

Abstracts of papers presented
at the 2025 Cold Spring Harbor Asia Conference

SHORT TANDEM REPEATS IN GENE REGULATION & DISEASES

November 3–November 7, 2025

Arranged by

Peng Jin, *Emory University (co-chair)*

Peter Todd, *University of Michigan (co-chair)*

Barbara Bardoni, *Institut de Pharmacologie Moléculaire et Cellulaire*

Linda Gu, *The Chinese University of Hong Kong*

Kinya Ishikawa, *Tokyo Medical and Dental University*

Beisha Tang, *Xiangya Hospital, Central South University*



Cold Spring Harbor Conferences Asia
Cold Spring Harbor Laboratory



SHORT TANDEM REPEATS IN GENE REGULATION AND DISEASES

Monday, November 3 – Friday, November 7, 2025

Monday	7:00 pm	1 Opening Keynote Session
Tuesday	9:00 am	2 Transposable Element-associated Repeat Disorders
Tuesday	2:00 pm	Poster Session
Tuesday	7:00 pm	3 CAG Repeat Expansion Disorders I
Wednesday	9:00 am	4 CAG Repeat Expansion Disorders II
Wednesday	1:30 pm	<i>Visit to Old Suzhou*</i>
Wednesday	7:00 pm	5 ALS-associated Repeat Expansions
Thursday	9:00 am	6 Repeats in Cellular Function and Human Disease
Thursday	2:00 pm	7 Short Tandem Repeat Variation in Human Traits
Thursday	5:00 pm	<i>Cocktails and Banquet</i>
Friday	9:00 am	8 Therapeutic Development for Repeat Expansion Disorders

Oral presentation sessions are located in the CSHA Auditorium

Poster session is in the Lake Front Hall.

Cocktail social hour is held outside in the Suz Garden.

Old Suzhou visits depart from the CSHA lobby

**optional tour requires additional fee.*

Meal locations and times are as follows:

Lunch: Main Cafeteria 12:00pm - 1:30pm

Dinner: Main Cafeteria 6:00pm - 7:30pm

Banquet: Suz Garden 6:00pm

More information will be available at CSHA office.

(Map at the end of this abstract book)

PROGRAM

MONDAY, November 3—7:00 PM

SESSION 1 OPENING KEYNOTE SESSION

Chairperson: **Peng Jin**, Emory University, Atlanta, Georgia, USA

KEYNOTE SPEAKER

Mechanistic Steps in Huntington's disease—From somatic repeat expansion to selective neuronal pathogenesis

X. William Yang [35'+10']

Presenter affiliation: University of California at Los Angeles, Los Angeles, California.

1

An emergent disease-associated motor neuron state precedes cell death in a mouse model of ALS

Aaron D. Gitler [20'+10']

Presenter affiliation: Stanford University, Stanford, California.

2

Understanding human disease through millions of tandem repeats across millions of genomes

Wei Li [10'+5']

Presenter affiliation: University of California, Irvine, Irvine, California.

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TUESDAY, November 4—9:00 AM

SESSION 2 TRANSPOSABLE ELEMENT-ASSOCIATED REPEAT DISORDERS

Chairperson: **Peter Todd**, University of Michigan, Ann Arbor, Michigan, USA

KEYNOTE SPEAKER

Expanding future of exploring tandem repeat expansions in human diseases

Shoji Tsuji [35'+10']

Presenter affiliation: International University of Health and Welfare, Chiba, Japan; The University of Tokyo, Tokyo, Japan.

4

Genetics and functional mechanisms of non-coding repeat expansion causing CANVAS and oculopharyngodistal myopathy

Andrea Cortese [20'+10']

Presenter affiliation: UCL Queen Square Institute of Neurology, London, United Kingdom; University of Milan, Pavia, Italy.

5

FNOP spectrum disorders caused by CGG and CCG repeat expansions

Hiroyuki Ishiura [20'+10']

Presenter affiliation: Okayama University, Okayama, Japan.

6

RE Pathways—A national program to improve diagnosis and management of cerebellar ataxia in Australia

Haloom Rafehi, Penny Snell, Kayli Davies, Liam Fearnley, Lauren Sanders, Ian Harding, Martin Delatycki, David Szmulewicz, Melanie Bahlo, Paul J. Lockhart [20'+10']

Presenter affiliation: Murdoch Children's Research Institute, Melbourne, Australia.

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Break

Small-molecules specific to GC-rich repeat DNA structures—A novel therapeutic avenue?

David A. Ripsman, Terence Gall Duncan, Mahreen Khan, Amit L. Deshmukh, Gagan B. Panigrahi, Isha Singh, Bimolendu Das, Stella Lanni, Mila Mirceta, Peixiang Wang, Masayuki Nakamori, Kazuhiko Nakatani, Yuh-Hwa Wang, Bitu Zamiri, Christopher E. Pearson [20'+10']

Presenter affiliation: The Hospital for Sick Children, Toronto, Canada.

8

Post-silencing pathogenic cascades in short tandem repeat initiated neurodegeneration

Richard M. Albertson, Abby Cheng, Peter K. Todd [10'+5']

Presenter affiliation: University of Michigan, Ann Arbor, Michigan.

9

RNA toxicity and interacting RNA-binding protein NOVA2 of (UUUCA)_{exp} RNA foci in familial cortical myoclonic tremor with epilepsy

Fan Zhang, Yiling Chen, Zhidong Cen, Wei Luo [10'+5']

Presenter affiliation: The Second Affiliated Hospital, Zhejiang University School of Medicine, Hangzhou, China.

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POSTER SESSION

Potential involvement of (TTCCA) repeat expansions in the pathology of spinocerebellar ataxia type 31

Yuki Aoki, Kohji Mori, Shoshin Akamine, Ryota Uozumi, Yoshitaka Nagai, Manabu Ikeda

Presenter affiliation: The University of Osaka, Osaka-Suita, Japan. 11

Developing a peptide inhibitor to alleviate GGGGCC-induced C9ALS/FTD by suppressing RNA and protein toxicities

Hui Zhen Chen, Gang Chen, Ho Yin Chan

Presenter affiliation: The Chinese University of Hong Kong, Hong Kong, China. 12

Characterizing schizophrenia-relevant tandem repeats through long-read sequencing

Zhengyu An, Jingqi Chen, Xing-Ming Zhao

Presenter affiliation: Fudan University, Shanghai, China. 13

Blood-based somatic CAG repeat expansion and clinical progression in Huntington's disease patients from Western China

Si-Rui Zhang, Yang-fan Cheng, Jean-Marc Burgunder, Hui-Fang Shang

Presenter affiliation: West China Hospital, Sichuan University, Chengdu, China. 14

Redefining the pathogenic threshold and clinical spectrum of CAG expansions in spinal and bulbar muscular atrophy using the UK Biobank

Chon Him Choy, Matteo Zanovello, Christopher Clarkson, Pietro Fratta, Arianna Tucci

Presenter affiliation: University College London, London, United Kingdom. 15

CGG repeat expansions in Charcot-Marie-Tooth disease—Insights from the 100 000 Genomes Project and 1000 Genomes Project

Stefano Facchini, Alessandro Bertini, Ilaria Quartesan, Riccardo Currò, Ricardo Parolin Schnekenberg, Natalia Dominik, Gustavo Alves, Lucia Ferullo, Arianna Tucci, Henry Houlden, Mary M. Reilly, Andrea Cortese

Presenter affiliation: UCL Institute of Neurology, London, United Kingdom; Università di Pavia, Pavia, Italy. 16

PreciseSTR—A GPU-accelerated algorithm for de novo detection and high-resolution profiling of short tandem repeats from long reads

Ruihan Wang, Li Fang

Presenter affiliation: Sun Yat-sen University, Guangzhou, China.

17

Intranasal administration of an expanded CAG RNA-targeting peptide ameliorates Huntington's disease neuronal apoptosis and motor deficits

Maggie Lok I Leong, Huizhen Chen, Tsz Kwan Yau, Zhefan Stephen Chen, Terence Gall-Duncan, Jiahao Li, Jun Zhang, Jiajia Zhao, Clinton Turner, Lynette Tippet, Maurice A. Curtis, Richard L. M. Faull, Zhong Zuo, Christopher E. Pearson, Ting-Fung Chan, Jacky Chi Ki Ngo, Ho Yin Edwin Chan

Presenter affiliation: The Chinese University of Hong Kong, Hong Kong, China.

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Short tandem repeat variations that impact multiple molecular phenotypes in esophageal squamous cell carcinoma represent candidate causal risk loci

Chenghao Li, Wenyan Sun, Jiangcong Ma, Xuanxuan Gou, Xijin Chang, Zhenhui Chen, Wei Li, Jianzhong Su

Presenter affiliation: Harbin Institute of Technology, Harbin, China.

19

Improving a peptide inhibitor against hexanucleotide repeat expansion in C9ALS/FTD

Mingxi Ou, Zhefan Stephen Chen, Kevin Talbot, Chi Ki Jacky Ngo, Ho Yin Edwin Chan

Presenter affiliation: The Chinese University of Hong Kong, Hong Kong, China.

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Using mouse models to study the pathogenesis of spinocerebellar ataxia type 51 (SCA51)

Eshu Ruan

Presenter affiliation: Jinan University, Guangzhou, China.

21

Repeat element–anchored enhancer–promoter interactions (REEPIs) shape the regulatory landscape of hormone-resistant breast cancer

Xufeng Shu, Masaki Kato, Piero Carninci

Presenter affiliation: RIKEN, Yokohama, Kanagawa, Japan.

22

Involvement of small nerve fibres in NOTCH2NLC-related neuronal intranuclear inclusion disease

Yun Tian, Yijie Song, Beisha Tang, Lei Li, Lu Shen

Presenter affiliation: Xiangya Hospital, Central South University, Changsha, China.

23

PML targets and resolves structured protein inclusions to mitigate neurodegeneration

Yang Wang, Jia-Xin Zhu, Fei-Xia Zhan, Yuchen Xia, Yinfeng Guo, Yan-Hao Chen, Xin-Hua Luan, Xi-Ya Shen, Yu-Wen Cao, Xin Zhang, Li Cao, Steven X. Hou

Presenter affiliation: Fudan University, Shanghai, China.

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Rapamycin alleviates neurodegeneration in a Drosophila model of spinocerebellar ataxia type 51

Cuijie Wei, Jianwen Deng, Hui Xiong

Presenter affiliation: Peking University First Hospital, Beijing, China.

25

Engineering a series of site-specific RNA endonucleases that degrade toxic RNA repeats

Tong Wei, Miaowei Mao, Zefeng Wang

Presenter affiliation: Southern University of Science and Technology, shenzhen, China.

26

Gene therapy for spinal-bulbar muscular atrophy

Chi Wai Wong

Presenter affiliation: NeuMed Pharmaceuticals, Hong Kong, China.

27

Developing a peptide inhibitor to suppress expanded CGG toxicities in the FXTAS model

Yitao Wu, Ho Yin Chan

Presenter affiliation: The Chinese University of Hong Kong, Hong Kong, China.

28

Expanded polyglycine protein aggregates alter the RNA splicing profile into a premature state partially via sequestering SF3B4

Jiaxing Yang, Youqi Zheng, Boxun Lu, Huijuan Feng, Yuhua Fu

Presenter affiliation: Fudan University, Shanghai, China.

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Pre- and postsynaptic upregulation of FasII synergistically underlies neuropathological and behavioral phenotypes in a *Drosophila* model of myotonic dystrophy

Alex Chun Koon, Ka Yee Winnie Yeung, Yitao Wu, Lok I Leong, John Tsun Po Cheung, Zhefan Stephen Chen, Shaohong Isaac Peng, Noah S. Armstrong, C. Andrew Frank, Paul Magneron, Mário Gomes-Pereira, Ariadna Bargiela, Aline Huguet, Rubén Artero, Genevieve Gourdon, Vivian Budnik, Brian D. McCabe, Ho Yin Edwin Chan
Presenter affiliation: The Chinese University of Hong Kong, Shatin, Hong Kong, China.

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TUESDAY, November 4—7:00 PM

SESSION 3 CAG REPEAT EXPANSION DISORDERS I

Chairperson: **Kinya Ishikawa**, Institute of Science Tokyo, Tokyo, Japan

Striatal pathology—A bridge between the CAG triplet repeat disorders HD and SCA1

Harry T. Orr [20'+10']

Presenter affiliation: University of Minnesota, Minneapolis and Saint Paul, Minnesota.

31

Pathogenic mechanisms and somatic instability in spinal and bulbar muscular atrophy

Diane E. Merry [20'+10']

Presenter affiliation: Thomas Jefferson University, Philadelphia, Pennsylvania.

32

PolyQ-related ataxia in China—From bedside to bench

Hong Jiang, Zhao Chen, Su Yang, Beisha Tang [20'+10']

Presenter affiliation: Xiangya Hospital, Central South University, Changsha, China.

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Using non-human primates to study the pathogenic mechanisms of Huntington's disease

Su Yang [10'+5']

Presenter affiliation: Jinan University, Guangzhou, China.

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Targeted 3'end mRNA sequencing uncovers cryptic polyadenylation in Huntington's disease to somatic instability and CAG repeat purity

Ane Velasco, Maria Manterola, Alvaro Herrero-Reiriz, María Carazo, Olatz Arnold, Martina Hallegger, Jernej Ule, Adolfo Lopez de Munain, Marta Olejniczak, Veronica Brito, Lorea Blazquez [10'+5']

Presenter affiliation: Biogipuzkoa Health Research Institute, San Sebastian, Spain; Centre for Network Biomedical Research on Neurodegenerative Diseases , Madrid, Spain.

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Investigation of tau pathology in neuronal intranuclear inclusion disease

Yun Tian, Sizhe Zhang, Beisha Tang, Lu Shen [10'+5']

Presenter affiliation: Xiangya Hospital, Central South University, Changsha, China.

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WEDNESDAY, November 5—9:00 AM

SESSION 4 CAG REPEAT EXPANSION DISORDERS II

Chairperson: **Su Yang**, Jinan University, Guangzhou, China

Short tandem repeat mutations can explain peculiar inheritance yet modifiers and mechanisms remain to be discovered

David L. Nelson, Lifang Li, Yanghong Gu, Emily Allen, Peng Jin, Peter Todd [20'+10']

Presenter affiliation: Baylor College of Medicine, Houston, Texas.

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Genetic modifiers of Fragile X-associated conditions

Peng Jin [20'+10']

Presenter affiliation: Emory University School of Medicine, Atlanta, Georgia.

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The roles of G-quadruplexes in repeat expansion disorders

Norifumi Shioda [20'+10']

Presenter affiliation: Kumamoto University, Kumamoto, Japan.

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Break

Molecular mechanisms and gene therapy of neuronal intranuclear inclusion disease

Yongcheng Pan, Nina Xie, Kailin Zhang, Lu Shen, Hong Jiang, Beisha Tang, Peng Jin, Shihua Li, Xiao-Jiang Li, Qiong Liu [20'+10']

Presenter affiliation: Xiangya Hospital, Central South University, Changsha, China.

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GGC trinucleotide repeat expansions hidden within small ORFs of the human “non-coding” genome are translated into toxic polyglycine proteins in oculopharyngodistal myopathy

Manon Boivin, Jiaxi Yu, Zhaoxia Wang, Jianwen Deng, Nicolas Charlet-Berguerand [10'+5']

Presenter affiliation: Peking University First Hospital, Beijing, China.

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Translation of GIPC1 5'UTR GGC repeat expansions into a toxic PolyG protein in oculopharyngodistal myopathy 2

Kexin Jiao, Xinyu Chen, Mengye Cao Cao, Tao Wang, Jianying Xi, Wenhua Zhu, Chongbo Zhao [10'+5']

Presenter affiliation: Huashan Hospital, Fudan University, Shanghai, and National Center for Neurological Disorders (NCND), China, Shanghai, China.

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WEDNESDAY, November 5—1:30 PM

Visit to Old Suzhou

WEDNESDAY, November 5—7:00 PM

SESSION 5 **ALS-ASSOCIATED REPEAT EXPANSIONS**

Chairperson: **Beisha Tang**, Xiangya Hospital, Central South University, Changsha, China

Understanding C9ORF72-ALS/FTD—Novel insights from Drosophila and iPSC-derived patient motor neurons

Fen-Biao Gao [20'+10']

Presenter affiliation: University of Massachusetts Chan Medical School, Worcester, Massachusetts.

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Mechanisms and modifiers of C9orf72 repeat expansion in ALS-FTD

Lian-Huan Wei, Suzhou Yang, Juliana M. Salgado, Tanian Arab, Udit Sheth, Tania Gendron, Junjie U. Guo [20'+10']

Presenter affiliation: Yale University, New Haven, Connecticut. 44

RNA chaperones regulate RAN translation in repeat expansion disorders

Yoshitaka Nagai [20'+10']

Presenter affiliation: Kindai University, Osaka-Sayama, Japan. 45

Epitranscriptomic and epigenetic dysfunctions in C9ORF72-ALS/FTD

Shuying Sun [20'+10']

Presenter affiliation: Johns Hopkins University School of Medicine, Baltimore, Maryland. 46

Knockdown screening of translation-related factors reveals distinct mechanisms of AUG and RAN translation in C9orf72-FTD/ALS

Kohji Mori, Shiho Gotoh, Shizuko Kondo, Ryota Uozumi, Koujin Miura, Yuki Aoki, Tesshin Miyamoto, Yoshitaka Nagai, Manabu Ikeda [10'+5']

Presenter affiliation: The University of Osaka, Suita, Osaka, Japan. 47

THURSDAY, November 6—9:00 AM

SESSION 6 REPEATS IN CELLULAR FUNCTION AND HUMAN DISEASE

Chairperson: **Junjie Guo**, Yale University School of Medicine, New Haven, Connecticut, USA

Translation of repeats in C9orf72 FTD/ALS—How it starts

Peter K. Todd, Ross Kaufhold, Shannon Miller, Wenzhao Dong, Clare Wieland, Indranil Malik, Bradley Crone, Amy Krans, Len Petrucelli, Sami Barmada, Alan Boyle, Jay Brito Querido [20'+10']

Presenter affiliation: University of Michigan, Ann Arbor, Michigan; Ann Arbor Veterans Administration Healthcare, Ann Arbor, Michigan. 48

RNA supply chain issues in myotonic dystrophy

Eric Wang [20'+10']

Presenter affiliation: University of Florida, Gainesville, Florida. 49

CGG repeat expansion mutations contribute towards the missing heritability of neurodevelopmental disorders

Dale Annear, Geert Vandeweyer, Frank Kooy [20'+10']

Presenter affiliation: University of Antwerp, Antwerp, Belgium.

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Break

Pervasive tandem repeat heterogeneity and homeostatic constraint in the male germline

Lucas Finot, Yilei Fu, Cera J. Aex, Egor Dolzhenko, Kaitlyn Wong, Michelle Quach, Ashkan L. Pourabhari, Lucia Ramirez, Michael L. Eisenberg, Michael P. Snyder, Fritz J. Sedlazeck, Graham S. Erwin [20'+10']

Presenter affiliation: Baylor College of Medicine, Houston, Texas.

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Exploring neurodegeneration mechanisms of repeat expansion disorders by targeted degradation of lipid droplets

Boxun Lu, Yuhua Fu [20'+10']

Presenter affiliation: Fudan University, Shanghai, China.

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Splicing dysregulation and behavioral deficits in a neuronal mouse model of myotonic dystrophy (DM1)

Gary J. Bassell, Bethlehem A. Bekele, Juan Arboleda, Eric T. Wang [10'+5']

Presenter affiliation: Emory University , Atlanta, Georgia.

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A novel 5' UTR CCG expansion in *TBC1D7* causes oculopharyngodistal myopathy

Riccardo Curro, Liedewei Van de Vondel, Stefano Facchini, Isaac Zu, Jonathan De Winter, Ilaria Quartesan, Mark Roberts, Stefen Brady, Ashirwad Merve, Kornelia Neveling, Alexander Hoischen, Peter De Jonghe, Henry Houlden, Michael G. Hanna, Enrico Bugiardini, Stephan Zuchner, Jonathan Baets, Andrea Cortese [10'+5']

Presenter affiliation: UCL Queen Square Institute of Neurology, London, United Kingdom.

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SESSION 7 **SHORT TANDEM REPEAT VARIATION IN HUMAN TRAITS**

Chairperson: **Linda Gu**, The Chinese University of Hong Kong, Hong Kong, China

Tandem repeat size and sequence modify gene function and disease risk

Ryan K. Yuen [20'+10']

Presenter affiliation: The Hospital for Sick Children, Toronto, Canada; University of Toronto, Toronto, Canada.

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Genomic approaches for studying the role of tandem repeat variations in human disease

Andrew J. Sharp [20'+10']

Presenter affiliation: Icahn School of Medicine at Mount Sinai, New York, New York.

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Profiling variation in and around tandem repeats

Egor Dolzhenko, Ben Weisburd, Matt C. Danzi, Adam English, Mark F. Bennett, Tom Mokveld, Guilherme Sena, William J. Rowell, Zev Kronenberg, Nehir E. Kurtas, Harrison Brand, Melanie Bahlo, Peng Jin, David L. Nelson, Fritz J. Sedlazeck, Stephan Zuchner, Heidi L. Rehm, Michael A. Eberle [20'+10']

Presenter affiliation: PacBio, Menlo Park, California.

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Break

Population and mutational insights of repeat expansion diseases from short-read genome sequencing

Arianna Tucci [20'+10']

Presenter affiliation: Queen Mary University of London, London, United Kingdom; UCL Queen Square Institute of Neurology, UCL, London, United Kingdom.

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Mosaic H3K9me3 at BREACHes predicts synaptic gene expression associated with fragile X syndrome cognitive severity

Kenneth Pham, Thomas Malachowski, Chuanbin Su, Linda Zhou, Ji Hun Kim, Jennifer E. Phillips-Cremins [10'+5']

Presenter affiliation: University of Pennsylvania, Philadelphia, Pennsylvania.

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Recurrent patterns of widespread neuronal genomic damage shared by major neurodegenerative disorders

Zinan Zhou, Lovelace J. Luquette, Guanlan Dong, Junho Kim, Jayoung Ku, Kisong Kim, Mingyun Bae, Diane D. Shao, Bezawit Sahile, Michael B. Miller, August Y. Huang, William J. Nathan, Andre Nussenzweig, Peter J. Park, Clotilde Lagier-Tourenne, Eunjung A. Lee, Christopher A. Walsh [10'+5']

Presenter affiliation: Boston Children's Hospital, Boston, Massachusetts; Harvard Medical School, Boston, Massachusetts. 60

C9orf72 RAN-translated proteins and quality control of membrane-less organelles

Zheng Ying [10'+5']

Presenter affiliation: Soochow University College of Pharmaceutical Sciences, Suzhou, China. 61

THURSDAY, November 6—5:00 PM

COCKTAILS and BANQUET

FRIDAY, November 7—9:00 AM

SESSION 8 THERAPEUTIC DEVELOPMENT FOR REPEAT EXPANSION DISORDERS

Chairperson: **Ryan Yuen**, The Hospital for Sick Children, Toronto, Canada

Targeted delivery for HD therapies

Beverly L. Davidson [20'+10']

Presenter affiliation: Children's Hospital of Philadelphia, Philadelphia, Pennsylvania; Perelman School of Medicine at the University of Pennsylvania, Philadelphia, Pennsylvania. 62

Investigation of HD pathogenesis using large animal models

Sen Yan, Su Yang, Peng Yin, Shihua Li, Xiao-Jiang Li [20'+10']

Presenter affiliation: Jinan University, Guangzhou, China. 63

Establishing sensory neurons as therapeutic targets in peripheral neuropathy driven by polyglutamine expanded ATXN3

Juan P. Mato, John Hayes, Maeha Sridharan, Aarsal Naeem, Toluwani Adelaja, Jacen Emerson, Alexandra F. Putka, Eva L. Feldman, Hayley S. McLoughlin [20'+10']

Presenter affiliation: University of Michigan, Ann Arbor, Michigan.

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Break

Treating myotonic dystrophy with artificial RNA endonucleases to specifically degrade toxic RNA expansions

Tong Wei , Hanyang Hu , Xin Jiang , Yuchen Li, Wenjian Han, Yi Yang, Xiao Xiao, Miaowei Mao, Zefeng Wang [10'+5']

Presenter affiliation: Chinese Academy of Sciences, Shanghai, China.

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Exosome as a carrier for delivery of mini-intrabody in treating HD

Tanqi Yang, Huichun Tong, Xiao-Jiang Li, Shihua Li [10'+5']

Presenter affiliation: Jinan University, Guangzhou, China.

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uN2CpolyG-DDX6 sequestration hijacking P-body in neuronal intranuclear inclusion disease

Ying Xiong, Kaiyan Jiang, Yuting He, Haixia Tang, Daojun Hong [10'+5']

Presenter affiliation: Nanchang University, Nanchang, China.

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