transporter 8 deficiency**

Hideyuki Iwayama<sup>1</sup>, Hiroki Kakita<sup>2</sup>, Masumi Iwasa<sup>1</sup>, Shinsuke Adachi<sup>3</sup>, Kyoko Takano<sup>4</sup>,

Masahiro Kikuchi<sup>5</sup>, Yasuko Fujisawa<sup>6</sup>, Hitoshi Osaka<sup>7</sup>, Yasumasa Yamada<sup>2</sup>, Akihisa Okumura<sup>1</sup>, Khemraj Hirani<sup>8</sup>, Roy Weiss<sup>8</sup>, Samuel Refetoff<sup>9</sup>

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<sup>3</sup>Dept. of Pediatr., Fukushiyama City Hosp., <sup>4</sup>Cent. for Med. Genet., Shinshu Univ. Hosp.,

<sup>5</sup>Dept. of Pediatr., Hitachi Gener. Hosp., <sup>6</sup>Dept. of Pediatr., Hamamatsu Univ. Sch. of Med.,

<sup>7</sup>Dept. of Pediatr., Jichi Med. Sch., <sup>8</sup>Dept. of Med., Univ. of Miami Miller Sch. of Med.,

<sup>9</sup>Dept. of Med., Univ. of Chicago

### **EO1-3 Central precocious puberty in a boy with pseudohypoparathyroidism type 1B**

Vlad Tocan, Yuichi Mushimoto, Kanako Kojima-Ishii, Naoko Toda, Shouichi Ohga

Dept. of Pediatr., Grad. School of Med. Sci., Kyushu Univ.

**EO1-4 The elucidation of genetic and phenotypic features of the mitochondrial genome in the Japanese population**

Kenichi Yamamoto<sup>1,2</sup>, Saori Sakaue<sup>2,3,4</sup>, Koichi Matsuda<sup>5</sup>, Yoshinori Murakami<sup>6</sup>,  
Yoichiro Kamatani<sup>3,7</sup>, Keiichi Ozono<sup>1</sup>, Yukihide Momozawa<sup>8</sup>, Yukinori Okada<sup>2,9,10</sup>

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<sup>5</sup>Dept. of Comput. Biol. and Med. Sci., Grad. Sch. of Front. Sci.,

<sup>6</sup>Div. of Mol. Path., the Inst. of Med. Sci., the Univ. of Tokyo,

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<sup>9</sup>Lab. of Stat. Immunol., Immunol. Front. Research Center,

<sup>10</sup>Integ. Front. Research for Med. Sci. Div., Institute for Open and Transdisciplinary Research Initiatives

**EO1-5 Haploinsufficiency of DMRT1 causes 46,XY gonadal dysgenesis**

Misa Honda<sup>1</sup>, Mizue Tsukui<sup>2</sup>, Hironori Shibata<sup>1</sup>, Satoshi Narumi<sup>3</sup>, Tomohiro Ishii<sup>1</sup>,  
Tomonobu Hasegawa<sup>1</sup>

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<sup>3</sup>Dept. of Molecular Endocrinology, National Center for Child Health and Development

**English Oral Session 2****October, 29(Fri.) 13:20 ~ 14:20 Room 4**

Chairs Noriyuki Namba (Division of Pediatrics and Perinatology, Faculty of Medicine, Tottori University)

Hiroshi Mochizuki (Division of Endocrinology and Metabolism, Saitama Children's Medical Center)

**EO2-1 Switch Data From the Open-Label Extension of the Pivotal Phase 3 Study of Once Weekly Somatrogon Compared to Daily Somatropin in Pediatric Patients with Growth Hormone Deficiency (pGHD)**

Michael Wajnrajch<sup>1</sup>, Bradley Miller<sup>2</sup>, Joel Steelman<sup>3</sup>, Lawrence Silverman<sup>4</sup>, Moshe Phillip<sup>5</sup>,  
Elpis Vlachopapadopoulou<sup>6</sup>, Renata Stawarska<sup>7</sup>, Ho-Seong Kim<sup>8</sup>, Oleg Malievskiy<sup>9</sup>,  
Cheol Woo Ko<sup>10</sup>, Srinivas Valluri<sup>1</sup>, Carrie Taylor<sup>1</sup>, Carl Roland<sup>11</sup>, John Choe<sup>12</sup>,  
Aleksandra Pastrak<sup>12</sup>, Cheri Deal<sup>13</sup>

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<sup>2</sup>University of Minnesota Masonic Children's Hospital, Minneapolis, MN, USA,

<sup>3</sup>Cook Children's Medical Center, Fort Worth, TX, USA,

<sup>4</sup>Goryeb Children's Hospital, Morristown, NJ, USA,

<sup>5</sup>Schneider Children's Medical Center, Petah Tiqwa, Israel,

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<sup>7</sup>Polish Mother's Memorial Hospital-Research Institute and Medical University of Lodz, Lodz, Poland,

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<sup>9</sup>Bashkir State Medical University, Ufa, Russian Federation,

<sup>10</sup>Kyungpook National University Children's Hospital, Daegu, Republic of Korea,

<sup>11</sup>Pfizer Inc, Sanford, NC, USA, <sup>12</sup>OPKO Health, Toronto, ON, Canada,

<sup>13</sup>Research Center, Sainte-Justine Mother-Child University Teaching Hospital and University of Montreal, Montreal, QC, Canada

## **EO2-2 Safety and effectiveness of pediatric growth hormone therapy: results from the full cohort in KIGS**

Mohamad Maghnie<sup>1</sup>, Michael B Ranke<sup>2</sup>, Mitchell E Geffner<sup>3</sup>, Elpis Vlachopapadopoulou<sup>4</sup>, Helmuth G Dörr<sup>5</sup>, Kerstin Albertsson Wikland<sup>6</sup>, Lourdes Ibáñez<sup>7</sup>, Martin Carlsson<sup>8</sup>, Wayne Cutfield<sup>9</sup>, Raoul Rooman<sup>10</sup>, Roy Gomez<sup>11</sup>, Michael P Wajnrajch<sup>8</sup>, Agnès Linglart<sup>12</sup>, Renata Stawerska<sup>13</sup>, Michel Polak<sup>14</sup>, Adda Grimberg<sup>15</sup>

<sup>1</sup>Dept of Pediatrics, Univ of Genova, Genova, Italy,

<sup>2</sup>Dept of Pediatric Endocrinology, University Children's Hospital, Tübingen, Germany,

<sup>3</sup>The Saban Research Institute, Children's Hospital Los Angeles, Los Angeles, CA, USA,

<sup>4</sup>Dept of Endocrinology, Growth and Development, Aglaia Kyriakou Children's Hospital, Athens, Greece,

<sup>5</sup>Division of Pediatric Endocrinology, Dept of Pediatrics and Adolescent Medicine, Friedrich-Alexander Univ of Erlangen-Nürnberg, Erlangen, Germany,

<sup>6</sup>Dept of Physiology/Endocrinology, Institute of Neuroscience and Physiology, Sahlgrenska Academy, Univ of Gothenburg, Gothenburg, Sweden,

<sup>7</sup>Endocrinology, Pediatric Research Institute Sant Joan de Déu, Barcelona, Spain,

<sup>8</sup>Endocrine Care, Pfizer, New York, NY, USA,

<sup>9</sup>Liggins Institute, Univ of Auckland, Auckland, New Zealand, <sup>10</sup>PendoCon, Putte, Belgium,

<sup>11</sup>European Medical Affairs, Pfizer, Brussels, Belgium,

<sup>12</sup>Dept of Pediatric Endocrinology and Diabetology for Children, APHP, Bicêtre Paris Saclay, Le Kremlin Bicêtre, France,

<sup>13</sup>Dept of Endocrinology and Metabolic Diseases, Polish Mother's Memorial Hospital-Research Institute, Lodz, Poland,

<sup>14</sup>Université de Paris; Hopital Necker Enfants Malades, Paris, France,

<sup>15</sup>The Children's Hospital of Philadelphia, Philadelphia, PA, USA

## **EO2-3 Global Pivotal Phase 3 Trial Comparing Once Weekly Somatrogen to Once Daily Genotropin in Children with Growth Hormone Deficiency (pGHD)**

Cheri Deal<sup>1</sup>, Aleksandra Pastrak<sup>2</sup>, Lawrence Silverman<sup>3</sup>, Srinivas Valluri<sup>4</sup>, Michael Wajnrajch<sup>4</sup>, Jose Cara<sup>4</sup>

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<sup>4</sup>Pfizer Inc, New York, NY, USA

## **EO2-4 Long-term safety of growth hormone in adults and adolescents with growth hormone deficiency: an overview of the full-cohort in KIMS**

Gudmundur Johannsson<sup>1</sup>, Philippe Touraine<sup>2</sup>, Ulla Feldt-Rasmussen<sup>3</sup>, Antonio Pico<sup>4,5,6</sup>, Greisa Vila<sup>7</sup>, Martin Carlsson<sup>8</sup>, André P van Beek<sup>9</sup>, Michael P Wajnrajch<sup>8</sup>, Roy Gomez<sup>10</sup>, Kevin CJ Yuen<sup>11</sup>

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<sup>2</sup>Sorbonne Université, Assistance Publique Hopitaux de Paris, Paris, France,

<sup>3</sup>Rigshospitalet, Copenhagen University Hospital, Copenhagen, Denmark,

<sup>4</sup>Biomedical Research Networking Center in Rare Diseases (CIBERER), Institute of Health Carlos III (ISCIII), Madrid, Spain,

<sup>5</sup>Hospital General Universitario de Alicante-Institute for Health and Biomedical Research (ISABIAL), Alicante, Spain,

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<sup>11</sup>Barrow Pituitary Center and Neuroendocrinology Clinic, Barrow Neurological Institute, University of Arizona College of Medicine, Creighton School of Medicine, Phoenix, AZ, USA

**EO2-5 Perception of Treatment Burden with Once Weekly Somatrogon vs Once Daily Genotropin in Pediatric Patients with Growth Hormone Deficiency (GHD): Results from a Randomized Phase 3 Study**

Aristides Maniatis<sup>1</sup>, Mauri Carakushansky<sup>2</sup>, Sonya Galcheva<sup>3</sup>, Gnanagurudasan Prakasam<sup>4</sup>, Larry Fox<sup>5</sup>, Adriana Dankovcikova<sup>6</sup>, Jane Loftus<sup>7</sup>, Andrew Palladino<sup>8</sup>, Maria Resa<sup>9</sup>, Carrie Taylor<sup>9</sup>, Mehul Dattani<sup>10</sup>, Jan Lebl<sup>11</sup>

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<sup>4</sup>Sutter Medical Center and Center of Excellence in Diabetes and Endocrinology, Sacramento, CA, USA,

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<sup>6</sup>Children's Faculty Hospital Kosice, Kosice, Slovakia, <sup>7</sup>Pfizer Ltd, Tadworth, UK,

<sup>8</sup>Pfizer Inc, Collegetown, PA, USA, <sup>9</sup>Pfizer Inc, New York, NY, USA,

<sup>10</sup>UCL Great Ormond Street Institute of Child Health, London, UK,

<sup>11</sup>Charles University and University Hospital Motol, Prague, Czech Republic

**EO2-6 Once-weekly somapacitan versus daily growth hormone in growth hormone deficiency: 3 year efficacy and safety results from REAL 3, a randomised controlled phase 2 trial**

Lars Savendahl<sup>1</sup>, Tadej Battelino<sup>2</sup>, Michael Hojby Rasmussen<sup>3</sup>, Paul Saenger<sup>4</sup>, Reiko Horikawa<sup>5</sup>

<sup>1</sup>Karolinska Inst., Karolinska Univ. Hosp.,

<sup>2</sup>Fac. of Med., UMC-Univ. Children's Hosp., Univ. of Ljubljana,

<sup>3</sup>Global Development, Novo Nordisk A/S, <sup>4</sup>NYU Langone Health, Mineola,

<sup>5</sup>Division of Endocrinology and Metabolism, National Center for Child Health and Development

English Oral Session 3

**October, 29(Fri.) 14:30 ~ 15:20 Room 4**

Chairs Tsuyoshi Isojima (Department of Pediatrics, Teikyo University School of Medicine)  
Shigeru Suzuki (Department of Pediatrics, Asahikawa Medical University)

**EO3-1 Efficacy, effectiveness, and tolerability of nasal glucagon in treating hypoglycemia in children and adolescents with type 1 diabetes**

Nan Zhang, Christopher Child, Qianqian Wang, Vivian Thieu  
Eli Lilly and Company

**EO3-2 A Japanese boy suspected of Simpson-Golabi-Behmel syndrome**

Masayo Yamazaki, Makiko Oguma, Toshihiro Tajima, Takanori Yamagata  
Dept. of Pediatr., Jichi Med. Univ.

**EO3-3 A mutation of the  $\beta$ -domain in *POU1F1* causes pituitary deficiency due to dominant PIT-1  $\beta$  expression**

Shigeru Suzuki<sup>1</sup>, Kumihiko Matsuo<sup>1,2</sup>, Yoshiya Ito<sup>1,3</sup>, Atsushi Kobayashi<sup>1,4</sup>, Osamu Ueda<sup>1,5</sup>, Tokuo Mukai<sup>1,6</sup>, Koichi Yano<sup>1,7</sup>, Kenji Fujieda<sup>1,8</sup>, Akimasa Okuno<sup>1,9</sup>, Yusuke Tanahashi<sup>1,10</sup>, Hiroshi Azuma<sup>1,9</sup>

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<sup>7</sup>City of Sapporo Minami Health and Welfare Center,

<sup>8</sup>(deceased) Former Prof., Dept. of Pediatr., Asahikawa Med. Univ.,

<sup>9</sup>Emeritus Prof., Asahikawa Med. Univ., <sup>10</sup>Dept. of Pediatr., Wakkanai City Hosp.

**EO3-4 A novel *NARS2* mutation detected in siblings of neonatal diabetes mellitus**

Hideaki Yagasaki, Hiromune Narusawa, Fumikazu Sano, Koichi Makino, Yumiko Mitsui,  
Kazumasa Sato, Koji Kobayashi, Tomoaki Sano, Masanori Ota, Takeshi Inukai

Dept. of Pediatr., Yamanashi Univ. Sch. of Med.

**EO3-5 Assisted reproductive technology represents a possible risk factor for development of epimutation-mediated imprinting disorders for mothers aged over 30 years**

Kaori Hara<sup>1</sup>, Keiko Matsubara<sup>1</sup>, Masashi Mikami<sup>2</sup>, Takahiro Arima<sup>3</sup>, Tsutomu Ogata<sup>4</sup>,  
Maki Fukami<sup>1</sup>, Masayo Kagami<sup>1</sup>

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