

8TH ANNUAL HUMAN GENOMICS SYMPOSIUM

UNIVERSITY OF CALIFORNIA DAVIS MEDICAL CENTER

NOVEMBER 19, 2021 ➤ Education Building, Lecture Hall 1222, 4610 X Street, Sacramento, CA

8:00 AM **Registration, Boxed Continental Breakfast, Online Poster Viewing**
Welcome
9:00 Katherine Rauen, MD, PhD, Division of Genomic Medicine, Department of Pediatrics, UC Davis MIND Institute, Symposium Chair

9:05 **KEYNOTE ADDRESS – FXTAS: An Older Face of the Fragile X Gene**
Paul J. Hagerman, MD, PhD – UC Davis Department of Biochemistry and Molecular Biology, UC Davis School of Medicine and MIND Institute

9:45 ➤ **PLATFORM PRESENTATIONS: Session Chair – Krishna R Singh, MD – UC Davis Department of Obstetrics and Gynecology**

1. **Split dCas9 AAV for Treatment of Huntington's Disease**
Jennifer Waldo et al
2. **Further Characterizing Regressive Autism from a Genetic Perspective**
Ariel Lee et al
3. **CRISPR-Based Epigenetic Editing for Targeted Escape from X Chromosome Inactivation (XCI) in Two Mouse Models of DCKL5 Deficiency Disorder (CDD)**
Fiona Buchanan et al
4. **Newborn Screen for Carnitine Palmitoyl Transferase 1A Deficiency in California: Rethinking Current Methodology**
Stacey Cole et al

10:45 **BREAK**

11:00 ➤ **PLATFORM PRESENTATIONS: Session Chair - Colette DeFilippo, MS, CGC – UC Davis Department of Pediatrics**

5. **Enhancers of Cancer Driver Genes are Enriched in Somatic Mutations in Cancer**
Marketa Tomkova et al
6. **BUB1B Germline Variant Predisposes to Cutaneous Melanoma and Multiple Other Primary Malignancies**
Maha Kazmi et al
7. **The Association Between Juvenile Xanthogranulomas in Neurofibromatosis Type 1 Patients and the Development of Leukemia: A Systematic Review**
Summer Meyer et al
8. **Recently Derived, Full-Length Gene Retrocopies Occur Frequently in Dogs**
Kevin Batchner et al

12:00 PM **Boxed Lunch, Online Poster Session**

1:00 **KEYNOTE ADDRESS - Targeted Treatments for Fragile X Disorders**
Randi Hagerman, MD – UC Davis Department of Pediatrics, MIND Institute

1:45 ➤ **PLATFORM PRESENTATIONS: Session Chair – Joseph Shen, MD, PhD - UC Davis Department of Pediatrics**

9. **Metabolomic Biomarkers are Associated with Area of the Pons in Fragile X Premutation Carriers at Risk for Developing FXTA**
Marwa Zafarullah et al
10. **Investigation of the Genetic Factors Contribution to the ASD Subphenotype Disproportionate Megalencephaly**
Sierra Nishizaki et al
11. **DPH5 Dysfunction: A Novel Cause of Embryonic Lethality or Profound Neurodevelopmental Disorder**
Suma Shankar et al
12. **Prediction of Neurodevelopmental Disorders Based on *de novo* Coding Variation**
Julie Chow et al

2:45 **Closing Remarks and Poster Prizes**

3:00 **Adjourn**