

Invited Lecture

November, 2(Wed.) 17:05 ~ 17:45 Room 1 (1F G2)

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

Molecular causes of primary adrenal insufficiency

John C. Achermann

UCL GOS Institute of Child Health, University College London, London, United Kingdom

KSPE-JSPE Plenary Lecture

November, 2(Wed.) 10:00 ~ 10:40 Room 1 (1F G2)

Chair Reiko Horikawa (Division of Endocrinology and Metabolism, National Center for Child Health and Development)

2022 Clinical Practice Guidelines for Central Precocious Puberty in Korea

Ji Eun Lee

Department of Pediatrics, division of Pediatric Endocrinology & metabolism,
Inha University Hospital, Inha university medical school

Luncheon Seminar 2

November, 1(Tue.) 12:00 ~ 12:50 Room 2 (3F G301+302)

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

IGFs and Growth Hormone Insensitivity: A Personal Voyage

Ron Rosenfeld

Professor and Chair of Pediatrics (emeritus), Oregon Health & Science University;
Professor of Pediatrics (emeritus), Stanford University

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English Oral Session 1 diabetes and others

November, 2(Wed.) 8:40 ~ 9:50 Room 4 (3F G304)



Chairs Tatsuhiko Urakami (Urakami Pediatric Endocrinology and Diabetes Clinic)
Misa Honda (Department of Pediatrics, Keio University School of Medicine)

EO1-1 Two sibling with LRBA deficiency presenting fulminant Type1 diabetes mellitus in infancy

Dai Suzuki¹, Eriko Totsune¹, Tomohiro Nakano¹, Akinobu Miura^{1,2}, Hirohito Shima¹,
Sayaka Kawashima¹, Ikumi Umeki^{1,3}, Youji Sasahara¹, Junko Kanno¹

¹Dept. of Pediatr., Tohoku Univ. Sch. of Med., ²Dept. of Pediatr., Sendai City Hosp.,

³Dept. of Pediatr., Iwate Pref. Central Hosp.

EO1-2 A case with a heterozygous protein-truncating *RFX6* variant diagnosed in the early stage of diabetes

Kazuhisa Akiba^{1,2,3}, Hiroaki Zukeran^{1,3}, Yukihiro Hasegawa^{1,4}

¹Div. of Endocrinol. Metab., TMCMC., ²Dept. of Mol. Endocrinol., NCCHD.,

³Dept. of Pediatr., Keio Univ. Sch. of Med., ⁴Dept. of Pediatr., Tama-Hokubu Med. Cent.

EO1-3 Efficacy and Safety of Dapagliflozin in Young Adults with Type 1 Diabetes Mellitus

Misa Honda^{1,2}, Noboru Uchida³, Tomohiro Ishii¹, Tomonobu Hasegawa¹, Goro Sasaki²

¹Dept. of Pediatr., Keio Univ. Sch. of Med., ²Tokyo Dental College Ishikawa General Hosp.,

³Saiseikai Utsunomiya Hosp.

- EO1-4 Experiences of Caregivers of Children and Young Adults with Type 1 Diabetes Related to Severe Hypoglycemia and Being Prepared with Nasal Glucagon - A Qualitative Study**
 Allyson Hughes¹, Emily Hankosky², Jiat Ling Poon², Katherine Chapman¹, Mark R Vanderwel³, Jeoffrey Bispham¹, Wendy Wolf¹, Beverly Falcon², Julie A Settles², Christopher J Child², Makoto Imori (non-author presenter)⁴
¹T1D Exchange, ²Eli Lilly and Company, ³Pediatric Endocrinology and Diabetes Specialists, ⁴Eli Lilly Japan K.K.
- EO1-5 A heterozygous familial hypercholesterolemia infant with a high polygenetic risk score showed severe high LDL-cho**
 Hirohito Shima¹, Kotaro Tayama¹, Tomohiro Nakagawa¹, Sayaka Kawashima¹, Dai Suzuki¹, Masaki Matsubara^{2,3}, Junko Kanno¹
¹Dept. of Pediatr., Tohoku Univ. Hosp.,
²Dept. of Diabetes and Lipid Metab., Natl. Cereb. and Cardiovasc. Ctr. Res. Inst.,
³Dept. of Gen. Med., Nara Med. Univ.
- EO1-6 Peripheral glucocorticoid metabolism may predict patient severity in PICU**
 Shuji Sai¹, Soichiro Wada¹, Shigetoshi Ogiwara¹, Takuya Tamura¹, Karen Chapman²
¹Dept. of Pediatr., Teine-Keijinkai Hosp., ²QMRI, Univ. of Edinburgh
- EO1-7 Achievement of Developmental Milestones Recorded in Real Time: A Mobile App-Based Study**
 Keiko Matsubara¹, Satoshi Narumi¹, Tomoyuki Hattori²
¹Dept. of Molecular Endocrinology, National Research Institute for Child Health and Development, ²First-Ascent Inc.

English Oral Session 2 growth, puberty and others

November, 2(Wed.) 14:30 ~ 15:30 Room 4 (3F G304)

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- Chairs Masayo Kagami (Department of Molecular Endocrinology, National Research Institute for Child Health and Development)
 Asako Tajima (Division of Endocrinology and Metabolism, Saitama Children's Medical Center)
- EO2-1 Zinc deficiency may affect to phenotypes of Trichorhinophalangeal syndrome type I harboring a novel *TRPS1* Variant**
 Hideaki Yagasaki¹, Koji Kobayashi¹, Hiromune Narusawa¹, Daisuke Watanabe¹, Mie Mochizuki¹, Kazumasa Sato¹, Tomoaki Sano¹, Hiroshi Mitsui², Masanori Ota¹, Takeshi Inukai¹
¹Dept. of Pediatr., Yamanashi Univ. Sch. of Med.,
²Dept. of Dermat., Yamanashi Univ. Sch. of Med.
- EO2-2 Outcomes in growth hormone-treated Noonan syndrome children: impact of PTPN11 mutation status**
 Alexander AL Jorge¹, Alberto Pietropoli², Nicky Kelepouris³, Reiko Horikawa⁴
¹Unidade de Endocrinologia-Genetica, LIM/25, Disciplina de Endocrinologia da Faculdade de Medicina da Universidade de Sao Paulo,
²Novo Nordisk Health Care AG, ³Novo Nordisk Inc.,
⁴Div. Endocrinol. Metab., Nat. Cent. for Child Health and Development

EO2-3 Molecular and clinical studies in 76 patients with central precocious puberty

Hiromune Narusawa^{1,2}, Keisuke Nagasaki³, Hideaki Yagasaki², Yasuhiro Naiki⁴, Shinichiro Sano⁵, Tomohiro Saito⁶, Shun Soneda⁷, Shintaro Terashita⁸, Saori Kinjo⁹, Mitsukazu Mamada¹⁰, Sumito Dateki¹¹, Satoshi Narumi¹, Reiko Horikawa⁴, Tsutomu Ogata¹², Maki Fukami¹, Masayo Kagami¹

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³Dept. Pediatr., Niigata Univ. Sch. Med. and Dent. Sciences,

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⁵Dept. Pediatr. Endo., Shizuoka Children's Hosp.,

⁶Dept. Pediatr., Yamanashi prefectural central Hosp., ⁷Tanaka Growth Clinic,

⁸Dept. Pediatr., Faculty of Medicine Univ. Toyama., ⁹Dept. Pediatr., Okinawa Chubu Hosp.,

¹⁰Dept. Pediatr., Tango Central Hosp., ¹¹Dept. Pediatr., Nagasaki Univ.,

¹²Dept. Pediatr., Hamamatsu Medical Center.

EO2-4 Array comparative genomic hybridization is useful for diagnosis of Turner syndrome with unbalanced translocation

Akie Nakamura¹, Shuntaro Morikawa¹, Takeshi Yamaguchi¹, Nozomi Hishimura¹, Kanako Nakayama¹, Naoya Kaneko¹, Katsura Ishizu²

¹Dept. of Pediatr., Hokkaido Univ. Sch. of Med., ²Kids Forest clinic

EO2-5 A case of Beckwith-Wiedemann syndrome with ACTH-independent Cushing's syndrome

Hiroko Yagi¹, Tomohiko Sato¹, Ken Higashimoto², Hidenobu Soejima², Kiminori Terui¹

¹Dept. of Pediatr., Hirosaki Univ. Sch. of Med., ²Dept. of Biomol. Sci., Saga Univ. Sch. of Med.

EO2-6 The high incidence of vitamin D deficiency in Hokkaido, especially Wakkani-Soya district, a high-latitude region of Japan

Yusuke Tanahashi¹, Shinsuke Fukui¹, Takamasa Miyoshi¹, Hideki Tamaru¹, Yuki Sasaki¹, Tomoka Tsubota¹, Takahide Kokumai², Shigeru Suzuki²

¹Dept. of Pediatr, Wakkani City Hosp., ²Dept. of Pediatr., Asahikawa Univ. Sch. of Med.

Web Poster Session 1 English Poster

November, 1(Tue.) 17:05 ~ 18:10 Zoom Breakout Room (web Poster Session Room 1) ㊤㊥

Chairs Kenichi Kashimada (Department of Developmental Biology and Pediatrics, Tokyo Medical and Dental University)

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

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

WP1-1-1 A case short stature case with a novel pathological variant of BMP2

Yogi Analia, Ryosei Iemura, Hisae Nakatani, Kei Takasawa, Kenichi Kashimada
Dept. of Pediatr. and Developmental Biology, Tokyo Medical and Dental Univ.

WP1-1-2 MODY With GCK Gene Heterozygous Missense Variant

Mazidah Noordin^{1,2}, Atsushi Hattori³, Maki Fukami³, Reiko Horikawa¹

¹Div. of Endocrinology, National Center for Child Health & Development (NCCHD), Tokyo, Japan,

²Dept. of Pediatr., Faculty of Medicine, Universiti Teknologi MARA (UiTM), Sungai Buloh Campus, Selangor, Malaysia

³Div. of Molecular Endocrinology, National Center for Child Health and Development (NCCHD), Tokyo, Japan

- WP1-1-3 Seasonal variation in vitamin D status of infants born in Saitama City emerges at 2 months of age**
Keigo Takahashi^{1,2}, Takeshi Arimitsu³, Kaori Hara-Isono³, Kazushige Ikeda²
¹Dept. of Pediatr., Washington Univ. in St. Louis,
²Div. of Neonat., Dept. of Pediatr., Saitama City Hosp.,
³Dept. of Pediatr., Keio Univ. Sch. of Med.
- WP1-1-4 A real-world study of persistence with daily growth hormone therapy among children with growth hormone deficiency in Japan**
Jane Loftus¹, Jenifer Wogen², Darrin Benjumea², Priti Jhingran², Yong Chen³, Jose Alvir⁴, Michael Wajnrajch^{4,5}
¹Pfizer, Walton Oaks, UK, ²Genesis Research, Hoboken, NJ, USA,
³Pfizer Inc, Collegeville, PA, USA, ⁴Pfizer Inc, New York, NY, USA,
⁵New York Univ. Grossman Sch. of Med., New York, NY, USA
- WP1-1-5 Growth outcomes from phase 2 and phase 3 studies of once-weekly somatrogen vs once-daily genotropin in pediatric patients with growth hormone deficiency: comparisons with literature and a growth study database**
Zvi Zadik¹, Jose Cara², Martin Carlsson², Michael Wajnrajch^{2,3}, Ronnie Wang⁴, Ron Rosenfeld⁵
¹Pediatric Endocrinology, Kaplan Medical Center, Rehovot, Israel,
²Pfizer Inc, New York, NY, USA,
³New York Univ. Grossman Sch. of Med., New York, NY, USA,
⁴Pfizer Inc, Groton, CT, USA, ⁵Oregon Health and Science Univ., Portland, OR, USA
- WP1-1-6 Efficacy and safety of once-weekly somatrogen in pediatric subjects with growth hormone deficiency: lack of impact of anti-drug antibodies**
Cheri Deal¹, Lawrence Silverman², Joan Korth-Bradley³, Carl Roland⁴, Carrie Taylor⁵, Jose Cara⁵, Michael Wajnrajch^{5,6}
¹Centre de recherche CHU Ste-Justine, Université de Montréal, Montréal, Canada,
²Goryeb Children's Hosp., Atlantic Health System, Morristown, NJ, USA,
³Pfizer Inc, Collegeville, PA, USA, ⁴Pfizer Inc, Sanford, NC, USA,
⁵Pfizer Inc, New York, NY, USA, ⁶New York Univ. Grossman Sch. of Med., New York, NY, USA
- WP1-1-7 Once-Weekly Somapacitan in Growth Hormone Deficiency: 4-Year Efficacy and Safety Results from REAL 3, a Randomised Controlled Phase 2 Trial**
Lars Savendahl¹, Tadej Battelino², Michael Hojby Rasmussen³, Paul Saenger⁴, Reiko Horikawa⁵
¹Dept. of Women's and Children's Health, Karolinska Inst. and Pediatr. Endocrinology Unit, Karolinska Univ. Hosp.,
²Fac. of Med., UMC-Univ. Children's Hosp., Univ. of Ljubljana,
³Clinical Drug Development, Novo Nordisk A/S, ⁴NYU Langone Health, Mineola,
⁵Div. Endocrinol. Metab., Nat. Cent. for Child Health and Development

WP1-1-8 Once-weekly Somapacitan is Effective and Well Tolerated in Children with GH Deficiency: a Randomized Phase 3 Trial

Bradley S. Miller¹, Joanne C. Blair², Michael Højby Rasmussen³, Volker Böttcher⁴, Rikke Beck Bang⁵, Rasmus Juul Kildemoes³, Aristides Maniatis⁶, Jun Mori⁷, Michel Polak^{8,9,10,11}, Stefano Stagi¹², Reiko Horikawa¹³

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³Novo Nordisk A/S, Søborg, Denmark, ⁴Endocrinology Clinic, Frankfurt am Main, Germany,

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⁶Rocky Mountain Pediatric Endocrinology, Centennial, CO, USA,

⁷Dept. of Pediatr., Graduate Sch. of Medical Science, Kyoto Prefectural Univ. of Med., Kyoto, Japan,

⁸Service d'Endocrinologie, Gynécologie et Diabétologie Pédiatriques, Hôpital Universitaire Necker Enfants Malades Paris, Assistance Publique-Hôpitaux de Paris, Paris, France,

⁹Université de Paris Cité, Paris, France, ¹⁰INSERM U1016, Institut Cochin, Paris, France,

¹¹Institut Imagine, Paris, France,

¹²Dept. of Health Sciences, Univ. of Florence, Anna Meyer Children's Univ. Hosp., Florence, Italy,

¹³Div of Endocrinology and Metabolism, National Center for Child Health and Development, Tokyo, Japan

WP1-1-9 Safety and efficacy of treatment with lonapegsomatropin in children with growth hormone deficiency at Week 130 in the enliGHten trial

Paul Hofman¹, Paul S. Thornton², Ulhas M. Nadgir³, Paul Saenger⁴, Elena D. Chertok⁵, Elena M. Aghajanova⁶, Wenjie Song⁷, Meng Mao⁷, Steven D. Chessler⁷, Allison S. Komirenko⁷, Michael Beckert⁸, Aimee D. Shu⁷, Aristides K. Maniatis⁹

¹Liggins Institute, Univ. of Auckland, ²Cook Children's Medical Center, Fort Worth, TX, USA,

³Center of Excellence in Diabetes and Endocrinology, Sacramento, CA,

⁴NYU Langone Health, New York, USA, ⁵Voronezh State Medical Univ., Voronezh, Russia,

⁶Yerevan State Medical Univ., Yerevan, Armenia, ⁷Ascendis Pharma, Palo Alto, CA, USA,

⁸Ascendis Pharma A/S, Hellerup, Denmark,

⁹Rocky Mountain Pediatric Endocrinology, Centennial, CO, USA

WP1-1-10 Once-Weekly Somapacitan Versus Daily Growth Hormone in Children Born Small for Gestational Age: Results From a Randomized Phase 2 Trial

Anders Juul^{1,2}, Philippe Backeljauw³, Michael Højby⁴, Masanobu Kawai⁵, Rasmus Juul Kildemoes⁶, Agnès Linglart⁷, Nehama Zuckerman-Levin⁸, Reiko Horikawa⁹

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³Div. of Endocrinology, Dept. of Pediatr., Cincinnati Children's Hosp. Medical Center, Univ. of Cincinnati College of Medicine, Cincinnati, Ohio, USA,

⁴Clinical Drug Development, Novo Nordisk A/S, Søborg, Denmark,

⁵Dept. of Gastroenterology, Nutrition and Endocrinology, Research Institute, Osaka Women's and Children's Hosp., Izumi, Japan,

⁶Data Science, Novo Nordisk A/S, Søborg, Denmark,

⁷AP-HP, Service d'Endocrinologie et Diabète de l'Enfant, Hôpital Bicêtre Paris Saclay, Université Paris Saclay, Le Kremlin-Bicêtre, France,

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