

Abstracts of papers presented
at the 2024 Cold Spring Harbor Asia Conference

AUTISM & NEURODEVELOPMENT DISORDERS: FROM GENETIC DISCOVERIES TO INTERVENTIONS

June 17–June 21, 2024

Arranged by

Eunjoon Kim, *Korea Advanced Institute of Science and Technology*
Zilong Qiu, *Shanghai Jiao Tong University*
Jonathan Sebat, *University of California, San Diego*
Toru Takumi, *Kobe University School of Medicine*
Xiang Yu, *Peking University*



Cold Spring Harbor Conferences Asia
Cold Spring Harbor Laboratory



**AUTISM & NEURODEVELOPMENT DISORDERS:
FROM GENETIC DISCOVERIES TO INTERVENTIONS**

Monday, June 17 – Friday, Jun 21, 2024

Monday	7:00 pm	Keynote Speaker 1 Developmental and Circuit Mechanisms I
Tuesday	9:00 am	2 Autism Genetics
Tuesday	2:00 pm	Poster Session
Tuesday	3:00 pm	<i>Chinese Tea and Beer Tasting</i>
Tuesday	7:00 pm	3 Molecular, Cellular, Synaptic Mechanisms I
Wednesday	9:00 am	4 Molecular, Cellular, Synaptic Mechanisms II
Wednesday	2:00 pm	<i>Visit to Old Suzhou*</i>
Wednesday	7:00 pm	5 Clinical / Translational Studies I
Thursday	9:00 am	6 Developmental and Circuit Mechanisms II
Thursday	2:00 pm	7 Clinical / Translational Studies II
Thursday	6:00 pm	Cocktails and Banquet
Friday	9:00 am	8 New Models and Translational Studies

Oral presentation sessions are located in the CSHA Auditorium
Poster session and Chinese Tea & Beer Tasting are 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 12:00am - 1:30pm

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

Banquet: Suz Garden 7:00pm

More information will be available at CSHA office.

(Map at the end of this abstract book)

PROGRAM

MONDAY, June 17—7:00 PM

SESSION 1 DEVELOPMENTAL AND CIRCUIT MECHANISMS I

Chairperson: **Zilong Qiu**, Shanghai Jiao Tong University School of Medicine, Shanghai, China

KEYNOTE SPEAKER

Advancements in neurodevelopmental disease models using genetically-modified marmosets

Hideyuki Okano [35'+10']

Presenter affiliation: Keio University, Kawasaki, Japan; RIKEN, Wako, Japan.

1

Break

Oxytocin and autism—Insights from PVH oxytocin neuron inputs and outputs

Xiang Yu [20'+5']

Presenter affiliation: Peking University, Beijing, China.

2

Abnormal theta oscillation induces social deficits via attenuating synaptic density

Shuai-Shuai Wang, Ying-Mei Lu [10'+5']

Presenter affiliation: Nanjing Medical University, Nanjing, China.

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TUESDAY, June 18—9:00 AM

SESSION 2 AUTISM GENETICS

Chairperson: **Jonathan Sebat**, University of California, San Diego, La Jolla, California, USA

Dissecting the genetic, biological and phenotypic heterogeneity of the autism spectrum

Jonathan Sebat [20'+5']

Presenter affiliation: University of California, San Diego, La Jolla, California.

Genetics of autism spectrum disorder in Chinese population

Kun Xia [20'+5']

Presenter affiliation: School of Life Sciences, Central South University
Changsha, Hunan, China; Hengyang Medical School, University of
South China, Hengyang, Hunan, China.

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**Abnormal cortical dynamics during behavioral transition in
15q11-13 CNV mice**

Nobuhiro Nakai [10'+5']

Presenter affiliation: Kobe University, Kobe, Japan.

5

**Integrative analyses of de novo mutations in 62,661 trios with
autism and developmental delay**

Shilin Gao, Xiaoli Rao, Yue Xu, Panhui Tian, Chaoyi Shan, Jiaqi Li,
Rong Zhang, Tianyun Wang [10'+5']

Presenter affiliation: Neuroscience Research Institute, School of Basic
Medical Sciences, Peking University, Beijing, China.

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Break

**Deciphering the genetic architecture of psychiatric disorders
leveraging the diversity of the human genome**

Hailiang Huang [20'+5']

Presenter affiliation: Broad Institute of MIT and Harvard, Cambridge,
Massachusetts; Massachusetts General Hospital, Boston,
Massachusetts; Harvard Medical School, Boston, Massachusetts.

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The polygenic architecture of autism

Jing Zhang, Jakob Grove, Caitlin E. Carey, Jack M. Fu, F K.

Satterstrom, Susan S. Kuo, Ajay Nadig, Swapnil Awasthi, Kaitlin E.

Samocha, Anders D. Børglum, Elise B. Robinson [20'+5']

Presenter affiliation: Broad Institute of MIT and Harvard, Cambridge,
Massachusetts; Massachusetts General Hospital, Boston,
Massachusetts.

8

**CRISPR screens in assembloids reveal autism spectrum disorder
genes that interfere with human interneuron development**

Xiangling Meng, David Yao, Kent Imaizumi, Xiaoyu Chen, Kevin
Kelley, Noah Reis, Mayuri Thete, Michael Bassik, Sergiu Pasca
[10'+5']

Presenter affiliation: Stanford University, Stanford, California.

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Protein phosphatase-1 and 2A in health and neurodevelopmental disorders

Houhui Xia, Karl Foley, Abigail Mayer, Cody McKee, Angus Nairn, Stefan Strack, Veerle Janssens [10'+5']

Presenter affiliation: University of Rochester, Rochester, New York.

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TUESDAY, June 18—2:00 PM

POSTER SESSION

Low-dose IL-2 improves neuronal mitochondrial function, reduces oxidative stress, and ameliorates autism symptoms

Penghao Yao, Xia Cao

Presenter affiliation: The Second Affiliated Hospital of Kunming Medical University, Kunming, China.

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Low-dose IL-2 therapy reverses Teff/Treg imbalance and improves core symptoms in BTBR mice

Meng Li, Xia Cao

Presenter affiliation: The Second Affiliated Hospital of Kunming Medical University, Kunming, China.

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Association of the retinol to all-trans retinoic acid pathway with autism spectrum disorder

Yu-Ru Feng, Qian Zhang, Jing-Kun Miao, Ting Yang, Jie Chen, Hong-Yu Chen, Qiu-Hong Mou, Xue-Li Xiang, Dan Long, Qiu-Hong Wei, Yuan Wu, Ting-Yu Li

Presenter affiliation: Children's Hospital of Chongqing Medical University, Chongqing, China.

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Disruption of developmental spontaneous neuronal activity mimics autism spectrum disorder model in *Drosophila*

Sarani Dey, Sharba Bandyopadhyay, Abhijit Das

Presenter affiliation: Indian Institute of Technology Kharagpur, Kharagpur, India.

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Scrap haploinsufficiency induced autistic-like behaviors in mice through disruption of *Satb2* expression

Chaodong Ding, Wei Zhou, Yuhan Shi, Shifang Shan, Yiting Yuan, Yuefang Zhang, Fei Li, Zilong Qiu

Presenter affiliation: Songjiang Research Institute, Shanghai, China; Institute of Neuroscience, Shanghai, China.

15

- Effective *in vivo* RNA base editing via engineered cytidine deaminase APOBECs fused with PUF proteins**
Wenjian Han, Bo Yuan, Xiaojuan Fan, Weike Li, Zefeng Wang, Zilong Qiu
 Presenter affiliation: Shanghai Jiao Tong University School of Medicine, Shanghai, China. 16
- Alteration of parvalbumin (PV) expression and activity of cortical PV neurons by azadiradione**
Sudipta Jana, Bhaskarjyoti Giri, Sagarika Das, Sharba Bandyopadhyay, Nihar R. Jana
 Presenter affiliation: IIT Kharagpur, Kharagpur, India. 17
- Early postnatal blockade of GABAergic receptor induces autism-like behavioral phenotypes in mice**
Se Jin Jeon, Chan Young Shin
 Presenter affiliation: Hallym University, Chuncheon, South Korea. 18
- CSDE1 mediates neurogenesis via post-transcriptional regulation of cell cycle**
Xiangbin Jia, Kun Xia, Hui Guo
 Presenter affiliation: Central South University, Changsha, China. 19
- Deletions of Cacna2d3 in parvalbumin expressing neurons leads to autistic-like phenotypes in mice**
 Wei Shao, Hang Zheng, Liang Jing, Kai Wang, Xiao Jiang
 Presenter affiliation: Anhui Medical University, Hefei, China. 20
- Postnatal age-differential ASD-related transcriptomic, synaptic, and behavioral deficits in *Myt1l*-mutant mice**
Seongbin Kim, Jae Jin Shin, Yeji Yang, Hyoseon Oh, Hyojin Kang, Jin Young Kim, Eunjoon Kim
 Presenter affiliation: Korea Advanced Institute for Science and Technology, Daejeon, South Korea. 21
- Sexually dimorphic behavior, neuronal activity and gene expression in *Ash1l* mutant mice**
Jisoo Kim, Seoyeong Kim, Seungjoon Lee, Yunho Lee, Yeji Yang, Muwon Kang, Soowon Lee, Young Woo Noh, Hyun Kim, Seong-Gi Kim, Hyojin Kang, Jin Young Kim, Eunjoon Kim
 Presenter affiliation: Korea Advanced Institute of Science and Technology (KAIST), Daejeon, South Korea. 22

Gene dosage-differential sexual dimorphism in *CHD8*-mutant mice

Jinkyong Kim, Seungjoon Lee, Eunkyoo Hwang, Hwajin Jung, Sooyeon Lee, Seongbin Kim, Heera Moon, Jisoo Kim, Gina Lee, Chanhee Lee, Sanghan Choi, Hyojin Kang, Seong-gi Kim, Eunee Lee, Eunjoon Kim

Presenter affiliation: Korea Advanced Institute for Science and Technology (KAIST), Daejeon, South Korea.

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Language subtypes in young autistic children and the influence of parental education, educational environment and diet

Qin Li, Weihua Zhao, Rong Zhang, Keith M. Kendrick

Presenter affiliation: Chengdu University of Traditional Chinese Medicine, Chengdu, China.

24

An EED/PRC2-H19 loop regulates cerebellar development

Pei-Pei Liu, Xiao Li, Lin-Fei Jiao, Zhao-Qian Teng, Yun-Gui Yang, Chang-Mei Liu

Presenter affiliation: Institute of Zoology, Chinese Academy of Sciences, Beijing, China.

25

Autism-like behaviors and impaired spine pruning in a *Pten* haploinsufficiency mouse model

Laixin Liu, Yuanyuan Zhang, Jiaqi Shen, Xiang Yu

Presenter affiliation: Peking University, Beijing, China; Peking University, Academy for Advanced Interdisciplinary Studies, China.

26

Reduced cortical connectivity and multisensory integration in the *Shank3*^{A14-16} mice

Seung-Mi Oh, You-Hyang Song, Hae-Yong Park, Jun-Ho Song, Jae-Hyun Kim, Eunjoon Kim, Se-Bum Paik, Seung-Hee Lee

Presenter affiliation: KAIST, Daejeon, South Korea; Institute for Basic Science, Daejeon, South Korea.

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Neural mechanisms and intervention of impaired somatosensory processing in autism-associated *Shank3* mutant dogs

Qi Shi, Liang Wu, Baolong Ren, Kun Guo, Yong-hui Jiang, Yong Q. Zhang, Li Hu

Presenter affiliation: Institute of Genetics and Developmental Biology, Chinese Academy of Sciences, Beijing, China.

28

Microbial engineering paves new ways in neurodevelopment therapy

Jing Wang, Weijun Feng, Bing Han

Presenter affiliation: Children's Hospital of Fudan University, Shanghai, China.

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Ash1l dysfunction in microglia induces autistic-like social behavior defects

Yuze Yan, Gang Zhou, Cheng Zhang, Hong Xie, Ji-Song Guan

Presenter affiliation: ShanghaiTech University, Shanghai, China.

30

Dysfunction of the primary visual cortex leads to visual impairments in a Grin2b+/C456Y mouse model of ASD

Jong-Yeon Yoo, Ilsong Choi, Eunjoon Kim, Seung-Hee Lee

Presenter affiliation: Korea Advanced Institute of Science and Technology (KAIST), Daejeon, South Korea; Institute for Basic Science (IBS), Daejeon, South Korea.

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TUESDAY, June 18—3:00 PM

Chinese Tea and Beer Tasting

TUESDAY, June 18—7:00 PM

SESSION 3 MOLECULAR, CELLULAR, SYNAPTIC MECHANISMS I

Chairperson: **Eunjoon Kim**, Korea Advanced Institute of Science and Technology, Daejeon, Korea

A novel transcription regulatory mechanism underlying brain development

Irina Voineagu [20'+5']

Presenter affiliation: University of New South Wales, Sydney, Australia.

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NMDAR dysfunction in mouse models of autism spectrum disorders

Eunjoon Kim [20'+5']

Presenter affiliation: IBS, Daejeon, South Korea; KAIST, Daejeon, South Korea.

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Turnover and replacement—From microglia with love

Bo Peng [10'+5']

Presenter affiliation: Fudan University, Shanghai, China.

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Reversal of neurological defects in a mouse model of microcephaly-capillary malformation syndrome

Meixin Hu, Jun Li, Jingxin Deng, Chunxue Liu, Yingying Liu, Huiping Li, Weijun Feng, Xiu Xu [10'+5']

Presenter affiliation: Children's Hospital of Fudan University, Shanghai, China.

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Break

Ash1l-expressing microglia gates social behavior

Yuze Yan, Gang Zhou, Cheng Zhang, Hong Xie, Ji-Song Guan [20'+5']

Presenter affiliation: ShanghaiTech University, Shanghai, China.

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Automated analysis of rodent behavior using IntelliProfiler—A potential tool for studying neurodevelopmental disorder-like model mice

Shohei Ochi, Sander van der Graaf, Hitoshi Inada, Noriko Osumi [10'+5']

Presenter affiliation: Tohoku University Graduate School of Medicine, Sendai, Japan.

37

MSL1 is involved in intellectual disability by facilitating acetylation of H4K16 through enriching histone H4 and acetyl-coenzyme A with liquid-liquid phase separation

Kan Yang, Zilong Qiu, Fei Li [10'+5']

Presenter affiliation: Shanghai Jiao Tong University School of Medicine, Shanghai, China.

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SESSION 4 MOLECULAR, CELLULAR, SYNAPTIC MECHANISMS II

Chairperson: **Zilong Qiu**, Shanghai Jiao Tong University School of Medicine, Shanghai, China

Neuronal gene regulation by alternative mRNA isoforms

Yalan Yang, Runwei Yang, Xiaochang Zhang [20'+5']

Presenter affiliation: The University of Chicago, Chicago, Illinois. 39

Organoid model of neonatal brain hypoxic injury uncovers circadian regulation of synapse engulfment in astrocytes

Li Li, Jong Bin Choi, Chi Hong Shin, Anna Voss, Erin M. Gibson, Julia A. Kaltschmidt, Steven A. Sloan, Won-Suk Chung, Anca M. Pasca [20'+5']

Presenter affiliation: Stanford University, Palo Alto, California. 40

Prefrontal-thalamic information processing during social interaction

Yang Zhan, Bailu Si, Xinnian Wang, Surui Xu [10'+5']

Presenter affiliation: Shenzhen Institute of Advanced Technology, CAS, Shenzhen, China. 41

Proteomic study of cell signaling in the primary cilium in brain development and diseases

Xiaoliang Liu, Eva Cai, Jingyi Zhang, Oscar Torres Gutierrez, Xuecai Ge [10'+5']

Presenter affiliation: University of California, Merced, Merced, California. 42

Break

Effectiveness of bumetanide treatment in children aged 3 to 6 years with autism spectrum disorder

Fei Li [20'+5']

Presenter affiliation: Xinhua Hospital, Shanghai Jiao Tong University School of Medicine, Shanghai, China. 43

Inhibiting proBDNF to mature BDNF conversion leads to ASD-like phenotypes in vivo

Feng Yang, He You, Toshiyuki Mizui, Yasuyuki Ishikawa, Keizo Takao, Tsuyoshi Miyakawa, Xiaofei Li, Ting Bai, Kun Xia, Lingling Zhang, Dizhou Pang, Yiran Xu, Changlian Zhu, Masami Kojima, Bai Lu [10'+5']

Presenter affiliation: Beijing Tiantan Hospital, Beijing, China.

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Neurodevelopmental abnormalities of human 15q11-13 chromosome duplication autism mouse model and potential intervention strategies

Chuan Xing, Tian-Jia Song, Yu-Yang Guo, Yi-Xuan Zhang, Pei-Yuan Jia, Dan Zhang, Han Zhang, Xiang Yu [10'+5']

Presenter affiliation: Peking University, Beijing, China.

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WEDNESDAY, June 19—2:00 PM

Visit to Old Suzhou

WEDNESDAY, June 19—7:00 PM

SESSION 5 CLINICAL / TRANSLATIONAL STUDIES I

Chairperson: Xiang Yu, Peking University, Beijing, China

Translational research approach to social orienting deficits in autism—The role of superior colliculus-ventral tegmental pathway

Camilla Bellone [20'+5']

Presenter affiliation: University of Geneva, Geneva, Switzerland.

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Hypo-arousal or hyperarousal? The mechanisms underlying social information processing in autism

Li Yi, Qiandong Wang, Ci Song, Zhuo R. Han [20'+5']

Presenter affiliation: Peking University, Beijing, China.

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Break

Discovering translational biomarkers in neurodevelopmental conditions—EEG profile in mouse models of idiopathic and syndromic autism

Ning Cheng [10'+5']

Presenter affiliation: University of Calgary, Calgary, Canada.

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Distinguishing autistic from typical developing children using multiple eye-tracking tasks

Dan Xu, Linghong Huang, Xiangqin Wang, Yanan Qing, Menghan Zhou, Lei Liu, Xiaojiao Yang, Weihua Zhao, Qin Li, Jiao Le, Keith M. Kendrick [10'+5']

Presenter affiliation: University of Electronic Science and Technology of China, Chengdu, China.

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Developing oxytocin as a therapeutic intervention for autism spectrum disorder

Keith M. Kendrick [20'+5']

Presenter affiliation: University of Electronic Science and Technology of China, Chengdu, China.

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Clinical phenotype and genetic characteristics of patients with SHANK3 gene variants

Chunxue Liu, Jingxin Deng, Yi Wang, Xiu Xu [10'+5']

Presenter affiliation: Children's Hospital of Fudan University, Shanghai, China.

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THURSDAY, June 20—9:00 AM

SESSION 6 DEVELOPMENTAL AND CIRCUIT MECHANISMS II

Chairperson: Eunjoon Kim, Korea Advanced Institute of Science and Technology, Daejeon, Korea

PQBP1 regulates social novelty through modulating glutamate signaling in nucleus accumbens medial shell D1-MSNs

Song Li, Zi-Chao Zhang, Junhai Han [20'+5']

Presenter affiliation: Southeast University, Nanjing, China.

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Neural circuit mechanisms of empathy impairments in autism spectrum disorders

Boxing Li [20'+5']

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

53

Dentate gyrus morphogenesis is regulated by an autism risk gene trio function in granule cells

Mengwen Sun, Xiaoxuan Sun, Dai Zhang, Jun Li [10'+5']

Presenter affiliation: Peking University Sixth Hospital, Peking University Institute of Mental Health, Beijing, China; Peking University, Beijing, China.

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Altered auditory cortical representation of context specific vocalization sequences in a mouse model of autism spectrum disorders

Srishti Jain, Purba Mandal, Swapna Agarwalla, Sharba Bandyopadhyay [10'+5']

Presenter affiliation: IIT Kharagpur, Kharagpur, India.

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Break

Sensory and multisensory deficits in mouse models of autism

Seung-Hee Lee [20'+5']

Presenter affiliation: KAIST, Daejeon, South Korea; IBS, Daejeon, South Korea.

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Basal forebrain circuitry for social interaction behavior

Han Xu [20'+5']

Presenter affiliation: Zhejiang University, Hangzhou, China.

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Spontaneous neuronal activity induced transcriptional regulation mediated by Mef2 in the origin of autism spectrum disorder

Sarani Dey, Sharba Bandyopadhyay, Abhijit Das [10'+5']

Presenter affiliation: Indian Institute of Technology Kharagpur, Kharagpur, India.

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Cntnap3 mutation impairs claustral function leading to abnormal social behavior

Zimeng Li, Zhuolei Jiao, Kan Yang, Zhifang Chen, Xiaohong Xu, Zilong Qiu [10'+5']

Presenter affiliation: Shanghai Jiaotong University School of Medicine, Shanghai, China; Chinese Academy of Sciences, Shanghai, China.

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THURSDAY, June 20—2:00 PM

SESSION 7 CLINICAL / TRANSLATIONAL STUDIES II

Chairperson: **Toru Takumi**, Kobe University School of Medicine, Kobe, Japan

Modeling autism

Toru Takumi [20'+5']

Presenter affiliation: Kobe University School of Medicine, Kobe, Japan.

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Connectopathy in the frontotemporal network of a schizophrenia rat model—Implications for auditory hallucinations

Hiroyuki Nawa, Manavu Tohmi, Hisaaki Namba [20'+5']

Presenter affiliation: School of Pharmaceutical Sciences, Wakayama Medical University, Wakayama, Japan.

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Clinically integrated research platforms for transformational science in neurodevelopment

Adam J. Guastella [20'+5']

Presenter affiliation: University of Sydney, Sydney, Australia.

62

Shank3 loss-of-function combined with abnormal nurturing environment led to active-but-odd social deficits-involvement of oxytocin system

Ming J. Li, Yu F. Wu, Xue W. Wang, Xing Y. Lan, Yong Zhang, Rong Zhang [10'+5']

Presenter affiliation: Peking University, Beijing, China; Ministry of Education/National Health Commission, Beijing, China; Peking University Health Science Center, Beijing, China.

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Application of nanotechnology for brain vascular targeting drug delivery

Jiukuan Hao [10'+5']

Presenter affiliation: University of Houston, Houston, Texas.

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Facial characteristics description and classification based on 3D images of Fragile X syndrome in a retrospective cohort of young Chinese male

Jieyi Chen, Siyuan Du, Xinran Dong, Wenhao Zhou, Qiong Xu [10'+5']

Presenter affiliation: Children's Hospital of Fudan University, Shanghai, China.

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THURSDAY, June 20—6:00 PM

COCKTAILS and BANQUET

SESSION 8 NEW MODELS AND TRANSLATIONAL STUDIES

Chairperson: **Xiang Yu**, Peking University, Beijing, China

Love, death, and oxytocin—The challenges of mouse maternal care

Robert C. Froemke [20'+5']

Presenter affiliation: NYU Grossman School of Medicine, New York, New York.

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Face processing impairment in autism-associated *Shank3* mutant dogs

Yong Q. Zhang [20'+5']

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

67

Anterior cingulate cortex-related functional hyperconnectivity underlies sensory hypersensitivity in *Grin2b*-mutant mice

Soowon Lee, Won Beom Jung, Heera Moon, Geun Ho Im, Young Woo Noh, Wangyong Shin, Yong Gyu Kim, Jee Hyun Lee, Seok Jun Hong, Yongwhan Jung, Sunjoo Ahn, Seong-Gi Kim, Eunjoon Kim [10'+5']

Presenter affiliation: Korea Advanced Institute of Science and Technology, Biological Sciences, South Korea.

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Break

Whole-brain *in vivo* base editing reverses autistic-like behavioral in *Mef2c* mutant mice

Wei-Ke Li, Shu-Qian Zhang, Tian-Lin Cheng, Zi-Long Qiu [20'+5']

Presenter affiliation: Shanghai Jiao Tong University School of Medicine, Shanghai, China.

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Atypical face processing in autism-associated *Shank3* mutant dogs

Siqi Yuan, Chenyu Pang, Liang Wu, Li Yi, Kun Guo, Yong Q Zhang, Shihui Han [10'+5']

Presenter affiliation: Chinese Academy of Sciences, Beijing, China; University of the Chinese Academy of Sciences, Beijing, China.

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Deciphering the molecular mechanisms of *MIR137* dysregulation in autism spectrum disorder

Ying Cheng [10'+5']

Presenter affiliation: Yunnan University, Kunming, China.

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