16th International Collaborative Forum
of Human Gene Therapy for Genetic Disease

第16回 国際協力遺伝病遺伝子治療フォーラム

Generate Innovation with Young Power

若い力で革新を

Date 開催日
January 23, 2026 (Fri.)
2026年1月23日（金）

Venue 会　場
The Jikei University School of Medicine, Building 1 Hall
東京慈恵会医科大学　1号館講堂

President 当番幹事
Prof. Hitoshi Osaka (Dept. of Pediatrics, Jichi Medical University)
小坂　仁（自治医科大学小児科学講座 教授）

Abstract Program Book

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Welcome Message

It is our great pleasure to announce that the **16th International Cooperative Forum on Gene Therapy for Genetic Diseases** will be held on Friday, January 23, 2026, at the Lecture Hall of the First Building, Jikei University School of Medicine in Tokyo.

In recent years, remarkable progress has been made in gene and cell therapies for genetic diseases, with the approval of new treatments accelerating worldwide. Through our own experience with AADC deficiency therapy, we have witnessed firsthand the great potential of gene therapy. Furthermore, gene therapy products for adrenoleukodystrophy (ALD) and metachromatic leukodystrophy (MLD) have been approved in Europe and the United States, and Duchenne muscular dystrophy (DMD) has also received approval in Japan. In addition, the development of **N-of-1 gene-editing therapies**, individually designed for single patients, has attracted international attention, further demonstrating the expanding possibilities of gene therapy.

At the same time, clinical application and approval of gene therapies in Japan continue to lag behind those in Western countries. In particular, the issues of drug lag and drug loss in the field of rare genetic diseases remain serious, and the establishment of infrastructure and acceleration of clinical implementation are urgent challenges. On the other hand, Japan has been internationally recognized for its pioneering achievements in regenerative medicine, especially with iPS cell research. It is therefore essential to build upon these strengths and further promote research that integrates diverse modalities, including gene therapy.

The theme of this Forum is **“Generate Innovation with Young Power.”** Our goal is to foster momentum for innovative therapeutic development led by the next generation of young researchers. We sincerely hope that this Forum will not only serve as an opportunity to share the latest knowledge on gene and cell therapies for genetic diseases, but also provide a platform where young researchers actively participate in research and development, respond to the expectations of patients and their families, and contribute to advancing gene and cell therapies into a future industry in Japan.

The 16th International Cooperative Forum on Gene Therapy for Genetic Diseases
Organizing Chair

Hitoshi Osaka

プログラム ／ Program

8:20 ～ 8:30

Opening Remarks ／ 開会あいさつ

Hitoshi Osaka (*President of the 16th International Collaborative Forum of Human Gene Therapy for Genetic Disease/ Department of Pediatrics, Jichi Medical University*)
小坂　　仁 (第16回当番幹事　国際協力遺伝病遺伝子治療フォーラム / 自治医科大学小児科学講座)

Masafumi Onodera (*President,International Collaborative Forum of Human Gene Therapy for Genetic Disease/ Graduate School of Engineering, Osaka University*)
小野寺雅史 (代表幹事　国際協力遺伝病遺伝子治療フォーラム / 大阪大学大学院工学研究科)

8:30 ～ 9:20

Morning Seminar ／ モーニングセミナー

Gene Therapy for Duchenne Muscular Dystrophy ／ デュシェンヌ型筋ジストロフィーの遺伝子治療

Chair ／ 座長

Kazuhiro Muramatsu (*Department of Pediatrics, Jichi Medical University*)
村松　一洋 (自治医科大学小児科学講座)

Speaker ／ 演者

Duchenne Muscular Dystrophy: Evolving Landscape of Gene Therapy and Clinical Insights 8
Yuko Shimizu-Motohashi (*Department of Child Neurology, National Center of Neurology and Psychiatry*)
本橋　裕子 (国立精神・神経医療研究センター脳神経小児科)

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Chugai Pharmaceutical Co., LTD ／ 中外製薬株式会社

9:30 ～ 11:00

Session 1

Gene Therapy for Metabolic Disease ／ 代謝性疾患の遺伝子治療

Topics ／対象疾患 : MPS IIIA, GSD Ia, Pompe Disease

Chairs ／ 座長

Toya Ohashi (*Health Science and Therapeutics ,The Jikei University School of Nursing*)
大橋　十也 (東京慈恵会医科大学医学部看護学科健康科学疾病治療学)

Hiroshi Kobayashi (*Division of Gene Therapy, Research Center for Medical Sciences, The Jikei University School of Medicine*)
小林　博司 (東京慈恵会医科大学総合医科学研究センター遺伝子治療研究部)

Speakers ／ 演者

Treatment with UX111 Reduced Cerebrospinal Fluid (CSF) Heparan Sulfate (HS) Exposure and Stabilized or Improved Functioning across Dose, Age, and Stage of MPS IIIA 10
Heather A. Lau (*Global Clinical Development, Ultragenyx Pharmaceutical*)

Results from a pivotal phase 3 double-blind placebo-controlled trial of DTX401 for the treatment of individuals with glycogen storage disease type Ia (GSDIa) 12
Richard Collis (*Global Clinical Development, Ultragenyx Pharmaceutical Inc.*)

AT845 gene replacement therapy for late-onset Pompe disease: an update on safety and preliminary efficacy data from FORTIS, a phase 1/2 open-label clinical study..... 14

Chieri Hayashi (*Astellas Pharma Global Development, USA*)

林　千江里 (アステラス製薬株式会社)

11:10 ～ 12:00

Session 2

Gene Therapy for Sensory Organs ／ 感覚器疾患の遺伝子治療

Chairs ／ 座長

Koji Nishiguchi (*Ophthalmology, Graduate School of Medicine, Nagoya University*)

西口　康二 (名古屋大学医学系研究科眼科)

Masafumi Onodera (*Graduate School of Engineering, Osaka University*)

小野寺雅史 (大阪大学大学院工学研究科)

Speakers ／ 演者

1) Gene-Specific and Gene-Agnostic Therapies for Inherited Retinal Diseases..... 16

Yusuke Murakami (*Department of Ophthalmology, Kyushu University*)

村上　祐介 (九州大学医学部 眼科学教室)

2) Preliminary Safety and Efficacy of AK-OTOF Gene Therapy for Otoferlin Gene (OTOF) -mediated Hearing Loss 18

Katie Wachtel (*Regulatory Affairs, Lilly Gene Therapy, Eli Lilly and Company*)

12:10 ～ 13:00

Luncheon Seminar ／ ランチョンセミナー

遺伝子治療用製品のウイルス安全性評価試験

Chairs ／ 座長

Hideto Chono (*CDM Business Development Division, Takara Bio Inc.*)

蝶野　英人 (タカラバイオ株式会社 CDM 推進本部)

Speakers ／ 演者

Testing service for evaluation of viral safety at Takara Bio 20

Yuko Kato-Mori (*ViSpot Division, Takara Bio Inc.*)

森　ゆうこ (タカラバイオ株式会社 ViSpot 事業部)

Sponsored by ／ 共催

Takara Bio Inc. ／ タカラバイオ株式会社

13:15 ～ 14:00

Session 3

Various Modalities for Genetic Diseases; ex vivo gene therapy ／ Nucleic Acid Therapy ／ 様々なモダリティで挑む遺伝病治療（遺伝子治療・核酸医薬）

Chairs ／ 座長

Torayuki Okuyama (*Genomic Medicine, Saitama Medical University*)
奥山　虎之 (埼玉医科大学ゲノム医療科)

Toru Uchiyama (*Department of Human Genetics ,National Center for Child Health and Development*)
内山　　徹 (国立成育医療研究センター成育遺伝研究部)

Speakers ／ 演者

1) Clinical Development of Atidarsagene Autotemcel, "arsa-cel," Autologous Hematopoietic Stem Cell Gene Therapy (HSC-GT) for Treatment of Early-Onset Metachromatic Leukodystrophy (MLD) , and Implications for MLD Newborn Screening (NBS) **22**
Kent Christopherson (*Global Medial Affairs, Orchard Therapeutics*)

2) Challenges toward personalized nucleic acid drug development for rare genetic disorders **24**
Hiroya Kuwahara (*Department of Neurology and Neurological Science / NucleoTIDE and PepTIDE Drug Discovery Center (TIDE) , Institute of Science Tokyo*)
桑原　宏哉 (東京科学大学脳神経病態学分野／核酸・ペプチド創薬治療研究センター)

14:10 ～ 15:00

共催セミナー

Various Modalities: Enzyme Replacement ／ 遺伝病の克服に向けて

Chairs ／ 座長

Karin Kojima (*Department of Pediatrics, Jichi Medical University*)
小島　華林 (自治医科大学 小児科学講座)

Motomichi Kosuga (*Division of Medical Genetics ,National Center for Child Health and Development*)
小須賀基通 (国立成育医療研究センター遺伝診療科)

Speakers ／ 演者

ERT, as a treatment approaches for genetic disorders **26**
Hiroshi Kobayashi (*Division of Gene Therapy, Research Center for Medical Sciences The Jikei University School of Medicine*)
小林　博司 (東京慈恵会医科大学総合医科学研究センター遺伝子治療研究部)

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15:10 ～ 16:00 **Session 4**

Empowering Young Leaders to Drive Genetic Medicine from Japan to the World ／ 遺伝病治療を日本から世界へ発信するために

Chairs ／ 座長

Hitoshi Osaka (*Department of Pediatrics ,Jichi Medical University*)
小坂　　仁 (自治医科大学小児科学講座)

Generate Innovation with Young Power : The Future of Gene Therapy Driven by the Next Generation. **28**

Speakers ／ 演者

Takafumi Nakamura (*Department of Genomic Medicine and Regenerative Therapeutics, Faculty of Medicine, Tottori University*)
中村　貴史 (鳥取大学医学部ゲノム再生医学講座)

Yoshiyuki Saito (*Faculty of Pharmaceutical Sciences, The University of Tokyo*)
齋藤　良行 (東京大学大学院 薬学系研究科)

Haruka Mizuno (*Meijo University, School of Pharmacy / student ambassadors from Innovation for NEW HOPE*)
水野　　遥 (名城大学／ Innovation for NEW HOPE 学生アンバサダー)

Ayaka Kikuchi (*Meiji Pharmaceutical University / student ambassadors from Innovation for NEW HOPE*)
菊池　彩華 (明治薬科大学／ Innovation for NEW HOPE 学生アンバサダー)

令和6年度 大学教育再生戦略推進費高度な臨床・研究能力を有する医師養成促進支援

16:10 ～ 17:00 **Sweets Seminar ／ スイーツセミナー**

Chairs ／ 座長

Takashi Okada (*Center for Gene and Cell Therapy, The Institute of Medical Science, The University of Tokyo*)
岡田　尚巳 (東京大学医科学研究所遺伝子・細胞治療センター)

Takanori Yamagata (*Pediatrics ,Tochigi Rehabilitation Center*)
山形　崇倫 (栃木県立リハビリテーションセンター小児科)

Speakers ／ 演者

Accelerating Gene Therapy Development with VectorBuilder **30**

Miho Matakatsu (*VectorBuilder Inc.*)
亦勝　実穂 (ベクタービルダー株式会社)

sponsored by ／ 共催

ベクタービルダー・ジャパン株式会社 ／ VectorBuilder Japan Inc.

17:10 ～ 18:00 **Session 5** Special Lecture ／ 特別講演

Chair ／ 座長

Yoshikatsu Eto (*Advanced Clinical Research Center ,Southern Tohoku Institute for Neuroscience*)

衛藤　義勝 (一般財団法人脳神経疾患研究所先進医療研究センター)

Speaker ／ 演者

ExpEditing AAV gene therapy **32**

Alberto Auricchio

(*President, European Society of Gene & Cell Therapy*（*ESGCT*))

18:00 ～ 18:10 **Closing remarks ／ 閉会あいさつ**

Hironori Nakagami (*Vice President, Japan Society of Gene and Cell Therapy/Department of Health and Medicine ,Osaka University Graduate School of Medicine*)

中神　啓徳 (一般社団法人日本遺伝子細胞治療学会　副会長 / 大阪大学大学院医学系研究科 健康発達医学寄附講座)

Motomichi Kosuga (*President of the 17th International Collaborative Forum of Human Gene Therapy for Genetic Disease/ Division of Medical Genetics, National Center for Child Health and Development*)

小須賀基通 (第 17 回当番幹事　国際協力遺伝病遺伝子治療フォーラム / 国立成育医療研究センター遺伝診療科)

Hitoshi Osaka (*President of the 16th International Collaborative Forum of Human Gene Therapy for Genetic Disease/ Department of Pediatrics, Jichi Medical University*)

小坂　　仁 (第 16 回当番幹事　国際協力遺伝病遺伝子治療フォーラム / 自治医科大学小児科学講座)

Abstracts & Curriculum Vitae

Yuko Shimizu-Motohashi

National Center of Neurology and Psychiatry, Department of Child Neurology, Chief Physician

CURRICULUM VITAE

Career History

2000 Graduated from Yokohama City University School of Medicine
2000 Pediatric Resident, National Center for Global Health and Medicine
2005 Pediatrician, Medical Center for Developmental Disabilities
2007 Resident in Department of Child Neurology, National Center of Neurology and Psychiatry (NCNP)
2010 Research Fellow, Department of Molecular Therapy, NCNP
2011 Research Fellow, Division of Genetics, Boston Children's Hospital
2012 Postdoctoral Associate, Department of Neurology, University of Minnesota Medical School
2014 Pediatric Neurologist, Department of Child Neurology, NCNP
2019- Chief Pediatric Neurologist, Department of Child Neurology, NCNP

Academic Awards Received

None

Publications

1) Gene therapy for Duchenne muscular dystrophy. Shimizu-Motohashi Y.
   Brain Dev. 2025 Oct;47(5):104424. doi: 10.1016/j.braindev.2025.104424.
2) Natural history of timed rise from floor in young individuals with Duchenne muscular dystrophy: A single-center retrospective study.
   Yoneno S, Takeshita E, Shimizu-Motohashi Y, Oba M, Hara T, Hirayama M, Komaki H.
   Brain Dev. 2025 Aug;47(4):104396. doi: 10.1016/j.braindev.2025.104396.
3) Phase 1/2 trial of brogidirsen: Dual-targeting antisense oligonucleotides for exon 44 skipping in Duchenne muscular dystrophy. Komaki H, Takeshita E, Kunitake K, Ishizuka T, Shimizu-Motohashi Y, Ishiyama A, Sasaki M, Yonee C, Maruyama S, Hida E, Aoki Y. Cell Rep Med. 2025 Jan 21;6(1):101901. doi: 10.1016/j.xcrm.2024.101901.

Duchenne Muscular Dystrophy: Evolving Landscape of Gene Therapy and Clinical Insights

Yuko Shimizu-Motohashi

National Center of Neurology and Psychiatry, Department of Child Neurology, Chief Physician

Duchenne muscular dystrophy (DMD) is a progressive X-linked neuromuscular disorder characterized by early-onset muscle weakness, loss of ambulation in late childhood, and subsequent cardiopulmonary decline. Advances in molecular therapeutics have expanded treatment options beyond traditional supportive care, ushering in a new era in which exon skipping and gene therapy have become clinically accessible.

Adeno-associated virus (AAV)–based microdystrophin gene replacement, typified by delandistrogene moxeparvovec (Elevidys), represents the most clinically advanced platform to date and was recently approved in Japan. Clinical trial data have identified a spectrum of immune-mediated adverse events, including acute liver injury, myocarditis, and microdystrophin epitope–specific T-cell–driven myositis, as well as rare cases of severe hepatic dysfunction. These findings underscore the importance of a safe operational framework for treatment administration, careful patient selection, and longitudinal follow-up guided by a in-depth understanding of the disease.

In Japan, clinical implementation is shaped by the regulatory requirements of the Cartagena Act, which mandates biosafety oversight, containment measures, and viral-shedding management. As such, practical deployment requires multidisciplinary coordination, institution-specific protocols, and extended inpatient monitoring following infusion.

Although expectations for this gene therapy are high, there remains a clear need for novel therapeutic strategies capable of delivering more robust and durable efficacy. Future directions include next-generation AAV capsids with enhanced muscle tropism; approaches to overcome vector size limitations, such as dual- or triple-AAV and non-viral platforms; and immune-modulating strategies that may expand eligibility for patients with pre-existing AAV antibodies or advanced disease.

Together, these clinical, mechanistic, and safety insights underscore the importance of a thorough understanding of the disease as we navigate the evolving DMD gene-therapy landscape and the responsibilities that accompany its implementation. Lastly, but most importantly, we must recognize that the full extent of safety and efficacy of currently available innovative therapies for DMD remains to be clarified. Continued efforts to generate high-quality evidence are essential to support future therapeutic progress.

Heather A. Lau, M.D., M.S.

Executive Director, Global Clinical Development, Ultragenyx Pharmaceutical

CURRICULUM VITAE

Education and Academic Carrier

1998:	BS, General Biology with honors, Cornell University, Ithaca, NY, USA
2001:	MS Biochemistry and Molecular Biology, New York Medical College, Valhalla, NY, USA
2005:	MD, School of Medicine, University of Rochester. Rocheter NY, USA
2013 – 2021:	Assistant Professor, Department of Neurology, NYU Grossman School of Medicine, New York, NY, USA
2021 – Present:	Adjunct Assistant Professor, Department of Internal Medicine, Yale University, USA

Publications in peer-review Journals (selected)

- **Lau HA**, Viskochil D, Tanpaiboon P, Lopez AG, Martins E, Taylor J, Malkus B, Zhang L, Jurecka A, Marsden D. Long-term efficacy and safety of vestronidase alfa enzyme replacement therapy in pediatric subjects < 5 years with mucopolysaccharidosis VII. Mol Genet Metab. 2022 May;136(1):28-37.
- Hughes DA, Deegan P, Giraldo P, Göker-Alpan Ö, **Lau H**, Lukina E, Revel-Vilk S, Scarpa M, Botha J, Gadir N, Zimran A; GOS Steering Committee. Switching between Enzyme Replacement Therapies and Substrate Reduction Therapies in Patients with Gaucher Disease: Data from the Gaucher
- Harmatz P, Prada CE, Burton BK, **Lau H**, Kessler CM, Cao L, Falaleeva M, Villegas AG, Zeitler J, Meyer K, Miller W, Wong Po Foo C, Vaidya S, Swenson W, Shiue LH, Rouy D, Muenzer J. First-in-human in vivo genome editing via AAV-zinc-finger nucleases for mucopolysaccharidosis I/II and hemophilia B. Mol Ther. 2022 Dec 7;30(12):3587-3600. doi: 10.1016/j.ymthe.2022.10.010. Epub 2022 Oct 25. PMID: 36299240
- **Lau, H**.; Harmatz, P.; Botha, J.; Audi, J.; and Link, B. Clinical characteristics and somatic burden of patients with mucopolysaccharidosis II with or without neurological involvement: An analysis from the Hunter Outcome Survey. Mol Genet Metabol Rep. 2023; 37:01005,
- Muenzer J, Ho C, **Lau H**, Dant M, Fuller M, Boulos N, Dickson P, Ellinwood NM, Jones SA, Zanelli E, O'Neill C. Community consensus for Heparan sulfate as a biomarker to support accelerated approval in Neuronopathic Mucopolysaccharidoses. Mol Genet Metab. 2024 Aug;142(4):108535.

Treatment with UX111 Reduced Cerebrospinal Fluid (CSF) Heparan Sulfate (HS) Exposure and Stabilized or Improved Functioning across Dose, Age, and Stage of MPS IIIA

Heather A. Lau, M.D., M.S.

Executive Director, Global Clinical Development, Ultragenyx Pharmaceutical

Background: MPS IIIA (Sanfilippo type A) is a progressive neurodegenerative disease characterized by developmental arrest, regression, and early death. UX111 (rebisufligene etisparvovec) is an AAV9 viral vector encoding human SGSH under investigation for MPS IIIA.

Methods: Data were analyzed across 2 open-label studies (NCT02716246, NCT0408873) and the ongoing long-term follow up (NCT04360265). Children received a single UX111 infusion IV at low ($0.5x10^{13}$ vg/kg, n=3), mid ($1x10^{13}$ vg/kg, n=3), or high dose ($3x10^{13}$ vg/kg, n=27). The mITT set (n=17) included children treated with high dose and either ≤2 years old or >2 years old with a BSID-III cognitive developmental quotient ≥60. CSF HS exposure was defined as time-normalized area under the curve of the percentage change from baseline. Data cutoff was 01Aug2024.

Results: 33 children received UX111, with a median (min-max) follow up of 43.96 (7.00-77.14) months. Median (95% CI) CSF HS exposure decreased by 57.86% (33.15, 61.44) relative to baseline for the low dose, 42.81% (39.74, 62.75) for the mid dose, 64.51% (56.29, 71.25, p<0.0001) for the high dose, and 65.96% (56.86, 74.16, p<0.0001) for the mITT set. In the mITT set, mean change in model-estimated BSID-III raw scores from 24-60 months of age was significantly improved for treated children vs Natural History for cognitive (22.7 points, p<0.0001), receptive communication (7.4 points, p=0.0212), and expressive communication (15.9 points, p=0.0011). Fine and gross motor skills showed separation from Natural History (7.3 points, p=0.050 and 2.0 points, p=0.360, respectively). Older children and those with more advanced disease at treatment (n=10) retained key communication, ambulation, and/or eating/self-feeding skills. Most UX111-related TEAEs were mild to moderate and resolved spontaneously.

Conclusions: UX111 showed a positive treatment effect and manageable safety in children with MPS IIIA across UX111 doses, age, and stage of disease at the time of treatment.

Richard Collis

Executive Medical Director, Global Clinical Development, Ultragenyx Pharmaceutical Inc.

CURRICULUM VITAE

- Collis R. Investigational DTX401 Phase 1/2 Trial Interviews to Explore Glycogen Storage Disease Type Ia Patient Experiences of Gene Therapy. Oral presentation at the *Scandinavian Association for Glycogen Storage Disease (SAGSD) 6th GSD Conference*; May 3–4, 2024; Ängelholm, Sweeden.
- Mitchell JJ, Riba-Wolman R, Rodriguez-Buritica DF, et al. Long-term Efficacy and Safety in Adults with Glycogen Storage Disease Type Ia (GSDIa) from a Phase 1/2 Clinical Trial and Long-term Follow-up Study of DTX401, an AAV8-mediated, Liver-directed Gene Therapy. Oral presentation at the *American Society of Gene & Cell Therapy (ASGCT)*; May 7–11, 2024, Baltimore, MD, USA.
- Mitchell JJ, Abdenur JE, de Boer F, et al. Efficacy and Safety Results from a Pivotal Phase 3 Trial of DTX401, an AAV8-mediated Liver-directed Gene Therapy, in Individuals with Glycogen Storage Disease Type Ia. Oral and poster presentation at the *Joint Congress of ESPE and ESE*; May 10-13, 2025; Copenhagen, Denmark.
- Collis R, Turner-Bowker DM, Egan S, et al. Qualitative Interviews to Characterize Disease and Treatment Burden at Baseline in Adult and Pediatric Patients Participating in a Pivotal Phase 3 Trial of DTX401 for the Treatment of Glycogen Storage Disease Type Ia. Poster presentation at the *Association for Glycogen Storage Disease Conference (AGSD)*; June 20–21, 2025; Denver, CO, USA.
- Blake A, Saavedra H, Mount M, et al. Nutritional Changes after an AAV8-mediated Liver-directed Gene Therapy in Participants with Glycogen Storage Disease Type Ia (GSDIa): Results from a Phase 3 Pivotal Trial. Presented as a poster at the *42nd Annual Meeting of the Southeastern Regional Genetics Group (SERGG)*; July 17-19, 2025, Asheville, NC, USA.

Results from a pivotal phase 3 double-blind placebo-controlled trial of DTX401 for the treatment of individuals with glycogen storage disease type Ia (GSDIa)

Richard Collis

Executive Medical Director, Global Clinical Development, Ultragenyx Pharmaceutical Inc.

Background: GSDIa is a rare, potentially life-threatening inherited carbohydrate metabolism disorder caused by biallelic pathogenic *G6PC* gene variants, resulting in deficiency of the glucose-6-phosphatase complex. DTX401 is an investigational adeno-associated virus serotype 8 vector containing the human *G6PC* gene.

Methods: DTX401-CL301 (GlucoGene Study; NCT05139316), is an ongoing, pivotal, phase 3, double-blind, randomized, placebo-controlled trial of DTX401 in patients 8 years and older with GSDIa. The primary endpoint was percent change from Baseline to Week 48 in daily cornstarch intake for the DTX401 group versus placebo group. Participants were randomly assigned (1:1) to receive blinded DTX401 or placebo. After the 48-week Primary Efficacy Analysis Period (PEAP), participants crossed over in a blinded manner, such that participants in the DTX401 group at Day 1 received placebo at Week 48, while participants in the placebo group at Day 1 received DTX401 at Week 48 (Crossover DTX401). Both the DTX401 and Crossover DTX401 groups were followed for an additional 48 weeks (Crossover Period). Here we present both the PEAP results and the Week 96 full dataset, as well as Week 24 results from an additional arm where 3 patients received open-label DTX401 in Japan.

Results: Following randomization, 21 participants received DTX401 and 25 received placebo. At Week 48, the mean (standard error [SE]) reduction in daily cornstarch intake from baseline was 10% (4.1) in the placebo group (n=23) versus 41% (4.6) in the DTX401 group (n=19); $p<0.0001$. At Week 96, the mean (SE) reduction in daily cornstarch intake from baseline was 61% (6.3) in the Crossover DTX401 group (n=18) and 61% (5.1) in the DTX401 group (n=15). This 61% reduction in cornstarch intake observed for both treatment groups at Week 96 generally exceeded the mean desired cornstarch reduction of 45% obtained from patient interviews at trial baseline, supporting the clinical meaningfulness of this change. Low levels of hypoglycemia were maintained in both groups despite reductions in cornstarch. Anticipated vector-induced liver enzyme elevations were managed with prophylactic corticosteroids. Elevations in triglycerides were observed in individual participants, requiring adjustments of cornstarch and dietary intake. Similar large reductions in daily cornstarch intake were observed at Week 24 in the Japan cohort (n=3), with a mean (SE) reduction of 95% (4.8) after open-label DTX401 treatment.

Conclusions: Treatment with DTX401 resulted in statistically significant and clinically meaningful reductions in cornstarch intake in the 48-week PEAP versus placebo. Greater reductions in cornstarch were observed in both groups in the Crossover Period at Week 96 and preliminary data from Japan. Experience with disease management post-gene therapy and confidence that all participants were treated with DTX401 may have contributed to greater cornstarch reductions in the Crossover Period versus those observed at the end of the 48-week PEAP. DTX401 was well tolerated and has an acceptable and manageable safety profile.

Chieri Hayashi

Astellas Pharma Global Development, Northbrook, IL, USA;

CURRICULUM VITAE

Chieri Hayashi earned her MD and PhD in Otolaryngology and Molecular Biology from Juntendo University School of Medicine in 2001 and 2007, respectively. From 2011 to 2014, Dr. Hayashi conducted research at the University of Pittsburgh as a visiting scholar in a neuroscience-focused laboratory. In March 2015, she joined Astellas, where she worked on late-phase clinical trials as a medical monitor before transitioning to Astellas Pharma Global Development in 2018. She currently serves as the Global Medical Lead for investigational gene therapy projects targeting Pompe disease.

AT845 gene replacement therapy for late-onset Pompe disease: an update on safety and preliminary efficacy data from FORTIS, a phase 1/2 open-label clinical study

Chieri Hayashi[1], Tahseen Mozaffar[2], Nicola Longo[3], Mark Walzer[1], Achim Steup[1], David Viskochil[4], Jordi Diaz-Manera[5]

[1] Astellas Pharma Global Development, Northbrook, IL, USA;
[2] University of California Irvine, Irvine, CA, USA;
[3] University of California Los Angeles, CA, USA;
[4] University of Utah, Salt Lake City, UT, USA;
[5] John Walton Muscular Dystrophy Research Centre, Newcastle University and Newcastle Hospitals NHS Foundation Trust, Newcastle upon Tyne, UK

INTRODUCTION: Pompe disease is a rare, autosomal recessive disease caused by deficiency of acid alpha-glucosidase (GAA), leading to the accumulation of lysosomal glycogen and damage to skeletal and cardiac muscles. AT845, a gene replacement therapy to express human GAA in muscle tissues, is in clinical development for late-onset Pompe disease (LOPD). METHODS: FORTIS (NCT04174105) is an ongoing multicenter, open-label, ascending dose phase 1/2 first-in-human clinical trial to determine if AT845 is safe and tolerable in adults with LOPD. Participants receive a single intravenous infusion of AT845 with subsequent monitoring of safety, clinical, and biochemical endpoints. RESULTS: At the data snapshot (September 30, 2024), the first six participants in FORTIS had received AT845 at a dose of $3x10^{13}$ vg/kg (n=2) or $6x10^{13}$ vg/kg (n=4) and had been followed for at least 9 months. The first three dosed participants had completed at least 3 years' follow-up, the fourth had completed at least 2 years, the fifth had been followed for at least a year, and the sixth had completed 36 weeks of follow-up. Most treatment-emergent adverse events (TEAEs) were mild or moderate (grade 1 or 2) and considered unrelated to study treatment. A medically significant grade 2 peripheral sensory neuropathy was reported in one participant in the $6x10^{13}$ vg/kg cohort. Five of the six participants developed steroid-responsive transaminitis that was mostly mild-to-moderate and deemed possibly related to AT845. Two participants not included in the snapshot reported grade 3 serious, possibly related, TEAEs characterized by elevated transaminases. All participants demonstrated transduction of AT845, with increases in muscle GAA levels and activity at greater than or equal to one post-dose timepoint. At the data snapshot, five of the six participants had discontinued enzyme replacement therapy (ERT). Final ERT doses were received 10, 15, 15, 17, and 24 weeks after AT845 dosing, respectively, meaning that the five participants had remained off ERT for approx. 1–3.5 years. For the first six dosed participants, forced vital capacity, 6-minute walk test, patient-reported fatigue scores (PROMIS-fatigue), and Pompe disease-specific patient-reported ability to perform daily activities and social participation per the Rasch-built Pompe-specific Activity (R-PAct) assessment were stable up to 3 years post-dosing, including following ERT withdrawal. CONCLUSIONS: In this study, AT845 has a manageable safety profile. Following treatment with AT845, most recipients have been able to stop ERT without an acute significant decline in clinical status.

Yusuke MURAKAMI

Department of Ophthalmology, Kyushu University, Associate Professor

CURRICULUM VITAE

1997-2003	**M.D.**, Faculty of Medicine, Kyushu University
2005-2009	**Ph.D**., Department of Pathology, Graduate School of Medical Sciences, Kyushu University
2009-2012	**Research fellow**, Massachusetts Eye and Ear Infirmary, Harvard Medical School
2012-2015	**Retina fellow**, Department of Ophthalmology, Kyushu University Hospital
2015-2019	**Assistant Professor in Ophthalmology**, Graduate School of Medical Sciences, Kyushu University
2019-2024	**Lecturer in Ophthalmology,** Graduate School of Medical Sciences, Kyushu University
2024-Present	**Associate Professor in Ophthalmology**, Graduate School of Medical Sciences, Kyushu University

Gene-Specific and Gene-Agnostic Therapies for Inherited Retinal Diseases

Yusuke MURAKAMI

Department of Ophthalmology, Kyushu University, Associate Professor

Inherited retinal dystrophies (IRDs), including retinitis pigmentosa (RP), are intractable retinal disorders characterized by progressive degeneration of rod and cone photoreceptors, ultimately leading to blindness. For many years, management relied largely on low-vision care and societal support. However, the recent approval in Japan of the gene therapy voretigene neparvovec (Luxturna®) for *RPE65*-associated IRD has marked a turning point. In parallel, genetic testing—previously limited to research settings—has now become available in clinical practice. Looking ahead, gene-specific therapies targeting other IRD genes, such as *RPGR*, *CEP290*, *CHM*, *CNGA3/CNGB3*, *USH2A* and *CYP4V2*, are actively being developed, and additional gene therapy products are expected to emerge in coming years.

While conventional gene therapies are applicable to only a small subset of patients, gene-agnostic approaches aimed at delaying disease progression may offer a more broadly applicable strategy for RP and other retinal degenerative diseases. Based on this concept, we are developing a neuroprotective gene therapy using a lentiviral vector encoding pigment epithelium–derived factor (PEDF) (development code: DVC1-0401). In an investigator-initiated phase 1/2a clinical trial, 12 patients with RP received subretinal administration of DVC1-0401 in a dose-escalation manner. The trial demonstrated no SAEs related to DVC-0401, and over a 3-year follow-up period, treated eyes showed significantly slower disease progression compared with fellow untreated eyes, supporting its neuroprotective effect.

In this lecture, I will summarize and discuss the latest advances in both gene-specific and gene-agnostic therapeutic strategies for IRDs.

Katie Wachtel

Vice President, Regulatory Affairs, Lilly Gene Therapy, Eli Lilly and Company

CURRICULUM VITAE

2023-	Vice President, Regulatory Affairs, Lilly Gene Therapy, Eli Lilly and Company
2020-2023	Vice President, Regulatory Affairs, Akouos

Education:

2010	The Johns Hopkins University, M.S. in Biotechnology
2005	The Pennsylvania State University, B.S. in Biology

Preliminary Safety and Efficacy of AK-OTOF Gene Therapy for Otoferlin Gene (OTOF)-mediated Hearing Loss

Katie Wachtel

Vice President, Regulatory Affairs, Lilly Gene Therapy, Eli Lilly and Company

Background: The otoferlin gene (OTOF) encodes otoferlin, a protein critical for signaling at the inner hair cell synapse. Individuals with mutations in OTOF typically present with congenital, severe to profound sensorineural hearing loss, with preserved otoacoustic emissions early in life. Recent advances in gene therapy and intracochlear delivery support the potential to restore physiologic hearing in individuals with OTOF-mediated hearing loss using a one-time, local administration of AK-OTOF (AAVAnc80-hOTOF). The objective of this gene therapy trial for OTOF-mediated hearing loss is to assess the safety, tolerability, and efficacy of escalating doses of investigational therapy AK-OTOF.

Methods: Clinical trial participants have profound hearing loss at baseline as assessed by auditory brainstem response (ABR) and receive a one-time intracochlear administration of AK-OTOF using a minimally invasive transcanal approach and the Akouos Delivery Device. Safety assessments and auditory function tests, including ABR and behavioral audiometry, are performed over the trial and long-term follow-up periods.

Results: The preliminary safety profile is favorable, with no related serious adverse events and no adverse events related to AK-OTOF gene therapy or the Akouos Delivery Device. Initial clinical evidence shows that AK-OTOF gene therapy has the potential to restore physiologic hearing in individuals with OTOF-mediated hearing loss. Safety and efficacy data (through at least 3 months post-administration) from participants will be presented including follow up beyond 1 year for some participants.

Conclusions: Available data suggest that AK-OTOF may be safely administered to participants, with onset of restoration of hearing as early as one month following administration.

Yuko Kato-Mori

Head of ViSpot Division, Takara Bio Inc.,

CURRICULUM VITAE

1989 B.Sc., Faculty of Agriculture, Hokkaido University
2000 Research Fellow, Japan Society for the Promotion of Science (JSPS)
2002 Ph.D., Graduate School of Agriculture, Hokkaido University
Senior Researcher, Meiji Feed Co., Ltd.
Researcher, Veterinary Virology Unit / Laboratory of Pathogen Management, School of Veterinary Medicine, Rakuno Gakuen University.
Since 2016, Associate Professor, Graduate School of Science, Technology and Innovation, Kobe University, engaged in research on the development of viral safety management technologies for the Manufacturing Technology Association of Biologics.
Joined ViSpot Inc. in 2018; current position since 2025.

Publications

“International Trends Toward a New Era of Viral Safety for Biopharmaceuticals,” PHARM TECH JAPAN (2024) 4:1661–1664.
“Concepts of Platform Validation and Utilization of Prior Knowledge,” Pharmacia (2024) 60:516–521.

Testing service for evaluation of viral safety at Takara Bio

Yuko Kato-Mori

Head of ViSpot Division, Takara Bio Inc.,

Biotechnology products, including biotherapeutics and other biological products derived from human or animal cell lines, inherently carry a risk of viral contamination due to the nature of their manufacturing processes. This risk is particularly critical for animal cell–derived products such as monoclonal antibodies, recombinant proteins, viral vectors, and cell-based therapies, which require appropriate management of both exogenous and endogenous viruses. Past incidents of viral contamination in manufacturing processes have demonstrated the potential for serious economic and clinical consequences, leading to the establishment of international regulatory frameworks such as ICH Q5A.

This presentation outlines the current framework, testing strategies, and recent revisions to guidelines for virus safety assessment in biotechnology products.

Virus safety is ensured through a comprehensive, three-pronged (“tripod”) approach consisting of testing of raw and starting materials, evaluation of virus removal by the manufacturing process, and testing of in-process intermediates and final products. Because product testing alone cannot statistically demonstrate the absence of viruses, virus clearance studies play a critical role in establishing process capability and ensuring overall viral safety.

While this approach is well established for many recombinant protein products, including antibody therapeutics, viral vector- and cell therapy–based products present additional challenges. These products often have limited opportunities for conventional virus inactivation or removal (clearance) steps during manufacturing, necessitating additional risk mitigation strategies.

The seminar will also discuss the emerging role of next-generation sequencing (NGS) in virus safety assessment. NGS provides a powerful tool for the detection of exogenous viruses and has been formally incorporated into the revised ICH Q5A(R2) guideline. Its application has the potential to significantly advance virus safety testing, and Takara Bio’s initiatives in this area will also be presented.

Kent Christopherson, PhD

Head of Global Medial Affairs, Orchard Therapeutics

CURRICULUM VITAE

Career History

1997	B.S. Biology, Bradley University
2001	Ph.D. BioChm/Mol Biol/Med Gen, Indiana University School of Medicine
2001-2003	Post-Doctoral Research Fellow, Indiana University School of Medine
2003-2006	Assistant Prof., Molecular Medicine, University of Texas Health Science Ctr.
2006-2012	Associate Prof., Div Hem/Onc/Cell Therapy, Rush University Medical Ctr.
2012-2017	Biogen
2017-2019	Ultragenyx Pharmaceutical
2019-Present	Orchard Therapeutics

Writings

30 peer reviewed manuscripts
7 reviews / book chapters

Clinical Development of Atidarsagene Autotemcel, “arsa-cel,” Autologous Hematopoietic Stem Cell Gene Therapy (HSC-GT) for Treatment of Early-Onset Metachromatic Leukodystrophy (MLD), and Implications for MLD Newborn Screening (NBS)

Kent Christopherson, PhD

Head of Global Medial Affairs, Orchard Therapeutics

Metachromatic leukodystrophy (MLD) is a rare, rapidly progressive, irreversible and ultimately fatal neurometabolic disease that affects approximately one in 100,000 live births globally. The disease is caused by a deficiency of arylsulfatase A (ARSA), leading to sulfatide accumulation and subsequent progressive demyelination, neurodegeneration, loss of motor and cognitive skills.

Atidarsagene Autotemcel, “arsa-cel,” formerly known as OTL-200, is an ex vivo autologous hematopoietic stem cell gene therapy (HSC-GT) that aims to correct the underlying genetic cause of MLD by inserting one or more functional copies of the human ARSA gene into the genome of a patient’s own hematopoietic stem cells using a lentiviral vector. The genetically repaired cells are infused back into the patient, where, once engrafted, they differentiate into multiple cell types, some of which migrate across the blood-brain barrier into the central nervous system and express the functional ARSA enzyme.

Over 20 years of pre-clinical and clinical research has culminated in an integrated Clinical Development Program data set from early-onset pediatric MLD patients enrolled in two single-arm, open-label clinical studies or treated under European expanded access frameworks. Outcomes of arsa-cel treated patients were compared with natural history data from untreated patients. The patient population collectively referred to as early-onset MLD includes pre-symptomatic late infantile (PSLI), pre-symptomatic early juvenile (PSEJ), and early-symptomatic early juvenile (ESEJ) sub-types. A fully enrolled clinical trial of arsa-cel treatment in late juvenile (LJ) MLD patients is ongoing. The arsa-cel program was originated by and developed in partnership with the San Raffaele-Telethon Institute for Gene Therapy (SR-Tiget) in Milan, Italy.

Arsa-cel was approved by the European Commission in December 2020 and UK Medicines and Healthcare products Regulatory Agency in January 2021 (tradename: Libmeldy®); and the Food and Drug Administration in the United States in March 2024 (tradename: Lenmeldy™) and is indicated for the treatment of early-onset MLD. Arsa-cel is considered an investigational drug product in all other geographies, including, but not limited to, Saudi Arabia and Japan. Recently arsa-cel has received Orphan Drug Designation and Priority Review from the Saudi Food and Drug Authority and Orphan Regenerative Medicine Product Designation by Japan's Ministry of Health, Labor and Welfare.

A critical element of newborn screening (NBS) adoption necessitates pilot studies. An international expert group of clinicians, scientists and advocates have come together through the MLD NBS Alliance for the past 5 years to support evidence generation for MLD NBS. Suitable and specific assays and algorithms for MLD NBS have been developed and published based on measurement of sulfatide by LC-MS/MS, ARSA enzyme activity, and confirmatory molecular testing. A robust package of necessary evidence has been generated to support the implementation of universal NBS for MLD that is based on prospective investigator-initiated NBS studies for MLD throughout the U.S., Europe and the Middle East. In addition, multiple manuscripts have been published underscoring consensus guidelines, feasibility and cost-effectiveness of NBS for MLD.

Hiroya Kuwahara

Associate Professor
Department of Neurology and Neurological Science / NucleoTIDE and PepTIDE
Drug Discovery Center (TIDE), Institute of Science Tokyo

CURRICULUM VITAE

Dr. Hiroya Kuwahara graduated from the Faculty of Medicine at Tokyo Medical and Dental University in 2002 and earned his Ph.D. in Medicine from the Graduate School of Tokyo Medical and Dental University in 2011. With over two decades of clinical experience in neurology, he provides specialized care for both common neurological disorders and rare, intractable diseases. He has been a translational researcher, focusing on nucleic acid therapeutics (Nat Commun 6: 7969, 2015, etc) and on drug delivery systems targeting the central nervous system (Nat Commun 8: 1001, 2017, etc). In addition, he served as a Medical Officer at the Japanese Ministry of Health, Labour and Welfare, contributing to the advancement of medical research and development nationwide. As a member of the Future Vision Committee of the Japanese Society of Neurology, he is dedicated to fostering drug discovery and innovation through collaborative initiatives linking industry, government, and academia.

Challenges toward personalized nucleic acid drug development for rare genetic disorders

Hiroya Kuwahara

Associate Professor
Department of Neurology and Neurological Science / NucleoTIDE and PepTIDE
Drug Discovery Center (TIDE), Institute of Science Tokyo

Recent breakthroughs in genome analysis technologies have dramatically increased the number of genetic disorders in which causative genes can be identified in individual patients. Personalized drug development for extremely ultra-rare conditions—often referred to as N-of-1+ therapeutics—is an emerging paradigm in which medicines are designed and manufactured for a single patient or a very small group, integrating safety and efficacy evaluation within a unified framework. Uniquely, this process completes all phases equivalent to Phase I–III clinical trials in just one patient.

For rare, intractable diseases affecting only a handful of patients worldwide, traditional industry-led drug development is often unfeasible, leaving an unmet medical need. However, when the pathogenic genetic mutation is known, academia-driven personalized nucleic acid therapeutics—such as antisense oligonucleotides—are attracting international attention as a promising strategy that could enable fundamental treatment.

In the United States, significant progress has already been made. The U.S. Food and Drug Administration (FDA) has issued guidance for conducting individualized clinical trials using antisense oligonucleotides in patients with poor prognosis or severe disabilities. The n-Lorem Foundation, a non-profit organization established by the founder of Ionis Pharmaceuticals, designs and manufactures patient-specific antisense oligonucleotides and has administered them to more than 40 individuals to date.

In Japan, we are working toward implementing personalized nucleic acid drug development by establishing a multi-stakeholder collaborative framework. Centered at the NucleoTIDE and PepTIDE Drug Discovery Center (TIDE), Institute of Science Tokyo, this initiative connects clinicians, researchers, pharmaceutical companies, regulatory authorities, and patient advocacy groups. We have also launched a plan for clinical trial using antisense oligonucleotides targeting a patient with extremely rare disease within Japan.

In this forum, I will present our progress to date and discuss future perspectives and challenges for personalized nucleic acid drug development tailored to rare genetic disorders.

Hiroshi Kobayashi, M.D.

Division of Gene Therapy, Research Center for Medical Sciences
The Jikei University School of Medicine.
Professor

CURRICULUM VITAE

Career History

1994 Assistant Professor, Department of Pediatrics, The Jikei University
2002 Study abroad at Research Immunology and BMT in the Children's Hospital LosAngeles, University of Southern California
2004 Returned to Japan and resumed the position at the Jikei Univ.
2006 Received a Doctor of Medicine degree, Lecturer and Chief Medical Officer
2012 Associate Professor, Division of Gene Therapy, Research Center for Medical Sciences
2020 Professor, Division of Gene Therapy, Research Center for Medical Sciences
2021 Director of the Gene Therapy Research Division

Academic Awards Received

2015. The 4th ACIMD (Asian Congress for Inherited Metabolic Diseases) Travel Award, 2015 Taipei
2017. 59th Japanese Society of Inherited Error of Metabolism (JSIEM), Academic Clinical Education Award

ERT, as a treatment approaches for genetic disorders

Hiroshi Kobayashi

Division of Gene Therapy, Research Center for Medical Sciences
The Jikei University School of Medicine.
Professor

Treatment strategies for genetic diseases have traditionally focused on symptomatic management. In recent decades, however, enzyme replacement therapy (ERT) has enabled direct intervention at the pathogenic level in inherited metabolic disorders by supplementing deficient enzymes at the protein level. ERT has been developed and clinically implemented for several lysosomal storage disorders, including Fabry disease, Pompe disease, mucopolysaccharidoses types I, II, IV, VI, and VII, and Gaucher disease. Regular exogenous administration of the deficient enzyme allows cellular uptake via specific receptors, thereby reducing substrate accumulation and preventing or slowing the progression of organ damage. This process is facilitated by the exposure of carbohydrate moieties, such as mannose-6-phosphate, on the surface of the enzyme, which enables receptor-mediated endocytosis into target cells. Clinically, ERT has been associated with improvements such as reductions in hepatosplenomegaly, enhancement of cardiopulmonary function, and improved skeletal and respiratory muscle performance, collectively contributing to improved patient quality of life (QOL). Importantly, early initiation of therapy during infancy or childhood is critical, as it may prevent irreversible organ damage. Recent advances in the field include the development of novel enzyme formulations such as modified enzymes designed to cross the blood–brain barrier, optimization of administration routes, and combination therapies incorporating pharmacological chaperones. Some of these approaches have already entered clinical practice. This lecture will review the efficacy and limitations of ERT and provide an integrated perspective on its role alongside next-generation therapeutic strategies for the treatment of genetic disorders.

Empowering Young Leaders to Drive Genetic Medicine from Japan to the World ／ 遺伝病治療を日本から世界へ発信するために

Takafumi Nakamura

Department of Genomic Medicine and Regenerative Therapeutics, Faculty of Medicine, Tottori University

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名城大学／ Innovation for NEW HOPE 学生アンバサダー

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Meiji Pharmaceutical University / student ambassadors from Innovation for NEW HOPE

菊池　彩華

明治薬科大学／ Innovation for NEW HOPE 学生アンバサダー

Generate Innovation with Young Power: The Future of Gene Therapy Driven by the Next Generation

Takafumi Nakamura[1], Yoshiyuki Saito[2], Haruka Mizuno[3], Ayaka Kikuchi[4], Hitoshi Osaka[5]

[1] Dept of Genomic Medicine and Regenerative Therapeutics, Faculty of Medicine, Tottori University, Yonago, Tottori
[2] Faculty of Pharmaceutical Sciences, The University of Tokyo, Tokyo
[3] Meijo University, School of Pharmacy, Nagoya, 6th-year undergraduate
[4] Meiji Pharmaceutical University, Tokyo, M1
[5] Department of Pediatrics, Jichi Medical University, Shimotsuke, Tochigi

This session aims to foster the next generation of leaders in gene and cell therapy by integrating three essential perspectives: education, industrialization, and engagement with civil society. We will discuss how innovation in gene therapy can be accelerated and sustainably translated into patient care in Japan.

First, Dr. Takafumi Nakamura (Tottori University) will provide an overview of the current state and challenges of education in gene therapy for young researchers. While structured training systems that integrate basic science, non-clinical development, clinical trials, and regulatory science have been established in Europe and the United States, Japan still faces fragmentation of educational resources and limited opportunities for hands-on translational training. This lecture will highlight where young scientists encounter the greatest barriers, what competencies are currently lacking, and how Japan must redesign its educational framework to nurture professionals capable of translating research into real-world therapies.

Next, Dr. Yoshiyuki Saito (The University of Tokyo) will discuss the innovation ecosystem required to transform gene therapy into a viable medical industry. Beyond advanced scientific technologies, gene therapy requires robust infrastructure in regulatory affairs, manufacturing, quality control, and international collaboration. Despite outstanding achievements in basic research, Japan continues to struggle with drug lag and drug loss, resulting in promising innovations failing to reach patients in a timely manner. Dr. Saito will present structural challenges in the current system and outline how academia, industry, and regulatory authorities must collaborate more closely. He will also emphasize how young researchers can and should engage in the process of industrialization and social implementation.

In the final part, student ambassadors from Innovation for NEW HOPE (managed by Astellas Pharma Inc.), Ms. Haruka Mizuno and Ms. Ayaka Kikuchi, will present their perspectives as non-researchers on the current state and future of gene therapy. Through their activities, they have encountered patients and families who received gene therapy, as well as physicians and researchers working at the forefront of this field. They were deeply moved by the hope that cutting-edge therapies bring to patients and simultaneously learned about the serious challenges Japan faces, including drug lag, drug loss, and information gaps between experts and the general public. Their presentation will share what they noticed, felt, and learned as citizens and young members of society, and will conclude with a message of encouragement to researchers and healthcare professionals.

This session seeks to visualize three major bottlenecks hindering the advancement of gene therapy in Japan—education, industrialization, and the disconnect with civil society—and to clarify the essential role of young power in overcoming these challenges. By bringing together researchers, industry leaders, students, patients, and citizens in a shared dialogue, this session aims to generate new collaborations and a shared vision for establishing gene and cell therapy as a core medical innovation of the next generation, delivered to patients as swiftly and equitably as possible.

Miho Matakatsu

VectorBuilder Inc.

CURRICULUM VITAE

Education

1995~1998 The Graduate University for Advanced Studies-SOKENDAI
Ph.D. in Life Sciences/Developmental Genetics

Professional Career

1998~2003 Post Doctoral Fellow, National Institute of Health, NIH, Bethesda, MD, USA

2003~2014 Research Professional, Ben May Cancer Department, The University of Chicago, Chicago, IL, USA

2014~2017 Senior Researcher, Dept. Otolaryngology head and neck surgery, Northwestern Medicine, Northwestern University, Chicago, IL, USA

2017~current Managing Director, VectorBuilder Japan Inc. VectorBuilder Inc. Chicago, IL, USA

Publication

1 ●Matsuoka AJ, Morrissey ZD, Zhang C, Homma K, Belmadani A, Miller CA, Chadly DM, Kobayashi S, Edelbrock AN, **Tanaka-Matakatsu M**, Whitlon DS, Lyass L, McGuire TL, Stupp SI, Kessler JA.: Directed differentiation of human embryonic stem cells toward placode-derived spiral ganglion-like sensory neurons. Stem Cells Translational Medicine. Mar; 6(3): 923-936, 2017.

2 ●Kato K., Dong B., Wada H., **Tanaka-Matakatsu M.**, Yagi Y and Hayashi S.: Microtubule-dependent balanced cell contraction and luminal matrix modification accelerate epithelial tube fusion. *Nature Communications*, Vol.7, 111141 Published 12 April, 2016.

3 ●**Tanaka-Matakatsu, M.**, Miller, J and Du W.: The homeodomain of Eyeless regulates cell growth and antagonizes the paired domain-dependent retinal differentiation function. *Protein & Cell*, Vol.6, 68-78, 2015.

4 ●**Tanaka-Matakatsu, M.**, Miller, J, Borger D, Tang W.-J. and Du W.: Daughterless homodimer synergizes with Eyeless to induce Atonal expression and retinal neuron differentiation. *Developmental Biology*, Vol. 392, 256-265, 2014.

5 ●**Tanaka-Matakatsu, M.** Xu J., Cheng L. and Du W.: Regulation of apoptosis of *rbf* mutant cells during *Drosophila* development. *Developmental Biology*, Vol. 326, 347-356, 2009.

Accelerating Gene Therapy Development with VectorBuilder

Miho Matakatsu

VectorBuilder Inc.

VectorBuilder is a full-service CDMO with extensive experience in cGMP vector manufacturing services. We operate multiple advanced facilities that provide support for customers throughout their drug-discovery processes, offering research-grade vectors for early discovery, GMP-like vectors for preclinical testing, and full GMP-grade vectors for clinical trials. We have provided IND-enabling vectors to a worldwide client base in the US, Europe, Japan, China and South Korea. Our CDMO services include process development, analytical development, cell banking, fill/finish, and regulatory support.

In this presentation, I will introduce VectorBuilder's CDMO services as well as brief results from our recent GMP AAV manufacturing services for Lantu Biopharma on their AAV gene therapy pipelines to Menkes disease Investigator-Initiated Trial (IIT) and Phase 1 Spinal muscular atrophy (SMA) gene therapy in Southeast Asia countries.

ALBERTO AURICCHIO, MD

- AAVantgarde Bio srl, Milan, Italy
- Telethon Institute of Genetics and Medicine (TIGEM) in Pozzuoli, Italy
- Department of Advanced Biomedical Sciences, “Federico II” University. Naples, Italy

CURRICULUM VITAE

Career History

- Professor of Medical Genetics at the Department of Advanced Biomedical Sciences, “Federico II” University.Naples, Italy (2017-present)
- Founder and Chief Scientific Officer of AAVantgarde Bio s.r.l (2021-present)
- President of the European Society of Gene and Cell Therapy (ESGCT, 2024–2026)
- Scientific Director of the Telethon Institute of Genetics and Medicine (TIGEM) in Pozzuoli, Italy (2024-present)

Academic Awards Received

- Telethon Fellowship 371/B for Italian Scientists Abroad (1999-2001).
- “Image of the Year” Award, 49th Annual Meeting of the Society of Nuclear Medicine, Los Angeles, CA, USA (2002).
- Ruth and Milton Steinbach Fund Award, NYC (2003-2006).
- Startcup Award for the “Angiotech” business plan, “Federico II” University, Naples, Italy (2005)
- “Outstanding New Investigator” Award, American Society of Gene Therapy (2006)
- Foundation “Fighting Blindness” Board of Director's Award (2009)
- “Visionary of the Quarter” award of the European Vision Institute (2009/2010)
- “European Research Council Consolidator Grant” (RetGeneTx #282085), European Research Council (2011)
- “European Research Council Advanced Grant” (EYEGET #694323), European Research Council (2016)
- “European Research Council Proof-of-Concept Grant” (GENEVISION #755075), European Research Council (2017)
- International Award for Scientific Research “Arrigo Recordati”, Rotterdam NL (2019)
- International Award “Bonifacio VIII” - XX edition, by the Accademia Bonifaciana, Anagni (FR), Italy (2022)
- “European Research Council Advanced Grant” (EXPEDITE #101097155), European Research Council (2023)
- Italian Knowledge Leader Award, Convention Bureau Italia, (2025)

ExpEditing AAV gene therapy

ALBERTO AURICCHIO, MD

- AAVantgarde Bio srl, Milan, Italy
- Telethon Institute of Genetics and Medicine (TIGEM) in Pozzuoli, Italy
- Department of Advanced Biomedical Sciences, “Federico II” University.Naples, Italy

In vivo gene therapy with adeno-associated viral (AAV) vectors is holding its promise for treatment of genetic diseases, yet some challenges remain that prevent to expand this approach to a larger patients population. These include: AAV cargo capacity limited to about 5 kb which prevents their application to conditions due to mutations in genes with a larger coding sequence; the episomal status of AAV genomes which results in short-term expression in proliferating tissues; efficient approaches to target toxic gain-of-function mutations that do not benefit from traditional gene addition. To overcome these limitations, we have developed platforms based on the co-delivery of 2 AAV vectors that either EXPand AAV transfer capacity or EDIT genomic loci by stably integrating therapeutic donor DNAs. We provide proof-of-concept of the efficacy and safety of these approaches in animal models of human inherited diseases, and, more recently, in humans.

Acknowledgments

共催

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（五十音順：2026 年 1 月 15 日）

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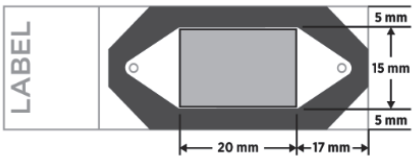

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Mallach A, et al.
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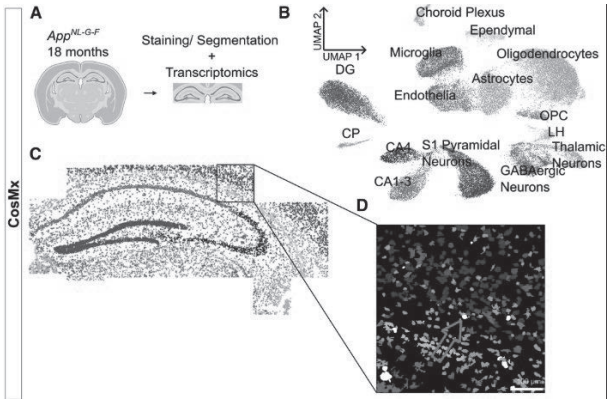

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	PanCK	Cytosol	3
	CD45	Leukocyte	4
Neuroscience (RNA)	DAPI	Nuclear	1
	rRNA	Cytosol	2
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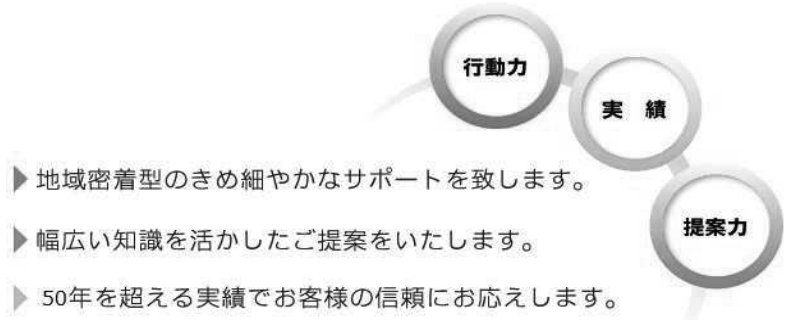

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