

**Day 2: TUESDAY, SEPT 17 2019 (CONTINUED)**

2:45–3 p.m. **Spina Bifida Sequencing Consortium to Identify the Impact of De Novo Mutations**—Sangmoon Lee

3–3:30 p.m. **Break**

**» METHODS FOR SHARING AND ANALYZING NEURAL TUBE DEFECT DATA**

3:30–3:45 p.m. **Novel Methodology to Analyze Gene-Gene Interactions in Neural Tube Defect Pathogenesis**—Kayla T.B. Fuselier

3:45–4 p.m. **Data Sharing and Analysis in the Neural Tube Defects International Community**—Vanessa Aguiar-Pulido

4–4:45 p.m. **Title TBA**—Betsy Ross

7–9 p.m. **Conference Dinner at Harvard Faculty Club**

**Day 3: WEDNESDAY, SEPT 18 2019**

7:45–8:15 a.m. **Breakfast**

**» EPIGENETICS OF NEURAL TUBE DEFECTS I**

8:15–8:30 a.m. **Investigating the Effect of Maternal Folic Acid Intake on Transcriptome and Methylome in a Neural Tube Defect Mouse Model**—Miho Ishida

8:30–8:45 a.m. **Folate Deficiency in Grandparents and Father Increases De Novo Mutations and Birth Defects Rates in C57BL/6 Mouse**—Yufang Zhen

8:45–9 a.m. **Role of Maternal Antioxidant Depletion-Induced Oxidative Stress in Embryo Malformation from Pregestational Obesity**—Mary Loeken

9–9:15 a.m. **Influence of Folic Acid Supplementation on Cilia Function in Neural Tube Closure**—Lee Niswander

9:15–9:45 a.m. **Break**

**» EPIGENETICS OF NEURAL TUBE DEFECTS II**

9:45–10 a.m. **MiR-324-5p Hypomethylation Induced by Folate Deficiency Affects Shh Pathway via Targeting Gli1 and Smo in Spina Bifida**—Li Wang

10–10:15 a.m. **The Gastrulating Embryo in its Environment: Expression of Nutrient Transporters**—Claudia Kapp

10:15–10:30 a.m. **Disruption of p53 Ubiquitination Mediates Neural Tube Defects Caused by Metal Micronutrient Insufficiency**—Huili Li

10:30–10:45 a.m. **The Effect of Folic Acid Deficiency on FGF Pathway via Brachyury Regulation in Neural Tube Defects**—Shaoyan Chang

10:45–11:15 a.m. **Break**

**» FRONTIERS IN PREVENTION OF NEURAL TUBE DEFECTS**

11:15–11:30 a.m. **Spina Bifida F and Anencephaly F: Need for Novel Fortification Vehicles to Address the Global Epidemic**—Godfrey P. Oakley Jr.

11:30–11:45 a.m. **Global Prevention of Neural Tube Defects with Folic Acid-Fortified Salt**—Vijaya Kancherla

11:45–12:15 p.m. **Prevention of Neural Tube Defects by Inositol**—Nicholas D.E. Greene

12:15 p.m. **Conference Close, Presentation of Awards**

12:15–2:30 p.m. **Lunch, Conference Room Available for Continued Discussions**

# 2019 International Neural Tube Defects Conference

Monday, September 16–Wednesday, September 18



**Boston Children's Hospital**  
Until every child is well™



**HARVARD MEDICAL SCHOOL**  
TEACHING HOSPITAL

## Day 1: MONDAY, SEPT 16 2019

7:45–8:15 a.m. **Breakfast**  
8:15–8:30 a.m. **Welcome**—Maitreyi Mazumdar

### » MECHANICS OF NEURAL TUBE CLOSURE

8:30–9 a.m. **Diversity Among NTDs and a Mechanism of Neural Tube Closure**  
—Andrew Copp  
9–9:15 a.m. **Human CLDN Variants Affect Different Phases of Neural Tube Closure: Functional Analysis in Chick Embryos**—Aimee K. Ryan  
9:15–9:30 a.m. **New Imaging Modalities Reveal Tissue Mechanics Changes During Cranial Neural Tube Closure**—Jitao Zhang  
9:30–9:45 a.m. **Hallmark of Primary Neurulation Observed in Zebrafish: Implications for Screening Genetic Variants Implicated in Cranial Neural Tube Defects**  
—Rachel Brewster  
9:45–10 a.m. **Questions**  
10–10:30 a.m. **Break**

### » NOVEL METHODS AND MODELS FOR UNDERSTANDING CLOSURE

10:30–11 a.m. **TBA**—Olivier Pourquié  
11–11:15 a.m. **Characterizing the Contribution of Individual Neuro-Mesodermal Progenitor Cells to the Vertebrate Body Axis**—Charlene Guillo  
11:15–11:30 a.m. **Semi-Cloned Mouse: A Powerful and Efficient Tool for NTD Research**—Lei Lu  
11:30–11:45 a.m. **The Role of Claudin-Dependent Glycoproteins in Neural Tube Closure**  
—Elizabeth-Ann Legere  
11:45–noon **Concerted Proteostatic and Metabolic Shifts in Early Forebrain During Neural Tube Closure Alter the Cerebrospinal Fluid Proteome and Depend on cMYC Downregulation for Mitochondrial Maturation and Ribosome Biogenesis**—Ryann M. Fame  
Noon–12:15 a.m. **A Plausible Mechanism for Neural Tube Defects Due to Dolutegravir Exposure**—Robert M. Cabrera  
12:15–2:15 p.m. **Poster Session and Lunch**

### » CELL BIOLOGY OF NEURAL TUBE CLOSURE I

2:15–2:30 p.m. **Ciliary Compartmentalization of Adenylyl Cyclases in Neural Tube Development and Closure**—Bandarigoda Nipunika Somatilaka  
2:30–2:45 p.m. **Investigating Interactions Between Wnt Signaling and Mitochondrial One Carbon Metabolism Using Mouse Models of Neural Tube Defects**  
—John Steele  
2:45–3 p.m. **Characterization and in Vitro Differentiation of Fetal Neuroprogenitor Cells Derived From Cerebral Spinal Fluid (CSF)**—Angela PH Burgess  
3–3:15 p.m. **MicroRNA Regulation of Neural Tube Development**—Ronald Parchem  
3:15–3:30 p.m. **Formate Rescues Neural Tube Defects Caused by Mutations in Slc25a32**  
—Yunping Lei  
3:30–4 p.m. **Break**

### » CELL BIOLOGY OF NEURAL TUBE CLOSURE II

4 p.m.–4:15 p.m. **Prevention from NTDS Linked With WNT-PCP Signalling Pathway**  
—Patricia Ybot-González

4:15–4:30 p.m. **Analysis of Neural Plate Transcriptomes in Mouse Models for Folate Responsive and Non-Folate Responsive Neural Tube Defects**—Meng Wu  
4:30–4:45 p.m. **The Role of Fuz on GPR161 Mediated Regulation of Shh Signaling in the Primary Cilia During Neural Tube Development**—Sung Eun Kim  
4:45–5 p.m. **ARMC5 as Part of a Novel RPB1-Specific Ubiquitin Ligase and its Roles in the Pathogenesis of Neural Tube Defects**—Jiangping Wu

## Day 2: TUESDAY, SEPT 17 2019

7:45–8:15 a.m. **Breakfast**

### » ADVANCES IN CLINICAL CARE

8:15–8:45 a.m. **Title TBA**—Benjamin Warf  
8:30–9 a.m. **Neural Tube Defect Care in Low-Income Settings: Challenges to Innovative Strategies in Global Neurosurgical Training and Treatment**—Sylvie Odent  
9–9:15 a.m. **Title TBA**—Sudipta Mukherjee  
9:30–9:45 a.m. **Title TBA**—Carlos Estrada  
9:45–10 a.m. **The Downside of Fetal Therapy for Myelomeningocele: This is Not a Cure**—William Whitehead  
10–10:30 a.m. **Break**

### » RISK FACTORS FOR NEURAL TUBE DEFECTS

10:30–10:45 a.m. **Title TBA**—Rebecca Zash  
10:45–11 a.m. **Perinatal Mortality from Neural Tube Defects in Botswana**—Modiegi D. Diseko  
11–11:15 a.m. **Socioeconomic Risk Factors for the Neural Tube Defects: Case-Control Study in Bogotá and Cali, Colombia 2001-2018**—Maria Manuela Sierra Breton  
11:15–11:30 a.m. **Neural Tube Defects (NTD) and Exposures to Low Doses of Radiation**  
—Wladimir Wertelecki  
11:30–11:45 a.m. **Interpregnancy Change in Body Mass Index (IPC-BMI) and Spina Bifida**  
—Laura E. Mitchell  
11:45–noon **Potential Modifiers of the Association Between Diabetes and Risk for Neural Tube Defects, National Birth Defects Prevention Study, 1997-2011**  
—Sarah C. Tinker  
Noon–12:15 a.m. **A Plausible Mechanism for Neural Tube Defects Due to Dolutegravir Exposure**—Robert M. Cabrera  
12:15–2:15 p.m. **Lunch, Mentor and Trainee Discusson**

### » GENE DISCOVERY

1:15–1:45 p.m. **Title TBA**—Suzanne Leal  
1:45–2 p.m. **The Role of Structural Variation in Human Predisposition to Spina Bifida**  
—Paul Wolujewicz  
2–2:15 p.m. **Whole Exome Sequencing in a Small Cohort of Neural Tube Defects: What Have We Learned so Far?**  
—Zoha Kibar  
2:15–2:30 p.m. **Examining Exomes of 507 Myleomeningocele Subjects Reveals Potential Roles of Novel Damaging Variants**—Kit Sing Au  
2:30–2:45 p.m. **Identification of Novel Candidate Risk Genes for Myelomeningocele in Genes of the Folate and One Carbon Metabolism Network**—Paul Hillman