



8TH ANNUAL PEDIATRIC RESEARCH SYMPOSIUM  
**TRANSLATING SCIENCE  
INTO CHILD HEALTH  
BY NEXT GENERATION FACULTY**

JAN | 12-13 | 2017

RADY CHILDREN'S HOSPITAL - SAN DIEGO

**CLOSING KEYNOTE SPEAKER:**

**ALBERT ("AL") PISANO, PHD**  
DEAN, JACOBS SCHOOL OF ENGINEERING, UCSD



**8th Annual Pediatrics Research Symposium**  
*Translating Science into Child Health*  
*By Next Generation Faculty*

**WELCOME**

January 12, 2017

Dear Friends and Colleagues:

The Department of Pediatrics at the University of California, San Diego and Rady Children's Hospital is proud to present the 8<sup>th</sup> Annual Pediatrics Faculty Research Symposium. Our goal today is to bring together many of our junior and mid-level investigators from our Department to present their progress made in areas of Bio-informatics, Stem Cells and Cell Biology, Allergy and Immunology and organogenesis. Speakers represent many disciplines using a broad-spectrum of translational research and methodologies in each area. This event is also inaugurating the Educational Building at Rady Children's this January, 2017.

We welcome you and we certainly hope that this occasion leads to active discussions and fruitful collaborations.

Sincerely yours,

Symposium Organizing Committee:

Christina Chambers, PhD, MPH

Jeffrey Schwimmer, MD

Stephen Spector, MD

Gabriel Haddad, MD

# TABLE OF CONTENTS

Program Schedule.....4

Presenter Abstracts.....11

Poster Presentation

- Allergy, Immunology & Rheumatology..... 44
- Biochemical Genetics & Metabolomics Lab.....44
- Cardiology.....45
- Cardiology & CV Surgery.....45
- Cardiology/Radiology.....46
- Cellular & Molecular Medicine.....46
- Child & Adolescent Services Research Center.....47
- Dermatology.....48
- Dysmorphology & Teratology.....48
- Emergency Medicine.....49
- Endocrinology.....50
- Gastroenterology, Hepatology & Nutrition.....50
- Genomic Medicine.....51
- Hospital Medicine.....52
- Host Microbe Systems & Therapeutics.....52
- Infectious Diseases.....52
- Intensive Care Unit.....53
- Neonatology.....53
- Nephrology.....54
- Neuro-Oncology.....54
- Neurosurgery.....54
- Nursing.....55
- Orthopedics.....55
- Otolaryngology.....56
- Pharmacology & Drug Discovery.....56
- Pharmacy.....57
- Pulmonology.....57
- Surgery.....58
- Urology.....59

# PROGRAM SCHEDULE

THURSDAY, JANUARY 12, 2017

1:00 - 1:20 **Welcome & Address**  
**Gabriel Haddad, MD**  
Distinguished Professor of Pediatrics and Neurosciences  
Chairman, Department of Pediatrics,  
University of California San Diego  
Physician-In-Chief & Chief Scientific Officer,  
Rady Children’s Hospital – San Diego

**Bioinformatics and Disease**  
**Moderator: Victor Nizet, MD**

1:20 – 1:40 **“Novel Insights into the Genetic Mechanisms of Diabetes Risk”**  
**Kyle Gaulton, PhD**  
Assistant Professor of Pediatrics  
Division of Gastroenterology

1:40 – 2:00 **“Challenges in the Discovery and Interpretation of Mutations in Disease Associated Genes”**  
**Vikas Bansal, PhD**  
Assistant Professor of Pediatrics  
Division of Genome Information Sciences

2:00 – 2:20 **“Engineering Better Therapeutics with Systems Analyses and Genome Editing”**  
**Nate Lewis, PhD**  
Assistant Professor of Pediatrics  
Division of Host-Microbe & Systems Therapeutics

2:20 – 2:40 **“Computationally-driven Answers to Disease”**  
**Debashis Sahoo, PhD**  
Assistant Professor of Pediatrics  
Division of Genome Information Sciences

**2:40 – 3:10 Keynote: “Genetic (mal-)adaptation: Nature's Spotlight on Human Disease”**  
**Vineet Bafna, PhD**  
Professor of Computer Science & Engineering  
Jacobs School of Engineering

3:10 – 3:20 *Break*

**Surgical Advances**  
**Moderator: Dennis Wenger, MD**

3:20 - 3:25

**Introduction**

**Scott Mubarak, MD**

Surgeon-in-Chief  
Clinical Professor of Orthopedics  
Emeritus Chair, Division of Pediatric Orthopedics  
Rady Children's Hospital – San Diego

3:25 – 3:35

***“The Utility of D-Dimer in the Evaluation of Pediatric Head Trauma”***

**Timothy Fairbanks, MD**

Associate Clinical Professor of Surgery  
Division Chief of Pediatric Surgery  
Vice President/Medical Director of Surgery  
Children's Specialists of San Diego

3:35 – 3:45

***“Infant Hip Dysplasia Outcomes in a Multi-Center Study”***

**Salil Upasani, MD**

Assistant Clinical Professor of Surgery  
Division of Pediatric Orthopedic Surgery

3:45 – 3:55

***“A Modified Technique for Strabismus Surgery in the Presence of a Scleral Buckle”***

**Anna Kim, MD**

Division of Pediatric Ophthalmology

3:55 – 4:05

***“Leveraging Informatics for Comparative Effectiveness Research”***

**George Chiang, MD**

Associate Clinical Professor of Surgery  
Division Chief of Urology

4:05 – 4:15

***“Robotic-assisted Endoscopic Third Ventriculostomy for the Treatment of Hydrocephalus”***

**David Gonda, MD**

Assistant Clinical Professor of Surgery  
Division of Pediatric Neurosurgery

4:15 – 4:25

***“Quality of Life Tool for Children with Facial Deformities”***

**Amanda Gosman, MD**

Clinical Professor of Surgery  
Division of Plastic Surgery

4:25 – 4:35

***“Laryngeal Stenosis in Children: Epidemiology Using a Population Based Approach”***

**Matthew Brigger, MD**

Associate Professor of Clinical Surgery  
Division of Otolaryngology

4:35 – 4:45

***“An Instrumented Glove for Improving Spasticity Assessment”***

**Andrew Skalsky, MD**

Associate Clinical Professor of Orthopaedic Surgery  
Dept of Orthopaedic Surgery  
Division of Pediatric Rehab Medicine

4:45 – 4:55

***“Prenatally Diagnosed Vasa Previa: A Single Institution Series of 96 Cases”***

**Val Catanzarite, MD, PhD.**

Chief of Perinatology/High Risk OB

**4:55 – 5:15**

***“How 3D Imaging of Scoliosis Changes What We Think We Know”***

**Keynote:**

**Peter Newton, MD**

Chief of Orthopedic Surgery & Scoliosis

5:15

***Closing Remarks***

**Donald Kearns, MD**

President and CEO  
Rady Children's Hospital – San Diego

## FRIDAY, JANUARY 13, 2017

- 8:30 - 8:50 **Welcome & Address**  
**Gabriel Haddad, MD**  
Distinguished Professor of Pediatrics and Neurosciences  
Chair, Department of Pediatrics,  
University of California San Diego  
Physician-In-Chief & Chief Scientific Officer,  
Rady Children's Hospital – San Diego
- Injury/Organogenesis**  
**Moderator: Christina Chambers, PhD**
- 8:50 – 9:10 **“Insights into Neonatal Hypoxic Injury from Animal Models of Human Disease”**  
**Farhad Imam, MD, PhD**  
Assistant Professor of Pediatrics  
Division of Neonatology
- 9:10 – 9:30 **“Novel Role of Macrophages in the Pathogenesis of Type 1 Diabetes”**  
**Wenxian Fu, PhD**  
Assistant Professor of Pediatrics  
Division of Endocrinology
- 9:30 – 9:50 **“Identification of Factors that Regulate Kidney Cell Fate: Hand2 Inhibits Intermediate Mesoderm Development”**  
**Elliott Perens, MD, PhD**  
Assistant Professor of Pediatrics  
Division of Nephrology
- 9:50 – 10:10 **“Genome-wide Assessment Reveals Marked Differences in the Transcriptome and Epigenome of Mononuclear Phagocytes in the Lung”**  
**Eniko Sajti, PhD**  
Assistant Clinical Professor of Pediatrics  
Division of Neonatology
- 10:15–10:45** **“Nuclear Receptors, Oxygen & the First Breath”**  
**Keynote: Ron Evans, PhD**  
Professor of Biology, Biomedical Sciences, and Neuroscience  
Director, Gene Expression Laboratory  
Howard Hughes Medical Institute Investigator  
March of Dimes Chair in Molecular and Developmental Biology  
Salk Institute for Biological Studies

10:45-11:05 **Break**

### **Infectious Diseases/Allergy/Immunology** **Moderator: Stephen Spector, MD**

- 11:05 – 11:25 **“Time to Unwind: A Novel B Cell Immunodeficiency”**  
**Lori Broderick, MD, PhD**  
Assistant Professor of Pediatrics  
Division of Allergy, Immunology & Rheumatology
- 11:25 – 11:45 **“New Approaches to the Evaluation of Neonatal Neutrophils and Sepsis Risk”**  
**Shelley Lawrence, MD, MS**  
Assistant Clinical Professor of Pediatrics  
Division of Neonatology
- 11:45 – 12:05 **“Eosinophilic Esophagitis – A Diagnosis That’s Hard to Swallow: Mechanisms and Clinical Impacts of Tissue Remodeling”**  
**Seema Aceves, MD, PhD**  
Associate Professor of Clinical Pediatrics  
Division of Allergy, Immunology & Rheumatology
- 12:05 – 12:25 **“The Emerging Story of Atorvastatin for Treatment of Acute Kawasaki Disease”**  
**Adriana Tremoulet, MD**  
Associate Professor of Clinical Pediatrics  
Division of Host Microbe Systems & Therapeutics
- 12:30-1:00** **“Hygiene Hypothesis to Early Human Microbiome: Understanding Asthma Origins”**  
**Keynote: Fernando Martinez, MD**  
Regents’ Professor of Pediatrics  
Division of Pulmonology/Allergy and Immunology  
Director, BIO5 Institute  
Director, Arizona Respiratory Center  
University of Arizona
- 1:00 – 1:30 **Lunch**
- 1:30 – 2:30** **Poster Session**



**Stem Cell/Cell Biology**  
**Moderator: Jeffrey Schwimmer, MD**

2:30 – 2:50 *“Hematopoietic Stem Cell Gene Therapy for the Lysosomal Storage Disorder Cystinosis: Clinical Translation and Application to Mitochondrial Diseases”*

**Stephanie Cherqui, PhD**

Associate Professor of Pediatrics  
Division of Genetics

2:50 – 3:10 *“Growth Factor Receptor Trafficking: Recycling Targets for Neuroblastoma Therapy”*

**Peter Zage, MD, PhD**

Associate Professor of Pediatrics  
Division of Hematology/Oncology

3:10 – 3:30 *“Accelerating Cures for Rare Diseases: Therapeutic Rescue of Niemann-Pick type C1 Stem Cell –derived Neurons Using Repurposed Drugs”*

**Paulina Ordonez, MD**

Assistant Professor of Pediatrics  
Division of Gastroenterology

3:30 – 3:50 *“Lost in Translation: Effects of a Mutant Protein in the Gut”*

**Mamata Sivagnanam, MD**

Associate Clinical Professor of Pediatrics  
Division of Gastroenterology

**3:55 – 4:25** *“Probing the Secrets of Alzheimer’s Disease with Stem Cells”*

**Keynote:**

**Larry Goldstein, PhD**

Distinguished Professor of Cellular and Molecular Medicine  
Department of Neurosciences  
Director, UC San Diego Stem Cell Program  
Scientific Director, Sanford Consortium for Regenerative Medicine  
Director, Sanford Stem Cell Clinical Center

**4:30 – 5:00** *Closing Keynote Speaker*

*“Medically-driven Engineering Innovation: The Best Days Yet to Come”*

**Albert (“Al”) Pisano**

Distinguished Professor, Mechanical & Aerospace Engineering and  
Electrical & Computer Engineering  
Walter J. Zable Endowed Chair of Engineering  
Dean, UCSD Jacobs School of Engineering

**5:00-5:15** **Award Winners Announced**

**5:15 – 6:30** **2<sup>nd</sup> Poster Session**

**JANUARY 12, 2017**  
**PRESENTER ABSTRACTS**

## KYLE GAULTON, PHD

Assistant Professor of Pediatrics  
Division of Gastroenterology



### *“Novel Insights into the Genetic Mechanisms of Diabetes Risk”*

**1:20 – 1:40**

Diabetes affects 30 million Americans and causes substantial morbidity and mortality. Genetic factors account for half of individual risk of developing diabetes, which recent studies have argued is largely due to many risk loci of small effect. As each risk locus entails a causal mechanism, translating this breadth of risk loci into their mechanisms can greatly inform on diabetes pathogenesis. The majority of diabetes risk loci map to non-coding sequence and likely affect gene regulation. In our work we aim to identify diabetes risk variants and determine how they affect gene regulatory processes using genome-wide regulatory maps from primary and stem cell-derived human tissue. By applying statistical models to integrate genetic and regulatory data, we have identified cellular systems broadly involved in diabetes risk as well as specific genomic regions and genes affected by risk variants in these systems. We are further pursuing molecular assay and cellular models of these variants, regions and genes to determine their functional and phenotypic effects. Our continued plans include collecting larger population and clinical genetic datasets, creating high-resolution regulatory maps in diabetes-relevant primary and stem cell-derived tissue across many samples, and developing and applying improved statistical models for data integration and inference. We aim finally to translate these genetic and mechanistic findings into new diabetes prevention and treatment strategies.

---

---

---

---

---

## VIKAS BANSAL, PHD

Assistant Professor of Pediatrics  
Division of Genome Information Sciences



### *“Challenges in the Discovery and Interpretation of Mutations in Disease Associated Genes”*

**1:40 – 2:00**

In spite of tremendous advancements in high-throughput sequencing technologies, the detection of pathogenic mutations in genes previously implicated in rare disorders remains challenging. Two major reasons for this are: (i) low sensitivity and specificity for detecting specific types of mutations such as insertions/deletions and (ii) inability to discriminate functional from neutral mutations using in-silico methods. In this talk, I will use monogenic diabetes as an example to illustrate these challenges, describe approaches to leverage large-scale sequence data to address some of these challenges, and highlight the complexity of genotype-phenotype relationship in genes known to be associated with rare human diseases.

---

---

---

---

---

---

---

---

## NATE LEWIS, PHD

Assistant Professor of Pediatrics  
Division of Host-Microbe & Systems Therapeutics



*“Engineering Better Therapeutics with  
Systems Analyses and Genome Editing”*

**2:00 – 2:20**

Over the past decades powerful new immunotherapies have emerged, allowing the treatment of complex diseases. However, the molecules in such therapies are highly complex and expensive. Therefore, there is a need to engineer the cells producing these drugs to increase drug safety, efficacy and affordability. To meet this need, we develop genomic resources and deploy systems biology technologies to study and engineer the pathways that influence mammalian cell growth and protein production. This includes metabolism, the secretory pathway, and glycosylation. In this talk I present recent work in which we mapped out all of the metabolic pathways in Chinese hamster ovary (CHO) cells, the primary host cells for the production of most biotherapeutics. I will further discuss how we have used knowledge from this work to eliminate one of the physiological traits of CHO that has significantly limited the quantity and quality of biotherapeutics: The Warburg effect.

---

---

---

---

---

---

---

---

---

---

## DEBASHIS SAHOO, PHD

Assistant Professor of Pediatrics  
Division of Genome Information Sciences



*“Computationally-driven Answers  
to Disease”*

**2:20 – 2:40**

The identification and characterization of different cell types within normal and diseased tissue are not only critical for the understanding of underlying biology but also in developing more effective therapeutic strategies. We have developed systematic computational approaches to identify genes expressed in these cells by analyzing publicly available, high-throughput gene expression datasets consisting of more than 2 billion measurement points. We developed a set of tools - StepMiner, BooleanNet (a network of Boolean implications), MiDReG (Mining Developmentally Regulated Genes) that uses Boolean implications to predict genes in developmental pathways, and HEGEMON (Hierarchical Exploration of Gene Expression Microarray Online) to identify genes expressed in both normal and malignant tissue development. We demonstrated that coordinated use of these tools could predict genes involved in developmental stages in human normal and cancer tissues. We use examples of human B cells, bladder cancer and colon cancer to show the power of this computational approach. Our recent discovery of a prognostic and predictive biomarker in human colon cancer is published in the New England Journal of Medicine.

---

---

---

---

---

---

---

---

---

---

## VINEET BAFNA, PHD

**KEYNOTE**

Professor of Computer Science & Engineering  
Jacobs School of Engineering

*“Genetic (mal-)adaptation: Nature's  
Spotlight on Human Disease”*

**2:40 – 3:10**



---

---

---

---

---

---

---

---

## TIMOTHY FAIRBANKS, MD

Associate Clinical Professor of Surgery  
Division Chief of Pediatric Surgery  
Vice President/Medical Director of Surgery  
Children's Specialists of San Diego

*“The Utility of D-Dimer in the  
Evaluation of Pediatric Head Trauma”*

**3:25 – 3:35**



---

---

---

---

---

---

---

---



## SALIL UPASANI, MD

Assistant Clinical Professor of Surgery  
Division of Pediatric Orthopedic Surgery



### *“Infant Hip Dysplasia Outcomes in a Multi-Center Study”*

3:35 – 3:45

**Background:** The use of a brace has been shown to be an effective treatment for hip dislocation in infants; however, previous studies of such treatment have been single-center or retrospective. The purpose of the current study was to evaluate the success rate for brace use in the treatment of infant hip dislocation in an international, multicenter, prospective cohort, and to identify the variables associated with brace failure.

**Methods:** All dislocations were verified with use of ultrasound or radiography prior to the initiation of treatment, and patients were followed prospectively for a minimum of 18 months. Successful treatment was defined as the use of a brace that resulted in a clinically and radiographically reduced hip, without surgical intervention. The Mann-Whitney test, chi-square analysis, and Fisher exact test were used to identify risk factors for brace failure. A multivariate logistic regression model was used to determine the probability of brace failure according to the risk factors identified.

**Results:** Brace treatment was successful in 162 (79%) of the 204 dislocated hips in this series. Six variables were found to be significant risk factors for failure: developing femoral nerve palsy during brace treatment ( $p = 0.001$ ), treatment with a static brace ( $p < 0.001$ ), an initially irreducible hip ( $p < 0.001$ ), treatment initiated after the age of 7 weeks ( $p = 0.005$ ), a right hip dislocation ( $p = 0.006$ ), and a Graf-IV hip ( $p = 0.02$ ). Hips with no risk factors had a 3% probability of failure, whereas hips with 4 or 5 risk factors had a 100% probability of failure.

**Conclusions:** These data provide valuable information for patient families and their providers regarding the important variables that influence successful brace treatment for dislocated hips in infants.

---

---

---

---

---

---

---

---



## ANNA KIM, MD

Division of Pediatric Ophthalmology

### *“A Modified Technique for Strabismus Surgery in the Presence of a Scleral Buckle”*

3:45 – 3:55

Ocular motility disturbance is a commonly reported complication following scleral buckling procedures for retinal reattachment. In the presence of extensive scar tissue and a buckling element, the response to standard strabismus surgery is difficult to predict. We describe the outcomes of a surgical technique for strabismus surgery in patients with a scleral buckle that allows for scleral reattachment of muscles and postoperative refinement of ocular alignment using adjustable sutures while preserving the integrity of the exoplant.

---

---

---

---

---

---

---

---

## GEORGE CHIANG, MD

Associate Clinical Professor of Surgery  
Division Chief of Urology



*“Leveraging Informatics for Comparative Effectiveness Research”*

**3:55 – 4:05**

**Background:** The use of a brace has been shown to be an effective treatment for hip dislocation in infants; however, previous studies of such treatment have been single-center or retrospective. The purpose of the current study was to evaluate the success rate for brace use in the treatment of infant hip dislocation in an international, multicenter, prospective cohort, and to identify the variables associated with brace failure.

**Methods:** All dislocations were verified with use of ultrasound or radiography prior to the initiation of treatment, and patients were followed prospectively for a minimum of 18 months. Successful treatment was defined as the use of a brace that resulted in a clinically and radiographically reduced hip, without surgical intervention. The Mann-Whitney test, chi-square analysis, and Fisher exact test were used to identify risk factors for brace failure. A multivariate logistic regression model was used to determine the probability of brace failure according to the risk factors identified.

**Results:** Brace treatment was successful in 162 (79%) of the 204 dislocated hips in this series. Six variables were found to be significant risk factors for failure: developing femoral nerve palsy during brace treatment ( $p = 0.001$ ), treatment with a static brace ( $p < 0.001$ ), an initially irreducible hip ( $p < 0.001$ ), treatment initiated after the age of 7 weeks ( $p = 0.005$ ), a right hip dislocation ( $p = 0.006$ ), and a Graf-IV hip ( $p = 0.02$ ). Hips with no risk factors had a 3% probability of failure, whereas hips with 4 or 5 risk factors had a 100% probability of failure.

**Conclusions:** These data provide valuable information for patient families and their providers regarding the important variables that influence successful brace treatment for dislocated hips in infants.

---

---

---

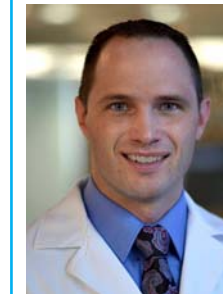
---

---

---

---

---



## DAVID GONDA, MD

Assistant Clinical Professor of Surgery  
Division of Pediatric Neurosurgery

*“Robotic-assisted Endoscopic Third Ventriculostomy for the Treatment of Hydrocephalus”*

**4:05 – 4:15**

---

---

---

---

---

---

---

---

## AMANDA GOSMAN, MD

Clinical Professor of Surgery  
Division of Plastic Surgery



### *“Quality of Life Tool for Children with Facial Deformities”*

**4:15 – 4:25**

**Aims:** Craniofacial conditions (CFCs) can have a severe impact on health-related quality of life (HRQoL) for children worldwide. Existing tools measuring HRQoL for children with CFCs are problematic because they have not been validated in this population or they are limited to one specific CFC. Currently there is no pediatric HRQoL tool that can be utilized across different types of CFCs. The aim of this study is to develop items and support the content validity for an HRQoL measure for children with diverse CFCs and their parents, in English and Spanish.

**Methods:** A semi-structured interview guide based on a comprehensive literature review and expert opinion was used to interview 70 patients ages 7-18+ and 57 parents of patients ages 0-18+. English and Spanish-speakers were equally represented. CFCs included were cleft lip/palate, craniosynostosis, craniofacial microsomia, microtia, and dermatological conditions. Researchers independently reviewed transcribed interviews and identified domains. Domains were then refined through group discussion. Interviews were organized into domains and their sub-categories (i.e. themes). Themes were operationalized into many items via group discussion. Ten items for each domain were chosen to present to subjects during cognitive interviews.

**Results:** Six bilingual and bicultural subscales were developed based on the following domains: (1) “Social Impact”; (2) “Psychological Function”; (3) “Physical Function”; (4) “Family Impact”; (5) “Appearance”; and (6) “Finding Meaning.” Though these domains were common across all groups, Spanish-speaking participants reported more instances of negative social impact and referred to spiritual search for meaning more often than English-speaking participants. Common barriers to treatment reported by families include treatment costs, regional shortage of medical and surgical providers, and the complexity of evaluation and treatment by multiple disciplines.

**Conclusions:** This is the first study to investigate the HRQoL concerns of families across diverse conditions and cultures and highlights the similarities and differences amongst these populations. The sixty chosen bilingual and bicultural items developed in this study will now undergo cognitive testing with subjects to further refine the items and determine their understandability and clarity for subjects. At the conclusion of this phase, thirty finalized items will undergo validation and reliability testing at multiple sites in the United States and Mexico. These iterative steps are consistent with recent FDA guidelines for the development and validation of new patient-reported outcomes measures. The tool will assist in assessment of treatment efficacy of craniofacial conditions from the patient’s perspective, the determination of best treatment plans, and will provide critical data on the HRQoL of underserved populations with CFCs in order to provide evidence-based advocacy for vulnerable communities.

---

---

---



## MATTHEW BRIGGER, MD

Associate Professor of Clinical Surgery  
Division of Otolaryngology

### *“Laryngeal Stenosis in Children: Epidemiology Using a Population Based Approach”*

**4:25 – 4:35**

**Objective:** Laryngeal stenosis is a condition of substantial morbidity to affected children and caretakers. The overall rarity and variable severity has resulted in significant difficulty identifying the epidemiologic composition of this patient population. The primary objective of this study is to determine the current epidemiology of children with laryngeal stenosis in the United States using population level data and develop a model for comparison across healthcare systems on national and international levels.

**Study Design:** Population level database analysis.

**Methods:** The 2012 Kids' Inpatient Database provides data on a sample of all pediatric discharges in the United States during that year. Children under the age of 18 diagnosed with laryngeal stenosis requiring hospitalization were identified by corresponding ICD-9 codes. Database analysis generated national estimates of summary statistics, which were compared to U.S. birth data during the same timeframe. Regression analysis was performed to identify patient characteristics associated with the diagnosis and undergoing a surgical procedure.

**Results:** The estimated prevalence of hospitalized children under age 18 diagnosed with laryngeal stenosis in the United States in 2012 is estimated to be 4001(95%CI: 3136;4866). Of these children, an estimated 916(95%CI: 642; 1191) underwent airway procedures including 282(95%CI:202;362) who underwent a tracheostomy. The estimated annual incidence of being diagnosed with laryngeal stenosis on a population level is 5.4(95%CI:4.2;6.6) per 100,000 children. Regression analysis suggests that male gender ( $p<0.0001$ ) and African American race ( $p<0.0001$ ) are associated with an increased disease burden. In this analysis, there are no specific patient characteristics associated with undergoing a procedure on the airway.

**Conclusions:** Despite well-documented successes in prevention, the true incidence of pediatric laryngeal stenosis in children in the United States is likely higher than previously indicated. Population based estimates are necessary to allow comparison and tracking across healthcare systems both nationally and globally. Comparison between various systems is critical to better understand the effects of prevention and identification on a global scale.

---

---

---

## ANDREW SKALSKY, MD

Associate Clinical Professor of Orthopaedic Surgery  
Dept of Orthopaedic Surgery  
Division of Pediatric Rehab Medicine



*“An Instrumented Glove for Improving Spasticity Assessment”*

**4:35 – 4:45**

Introduction: An instrumented glove worn by clinicians that can augment subjective assessments of muscle hypertonicity with an objective, repeatable metric with reduced inter- and intra- rater variability and improved resolution over existing standards is highly needed. Methods: We present the design and validation of such a system using commercial, off the shelf components. The glove includes spatially-resolved, force-dependent resistive sensor elements and an inertial measurement unit. We developed a mock patient that is equipped with a mechanism to adjust the arm stiffness, a load-cell and a potentiometer to measure the work done to move the arm. The mock-patient provides ground truth to validate the proposed concept. Results: The Pearson correlation coefficient between the glove estimates and mock patient measurements was 0.97. There was also consistent linear agreement between the glove measurements and the subjective physician measurement of Hypertonicity. Conclusions: Preliminary assessments by clinicians indicate that the proposed glove can provide objective metrics of hypertonicity with superior resolution compared to the current gold standard.

---

---

---

---

---

---

---

---

---

---

## VAL CATANZARITE, MD, PHD

Chief of Perinatology/High Risk OB



*“Prenatally Diagnosed Vasa Previa: A Single Institution Series of 96 Cases”*

**4:45 – 4:55**

OBJECTIVE: To describe outcomes for a large cohort of women with prenatally diagnosed vasa previa, determine the percentage in patients without risk factors, and compare delivery timing and indications for singletons and twins.

METHODS: This was a retrospective case series of women with prenatally diagnosed vasa previa delivered at a single tertiary center over 12 years. Potential participants were identified using hospital records and perinatal databases. Patients were included if vasa previa was confirmed at delivery and by pathologic examination. Maternal and newborn data were gathered from medical records.

RESULTS: There were 77 singleton and 19 twin pregnancies with a prenatal diagnosis of vasa previa. There was one neonatal death from congenital heart disease. Perinatal management of recommended elective hospitalizations with corticosteroid administration and elective early delivery resulted in average gestational age for delivery in singletons at 34.7+/-1.6 weeks and 32.8+/-2.2 weeks for twins. Among the 77 singletons, delivery was elective in 48, as a result of contractions or labor in 21, bleeding in four, nonreassuring tracing in two, asymptomatic cervical shortening in one, and preeclampsia in one. Among 19 twins, delivery was elective in six and for contractions or labor in 13. Sixty-eight percent of twins compared with 37% of singletons had nonelective delivery (P<.05). Delivery occurred by 32 weeks of gestation in 6.4% of singletons and 26% of twins (P<.05) and by 34 weeks of gestation in 11% of singletons and 58% of twins (P<.001). Six neonates (5.2%) had major anomalies, all prenatally detected. Respiratory distress syndrome occurred in 57.1% of singletons and 65.7% of twins. Nineteen singletons (24.7%) had no risk factors for vasa previa.

CONCLUSION: Planned preterm delivery for women with prenatally diagnosed vasa previa resulted in elective delivery for singletons in 62% and for twins 32%.

Gestational age at birth on average was 34.7 weeks for singletons and 32.8 weeks of gestation for twins. Major anomalies were frequent as was respiratory distress syndrome. Elective delivery between 34 and 35 weeks of gestation for singletons is reasonable. As a result of the high rate of nonelective delivery in twins, delivery at 32– 34 weeks of gestation may be risk-beneficial. The high rate of singletons without risk factors for vasa previa reinforces the recommendation to screen routinely for cord insertion site.

---

---

---

---

---

---

---

---

---

---

**PETER NEWTON, MD**

**KEYNOTE**

Chief of Orthopedic Surgery & Scoliosis

*“How 3D Imaging of Scoliosis Changes  
What We Think We Know”*

**4:55-5:15**



Traditional imaging of scoliosis has been with 2D plain radiographs and the Cobb angle. The importance of the sagittal plane has certainly been appreciated for many decades of the post Harrington era. Stagnara and others for years have noted the challenge of measuring the coronal and sagittal plane deformities due to the associated axial plane rotation. 3D segmental analysis particularly of the sagittal plane in thoracic AIS suggests many of the segments are in relative or even frank lordosis. The etiology of this “relative anterior overgrowth” is unknown however the loss of kyphosis is greater in larger coronal curves. This finding also holds true for non-idiopathic curves, suggesting a secondary rather than primary cause. It is postulated that increasing coronal deformity associated with greater axial rotation out of the normal sagittal plane, reduces normal forces on the vertebral endplates particularly in the convex and anterior aspects. The Heuter Volkman principle would support more growth in these regions as a result of the altered loading. This theory has been applied to the coronal progression in the past (Stokes viscous cycle), but this also seems to apply in sagittal plane. 3D imaging now allows a segmental analysis of spinal deformity that was not possible with standard radiographs with relevance for both understanding the etiology as well as the treatment of scoliosis.

---

---

---

---

---

---

---

---

# JANUARY 13, 2017

## PRESENTER ABSTRACTS





## FARHAD IMAM, MD, PHD

Assistant Professor of Pediatrics  
Division of Neonatology

*“Insights into Neonatal Hypoxic Injury from  
Animal Models of Human Disease”*

**8:50 – 9:10**

Fine metabolic regulation to adjust for changes in oxygen and energy availability is a conserved, ubiquitous survival strategy of cells and tissues to unpredictable environments. Severe oxygen deprivation can overwhelm these protective strategies and is a common cause of major brain, heart, and kidney injury in adults and newborns alike. Intriguingly, mild hypoxia can be preventative against a later, more severe hypoxia exposure via “hypoxic preconditioning”—a protective phenomenon that is not yet fully understood. We have therefore established and optimized an embryonic zebrafish model to study hypoxic preconditioning in detail using a functional genomic approach. Using this developmental zebrafish model, we validated five novel hypoxia-protective genes from hundreds of hypoxia-regulated genes we identified via differential expression microarray: *irs2a*, *crtc3*, and *camk2g2* have been previously implicated in insulin and glucose metabolism, while *btr01* and *ncam2* are previously uncharacterized. Furthermore, we have generated null mutants using CRISPR in three of these genes (*irs2a*, *crtc3*, *btr01*) and have begun to characterize hypoxia-induced cellular, transcriptional, and metabolic nature of the mutant phenotypes. These results extend our understanding of the mechanisms of hypoxic preconditioning and affirm the discovery potential of this novel vertebrate hypoxic stress model.

---

---

---

---

---

---

---

---

## WENXIAN FU, PHD

Assistant Professor of Pediatrics  
Division of Endocrinology

*“Novel Role of Macrophages in the Pathogenesis of Type 1 Diabetes”*

**9:10 – 9:30**



Tissue-resident macrophages represent a key constituent of the innate immune system in orchestrating inflammation and regulating tissue homeostasis. However, how they regulate adaptive immune responses under inflammatory conditions remains poorly understood. We here report a newly identified subset of tissue-resident macrophages that impact the development and function of CD4<sup>+</sup> Foxp3<sup>+</sup> regulatory T (Treg) cells—a central mechanism in the prevention of lethal autoimmunity and excessive inflammation. This subset of tissue-resident macrophages distinctively express complement receptor of the immunoglobulin superfamily (CR1g). We find that in addition to dampening pathogenic T cell proliferation, CR1g promotes the differentiation of induced Treg (iTreg) cells, by synergizing with TGF- $\beta$  signaling. CR1g also stabilizes Foxp3 expression in iTreg cells. *In vivo* CR1g-Ig treatment increases Treg proportion, particularly in pancreatic islets of NOD mice, a primary animal model for juvenile autoimmune diabetes. Therefore, CR1g may represent a novel therapeutic target to treat diabetes-associated islet inflammation, in that it evokes a synergistic effect by suppressing effector T (Teff) cells and promoting Treg cells.

---

---

---

---

---

---

---

---



## ELLIOT PERENS, MD, PHD

Assistant Professor of Pediatrics  
Division of Nephrology

*“Identification of Factors that Regulate Kidney Cell Fate: Hand2 Inhibits Intermediate Mesoderm Development”*

**9:30 – 9:50**

**Background:** The kidneys and urinary tract are derived from the intermediate mesoderm (IM), yet the regulatory pathways that determine precise IM dimensions and that separate the IM from neighboring portions of the posterior mesoderm are poorly understood.

**Methods:** To study the genetics of early kidney development, we are using zebrafish. Like mammalian kidneys, zebrafish kidneys are derived from the IM, which expresses the same conserved transcription factors (such as *lim1* and *pax2*) as the mammalian IM. Using a combination of loss-of-function and gain-of-function analysis, we have sought to determine the role of *hand2* in IM development.

**Results:** We have found that the bHLH transcription factor Hand2 limits the size of the embryonic kidney by refining IM dimensions. In zebrafish *hand2* mutants, the IM is expanded, and it is diminished when *hand2* is overexpressed. *hand2* is expressed within the posterior mesoderm, laterally adjacent to the IM. A set of venous precursors arise at the interface between these two territories, and *hand2* promotes their development while suppressing IM formation in this region. Furthermore, Hand2 and the similarly localized zinc-finger transcription factor *Osr1* have functionally antagonistic influences on pronephron formation.

**Conclusions:** Together, our data illuminate a previously unrecognized regulation of IM development and suggest a model in which *hand2* functions in opposition to *osr1* to balance the formation of IM and venous progenitors by regulating cell fate decisions in the posterior mesoderm. Our findings have implications for understanding the genetic basis of congenital anomalies of the kidney and urinary tract (CAKUT) and for developing new approaches in regenerative medicine.

---

---

---

---

---

---

---

## ENIKO SAJTI, PHD

Assistant Clinical Professor of Pediatrics  
Division of Neonatology

*“Genome-wide Assessment Reveals Marked Differences in the Transcriptome and Epigenome of Mononuclear Phagocytes in the Lung”*

**9:50 – 10:10**



**Background:** The specialized microenvironment of the airspaces harbors different subsets of mononuclear phagocytes (MP) that are critical for lung development and function. These cells participate in a variety of processes including alveolarization, angiogenesis, surfactant turnover and immune response. However, the phenotypic and functional characteristics of these MP populations are poorly characterized.

**Objectives:** Determine the gene expression profile of four MP, alveolar macrophages (AM), interstitial macrophages (IM), dendritic cells (DC) and monocytes in the mouse lung at different time points during postnatal development.

**Method:** Lungs of C57BL/6 mice were harvested in the sacular phase on postnatal day 2 (PND2), alveolar phase on PND10 and as adults at 2 months of age. MP populations were sorted by fluorescence-activated cell sorting. Gene expression of MP populations at the three time points was determined by RNA-sequencing. Open regions of chromatin were determined using the transposase-accessible-chromatin-sequencing (ATAC-seq) assay. Data were analyzed with HOMER.

**Results:** Significant differences in gene expression were present in the four MP populations with hundreds of mRNA transcripts selectively increased or decreased in only one MP population. Gene ontology annotation of the uniquely expressed genes revealed an enrichment of ion transport and cell adhesion related genes in AM, enrichment of cell adhesion and angiogenesis related genes in DC and a strong enrichment of inflammatory genes in IM and monocytes. AM had reduced expression of genes involved in an inflammatory immune response. Importantly, a substantial number of genes were uniquely regulated in each cell subset during development. In young animals AM were most enriched in cell adhesion and angiogenesis related genes, whereas in adult animals these genes were more enriched in IM. Motif enrichment analysis of open chromatin regions identified several MP subset specific transcription factors binding motifs.

**Conclusion:** Transcriptomic analysis of resident lung MP revealed a marked diversity of these cells in steady state and during postnatal development. Signature genes of each population residing in their distinct microenvironment highlighted several aspects of their functional specialization in the healthy lung. AM had a lower expression of inflammatory genes suggesting transcriptional mechanisms that prevent them from mounting an excessive and pathological immune response. In contrast, IM and monocytes were enriched in immune response genes, enabling them to provide a robust defense mechanism if a pathogen passes the alveolar barrier. These studies provide insights into the unique contributions of each MP population during lung development and a basis for their distinct roles in lung pathology.

---

---

---

---

---



## RON EVANS, PHD

Professor of Biology, Biomedical Sciences, and Neuroscience  
Director, Gene Expression Laboratory  
Howard Hughes Medical Institute Investigator  
March of Dimes Chair in Molecular and Developmental Biology  
Salk Institute for Biological Studies

### KEYNOTE

*“Nuclear Receptors, Oxygen & the First Breath”*

**10:15–10:45**

Although the lung is a defining feature of air-breathing animals, the pathway controlling the formation of type I pneumocytes, the cells that mediate gas exchange, is poorly understood. In contrast, the glucocorticoid receptor (GR) and cortisone related drugs have long been known to promote type II pneumocyte maturation; prenatal administration of betamethasone is commonly used to attenuate the severity of neonatal respiratory distress syndrome (NRDS). We show that knock-in mutations of the nuclear co-repressor SMRT (silencing mediator of retinoid and thyroid hormone receptors) in mice (SMRTmRID) produces a previously unidentified respiratory distress syndrome caused by prematurity of the type I pneumocyte. Though unresponsive to glucocorticoids, treatment with anti-thyroid hormone drugs (propylthiouracil or methimazole) completely rescues SMRT-induced RDS, suggesting an unrecognized and essential role for the thyroid hormone receptor (TR) in lung development. We show that TR and SMRT controls Klf2, a master-regulator of the type I pneumocyte gene program. Conversely, mice without lung Klf2 lack mature type I pneumocytes and die shortly after birth, closely recapitulating the SMRTmRID phenotype. These results identify TR as a second nuclear receptor involved in lung development, specifically type I pneumocyte differentiation, and suggest a possible new type of therapeutic option in the treatment of NRDS that is unresponsive to glucocorticoids.

---

---

---

---

---

---

---

---

---

---

## LORI BRODERICK, MD, PHD

Assistant Professor of Pediatrics  
Division of Allergy, Immunology  
& Rheumatology



*“Time to Unwind: A Novel B Cell Immunodeficiency”*

**11:05 – 11:25**

B cell development in humans requires active selection for immunologic maturity and elimination or inactivation of autoreactive cells, yet many details regarding this selection remain unknown. We previously described a presentation of complete B cell immunodeficiency, associated with facial dysmorphism, limb and genital anomalies. We used whole genome sequencing of affected patients and unaffected first-degree relatives from two families to identify genetic mutations in a common gene. Variant analysis revealed mutations in *TOP2B*, which encodes topoisomerase II $\beta$  (Top2b), a highly conserved enzyme responsible for double-stranded DNA breaks crucial to DNA replication and gene transcription. A role for Top2b in B cell development and differentiation has not previously been described. We hypothesize that mutations in *TOP2B* are responsible for the immunoclinical phenotype observed in patients due to negative effects on B cell differentiation, and elimination of immature B cells. Here, we use *in vitro*, *ex vivo* and *in vivo* models to 1) analyze how our identified mutations in *TOP2B* affect protein expression and function, and 2) examine how loss of Top2b function affects regulation of B cell development. Our goal is that an understanding of the role of Top2b in B cell development will have far reaching effects on our ability to treat B cell-driven diseases.

---

---

---

---

---

---

---

---



**SHELLEY LAWRENCE, MD, MS**

Assistant Clinical Professor of Pediatrics  
Division of Neonatology

*“New Approaches to the Evaluation of Neonatal Neutrophils and Sepsis Risk”*

**11:25 – 11:45**

Infection is the second leading cause of neonatal mortality worldwide and results in the deaths of over 1 million newborns annually, more than half during the first week of life. Technologies presently used to diagnose neonatal sepsis, such as manual differentials and routine blood culture methodologies, are antiquated, time consuming, and in the case of manual differentials, very subjective. Prolonged delays in pathogen identification and antibiotic sensitivity testing (up to 4-5 days) can lead to unnecessary exposure to broad-spectrum antibiotics resulting in bacterial antibiotic resistance in non-infected neonates, while preventing targeted antimicrobial therapy in septic neonates. Prolonged antibiotic use, while critical and potentially lifesaving in the treatment of actual infections in this vulnerable patient population, can result in invasive fungal (Candida) infections, necrotizing enterocolitis, and death. Studies of the human microbiome also raise grave concerns about epidemiological links to the development of asthma, allergies, and metabolic disorders (such as obesity and diabetes). Importantly, even a single course of antibiotics can permanently change the human gastrointestinal microbiota. New technologies such as High Resolution Heat Melt for the detection of bacteria and flow cytometric methods for the calculation of neutrophil differentials can provide accurate, low-cost, robust, and valid tools for the rapid diagnosis of neonatal sepsis on minimal blood volume. Current clinical results and future directions of these two platforms will be presented.

---

---

---

---

---

---

**SEEMA ACEVES , MD, PHD**

Associate Professor of Clinical Pediatrics  
Division of Allergy, Immunology & Rheumatology



*“Eosinophilic Esophagitis – A Diagnosis That’s Hard to Swallow: Mechanisms and Clinical Impacts of Tissue Remodeling”*

**11:45 – 12:05**

Eosinophilic esophagitis (EoE) is a relatively newly described antigen driven disease of the esophagus which is increasing in incidence and prevalence in both children and adults. EoE requires repeated endoscopy with biopsy for diagnosis and management. Disease complications include esophageal strictures, rigidity, and food impactions. These complications arise from the process of tissue remodeling and can be assessed using new functional endoscopic tools. The NIH funded Aceves laboratory works to understand the mechanisms of esophageal remodeling including novel inflammatory cell populations and the effects of fibrotic growth factors and esophageal rigidity on structural cell function. In order to answer these mechanistic questions, the laboratory focuses on understanding the disease course in children and utilizes novel human model platforms and cells to dissect these processes.

---

---

---

---

---

---

---

---





### ADRIANA TREMOULET, MD

Associate Professor of Clinical Pediatrics  
Division of Host Microbe Systems & Therapeutics

*“The Emerging Story of Atorvastatin for Treatment of Acute Kawasaki Disease”*

**12:05 – 12:25**

Since the 1980s, the primary treatment of acute Kawasaki disease (KD), the most common cause of acquired heart disease in children, has been intravenous immunoglobulin (IVIG) and aspirin. However, up to 20% of children with acute KD will not respond to this initial therapy and be at higher risk for coronary artery abnormalities (CAA). Repurposing of existing medications that could target key pathways in the CAA pathogenesis of KD could lead to quickly finding safe interventions for CAA in KD. Atorvastatin has many appealing properties that could be beneficial in preventing or abrogating CAA formation including, increasing the number and suppressive function of regulatory T-cells and reducing endothelial to mesenchymal transition that results in myofibroblasts which lead to CAA. Thus, given that atorvastatin could prove to be a powerful anti-inflammatory therapy for CAA in KD, we embarked on a Phase I/IIa trial and have completed the Phase I dose-escalation study which demonstrated that a short course of atorvastatin is safe in acute KD. Simultaneously we are elucidating the anti-inflammatory and vessel healing mechanisms of atorvastatin in using human umbilical vein endothelial cells in tissue culture.

---

---

---

---

---

---

---

---

---

---

### FERNANDO MARTINEZ, MD

**KEYNOTE**

Regents’ Professor of Pediatrics  
Division of Pulmonology/Allergy and Immunology  
Director, BIOS Institute  
Director, Arizona Respiratory Center  
University of Arizona



*“Hygiene Hypothesis to Early Human Microbiome: Understanding Asthma Origins”*

**12:30–1:00**

Dr. Fernando D. Martinez is a Regents’ Professor and Director of the Asthma and Airway Disease Research Center at the University of Arizona in Tucson.

Dr. Martinez is a world-renowned expert, and one of the most highly regarded researchers, in the field of childhood asthma. His primary research interests are the natural history, genetics, and treatment of childhood asthma.

His groundbreaking research has had an impact on his field in numerous ways, most prominent among them the development of the concept of the early origins of asthma and COPD. This concept is now widely accepted as the potential basis for the design of new strategies for the prevention of these devastating illnesses affecting millions of children and adults worldwide. In addition, Dr. Martinez has made important contributions to our understanding of the role of gene-environment interactions in the development of asthma and allergies. He has also been the principal investigator of one of the Clinical Centers that are part of the NHLBI Asthma Treatment Networks, which have contributed fundamental new evidence on which to base national guidelines for the treatment of the disease.

Dr. Martinez currently serves on national scientific boards including the NHLBI National Advisory Council and the National Scientific Council on the Developing Child. He was a member of the National Asthma Education and Prevention Program that was responsible for the development of the Expert Panel Report: Guidelines for the Diagnosis and Management of Asthma in 1997 and its first revision in 2001. He also has been a member of the FDA Pulmonary-Allergy Drugs Advisory Committee and the Board of Extramural Advisors of the National Heart, Lung, and Blood Institute (NHLBI).

---

---

---

---

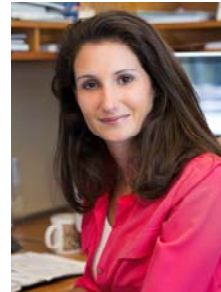


## STEPHANIE CHERQUI, PHD

Associate Professor of Pediatrics  
Division of Genetics

*“Hematopoietic Stem Cell Gene Therapy for the Lysosomal Storage Disorder Cystinosis: Clinical Translation and Application to Mitochondrial Diseases”*

**2:30 – 2:50**



Hematopoietic stem and progenitor cells (HSPCs) are ideal candidates for use in regenerative medicine and cell replacement therapies because of self-renewal capacity and safety. While the ability of HSPC transplantation to rescue non-hematopoietic tissue remains controversial, we previously demonstrated that a single systemic transplantation of wild-type (WT) HSPCs led to long-term kidney, eye and thyroid preservation in a mouse model of the multi-systemic lysosomal storage disorder, cystinosis.

An autologous HSPCs gene therapy approach has then been developed with Ctns<sup>-/-</sup> HSPCs gene-modified ex vivo to express a functional CTNS cDNA using the lentiviral vector pCCL-CTNS. Preclinical studies showed that tissue cystine decrease and kidney function rescue were also achieved with this strategy. The toxicology and pharmacology studies required by the FDA are in progress to obtain an Investigational New Drug for a phase I clinical trial. For the design and conduct of the future clinical trial, the Cystinosis Stem Cell and Gene Therapy Consortium was recently created and is composed of experts in cystinosis, bone marrow transplant and gene therapy, most of them being from the Pediatrics Department at UC San Diego.

The cellular mechanism for stem cell mediated-tissue repair involved the differentiation of WT HSPCs into macrophages that delivered lysosomes bearing functional cystinosis into diseased host cells via long tubular extensions known as “tunneling nanotubes” (TNTs). This finding allowed us to successfully apply HSPC transplantation to the mitochondrial neurodegenerative disorder, Friedreich’s Ataxia, for which there is no treatment, and open new perspective in the treatment of mitochondrial disorders.

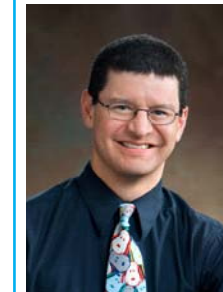
---

---

---

---

---



## PETER ZAGE, MD, PHD

Associate Professor of Pediatrics  
Division of Hematology/Oncology

*“Growth Factor Receptor Trafficking: Recycling Targets for Neuroblastoma Therapy”*

**2:50– 3:10**

Children with high-risk neuroblastoma have poor outcomes, and improved understanding of the mechanisms underlying neuroblastoma pathogenesis, recurrence, and treatment resistance will identify novel targets for future therapies. Aberrant growth factor receptor (GFR) expression and tyrosine kinase signaling are associated with the pathogenesis of many malignancies, and these kinases and their downstream signaling pathways are targets for novel therapies. We previously demonstrated that the E3/E4 ubiquitin ligase UBE4B, whose gene is found in the chromosome 1p36 region commonly deleted in neuroblastoma tumors, is critical for appropriate GFR trafficking, degradation, and signaling, suggesting a previously undiscovered link between GFR trafficking and neuroblastoma pathogenesis. However, the role of UBE4B and GFR trafficking in neuroblastoma pathogenesis has not previously been investigated.

Using neuroblastoma tumor expression array data-sets, we have demonstrated associations between *UBE4B* gene expression and neuroblastoma patient outcomes and prognostic features. We have also shown that reduced UBE4B protein expression in neuroblastoma tumors was associated with high-risk features, a lack of differentiation, and with ERK activation. These results suggest UBE4B-mediated GFR trafficking and degradation may contribute to the poor prognosis of neuroblastoma tumors with 1p36 deletions and that UBE4B expression may mediate neuroblastoma differentiation. We further evaluated the role of GFR trafficking in neuroblastoma tumors via investigation of a germline polymorphism in the *FGFR4* gene that results in altered FGFR4 trafficking, degradation, and signaling. This *FGFR4* Arg388 polymorphism has been shown to be associated with decreased survival, treatment resistance, and aggressive disease in many malignancies. We identified an association of this *FGFR4* polymorphism with altered FGFR4 trafficking, degradation, and signaling in neuroblastoma cells, further suggesting a key role for GFR trafficking in neuroblastoma pathogenesis.

Based on the altered activity of the Ras/MAPK signaling pathway mediated by GFR trafficking and degradation, we evaluated the efficacy of a novel MEK inhibitor binimetinib in neuroblastoma tumor cell lines. Neuroblastoma tumor cells show a range of sensitivities to binimetinib, with sensitive cells demonstrating complete loss of phosphorylated ERK, while resistant cells demonstrated either incomplete loss of ERK phosphorylation or minimal effects on MEK phosphorylation, suggesting alternative mechanisms of resistance. UBE4B and NF1 protein expression levels correlated with responses to binimetinib, supporting the use of UBE4B and NF1 as additional biomarkers to identify patients that may respond to MEK inhibition. These results further suggest that GFR trafficking regulated by UBE4B leads to altered activity of downstream intracellular signaling pathways that can be used as effective targets for novel therapies.

---

---

## PAULINA ORDONEZ, MD

Assistant Professor of Pediatrics  
Division of Gastroenterology



*“Accelerating Cures for Rare Diseases: Therapeutic Rescue of Niemann-Pick type C1 Stem Cell-derived Neurons Using Repurposed Drugs”*

**3:10 – 3:30**

There are approximately 7,000 rare diseases and less than 5% have an FDA approved therapy. Out of the 30 million people affected by rare diseases in the United States half are children, and 30% of these children will not live to see their 5<sup>th</sup> birthday. The challenges of drug development for rare diseases are made even more significant by high research costs, small populations and lack of incentive. Human stem cell research offers the unprecedented opportunity to accelerate therapeutic development for rare diseases through rigorous mechanistic studies and compound screening in disease relevant cells. One example is Niemann-Pick type C1 (NPC1), a severe lysosomal cholesterol storage disease that causes significant neurodegeneration leading to death, typically in childhood. There is no FDA approved treatment for NPC1, and drugs that are currently being tested have a suboptimal safety profile and insignificant brain penetration. Hence, there is a dire need to identify new therapies that are efficient, safe and have an adequate bioavailability profile.

Using a small bank of human induced pluripotent stem cells (hiPSC) derived from patients carrying NPC1 disease-causing mutations, we found that cholesterol sequestration in the lysosome results in: (1) hyperactivation of autophagy in response to cholesterol “starvation” and (2) decreased cellular viability due to substantial disruption of mitochondrial turnover by autophagy, which leads to mitochondrial membrane depolarization and generation of reactive oxygen species. We used a hypothesis-driven drug screening approach to discover a class of compounds that protect against mitochondrial dysfunction and improve viability in human NPC1 neurons. Two of the most promising compounds are already approved for use in humans by the FDA and are known to cross the blood-brain barrier, offering immediate therapeutic potential for NPC1 and related lysosomal storage diseases. Our studies illustrate the critical value of using mechanistic studies in human cells to innovate therapies for rare diseases through the repurposing of FDA-approved compounds.

---

---

---

---

---

---

---

---

## MAMATA SIVAGNAM, MD

Associate Clinical Professor of Pediatrics  
Division of Gastroenterology



*“Lost in Translation: Effects of a Mutant Protein in the Gut”*

**3:30 – 3:50**

**Background:** Congenital tufting enteropathy (CTE) is an intractable diarrheal disease of infancy presenting with profuse watery diarrhea, electrolyte imbalances, and impaired growth. Intestinal pathology includes villous atrophy, crypt hyperplasia and epithelial tufts leading to intestinal failure. We have discovered mutations in *epithelial cell adhesion molecule (EpCAM)* as the cause of disease in CTE patients but we have yet to understand how these mutations are responsible for structural abnormalities of the villi in CTE. Recently, we made an observation that mutant forms of EpCAM are sequestered in the endoplasmic reticulum (ER) compared with the normal localization of WT EpCAM at the plasma membrane. We hypothesize mutated EpCAM causes increases in key molecular pathways involved in the Unfolded Protein Response (UPR), due to ER stress.

**Methods:** An *in vivo* mouse model, based on a mutation found in CTE patients, was developed allowing for inducible deletion of exon 4 in *Epcam* resulting in a mutant protein with decreased expression. Protein lysate and RNA from tamoxifen-induced *Epcam*<sup>Δ4/Δ4</sup> mice was used to evaluate UPR mediators, GRP-78 via western blot, and Blos1 and Scara3 via qPCR. The level of apoptosis was evaluated by immunofluorescence of caspase-3. Data presented are means + SEM and statistical significance was assessed by student's unpaired t-test.

**Results** GRP-78, an indicator of activation of UPR, was found to be significantly increased in ind.*Epcam*<sup>Δ4/Δ4</sup> mice (p=0.0227, n=5). Scara3 and Blos1 RNA was found to be significantly reduced by 51% and 46% respectively in mutant mice tissue (p<0.001, n=5). Apoptosis was not found to be elevated because no significant change in the levels of caspase-3 was seen between mutant and littermate control mice.

**Conclusion:** EpCAM causes elevated ER stress, indicated by an increase in GRP-78 and decreases in Scara3 Blos1 RNA transcripts. However, the level of ER stress is still below the apoptotic threshold, indicated by the lack of increase in caspase-3. These conditions indicate that the sequestration of EpCAM in the ER is causing activation of the UPR. UPR activation may be contributing to the structural and functional abnormalities of the villi in CTE patients.

---

---

---

---

---

---

---

---

**LARRY GOLDSTEIN, PHD**

**KEYNOTE**

Distinguished Professor of Cellular and Molecular Medicine  
Department of Neurosciences  
Director, UC San Diego Stem Cell Program  
Scientific Director, Sanford Consortium for Regenerative Medicine  
Director, Sanford Stem Cell Clinical Center

*“Probing the Secrets of Alzheimer’s Disease with Stem Cells”*

**3:55 – 4:25**



The use of human induced pluripotent stem cells has allowed us to gain an understanding of the molecular mechanisms leading to hallmark AD pathologies, in differentiated neurons and astrocytes. Our “disease in a dish” models, using isogenic backgrounds, provide an ideal starting point to define the effects of known and recently described AD risk factors.

---

---

---

---

---

---

---

---

---

---

**ALBERT “AL” PISANO, PHD**

**KEYNOTE**

Distinguished Professor, Mechanical & Aerospace Engineering and Electrical & Computer Engineering  
Walter J. Zable Endowed Chair of Engineering  
Dean, UCSD Jacobs School of Engineering

*“Medically-driven Engineering Innovation: The Best Days Yet to Come”*

**4:30 – 5:00**



Beginning with a very short review of the Jacobs School of Engineering, this talk will proceed through a review of medical/engineering projects currently in progress. Building upon a growing strength in data streams, data analytics and new actions/agents, this medical/engineering research will be harnessed to prosecute a vision for “The Collaboratories for the Digital Future.” The research direction has promise to bring real-time analytics via multiple data streams over new networks to enable patients and doctors to make decisions and take actions with unprecedented speed and accuracy.

---

---

---

---

---

---

---

---

---

---

JANUARY 13, 2017  
POSTER PRESENTATION  
1:30-2:30 & 5:15-6:30

## ALLERGY, IMMUNOLOGY, AND RHEUMATOLOGY

(1) ABDULLA ALSAGGAF

*PROFOUND B-CELL DEFICIENCY IN A CHILD WITH WOLF-HIRSCHHORN SYNDROME*

(4) BOB GENG

*PENICILLIN SKIN TESTING IN THE EVALUATION AND MANAGEMENT OF PENICILLIN ALLERGY IN AN OUTPATIENT PEDIATRIC*

(5) ARIELA HAIMOVICH

*L-SULFORAPHANE, A POTENTIAL NATURAL INHIBITOR OF THE NLRP3 INFLAMMASOME "EAT YOUR BROCCOLI, A CURE FOR INFLAMMATION"*

(46) JUN OYAMADA

*GENETIC VARIATION IN THE SLC8A1 CALCIUM SIGNALING PATHWAY IS ASSOCIATED WITH BIFID T WAVES ON THE ECG FROM ACUTE KAWASAKI DISEASE PATIENTS*

(32) CHISATO SHIMIZU

*WHOLE GENOME SEQUENCING OF AN AFRICAN AMERICAN FAMILY HIGHLIGHTS TOLL LIKE RECEPTOR 6 VARIANTS IN KAWASAKI DISEASE SUSCEPTIBILITY*

## BIOCHEMICAL GENETICS AND METABOLOMICS LAB

(101) BRUCE BARSHOP

*TARGETED METABOLOMIC PATHWAY ANALYSIS. A CASE STUDY IN CYSTINOSIS*

(102) ILYA GERTSMAN

*METABOLOMICS STUDIES OF TYROSINE CATABOLISM DISORDERS REVEAL PERTURBATION OF TRYPTOPHAN METABOLISM IN BOTH HOST AND MICROBIOME*

## CARDIOLOGY

**(35) DAVID AURIGEMMA**

*RADIOFREQUENCY ABLATION/BALLOON DILATION AND RIGHT VENTRICULAR OUTFLOW TRACT STENTING AS ALTERNATIVE PALLIATION FOR INFANTS WITH PULMONARY ATRESIA AND TETRALOGY OF FALLOT*

**(37) MOHAMMAD EBRAHIM**

*CONFIRMED LONG QT SYNDROME IN PATIENTS WITH CONGENITAL HEART DISEASE: MODES OF PRESENTATION AND DIAGNOSIS*

**(38) HOWAIDA EL-SAID**

*3D ROTATIONAL ANGIOGRAPHY FOR ASSESSMENT OF CORONARY ARTERIES DURING MELODY VALVE IMPLANTATION: INTRODUCING A TECHNIQUE THAT MAY IMPROVE OUTCOMES*

**(39) YUDY FONSECA**

*CORRELATION OF TRANSJUGULAR LIVER BIOPSY IN FONTAN PATIENTS WITH MAGNETIC RESONANCE ELASTOGRAPHY & HEMODYNAMICS*

**(49) JOSE SILVA-SEPULVEDA**

*LATE FOLLOW UP IN THE DAMUS-KAYE-STANSEL PROCEDURE: RISK FACTORS FOR A HIGH INCIDENCE OF SUDDEN CARDIAC DEATH*

**(50) HEATHER SUN**

*PRENATAL DETECTION OF CRITICAL CONOTRUNCAL DEFECTS REMAINS LOW DESPITE REVISED OBSTETRICAL IMAGING GUIDELINES*

## CARDIOLOGY & CV SURGERY

**(36) ANDRÁS BRATINCSÁK**

*RE-STENTING IMPROVES VESSEL PATENCY, LUMEN AREA AND ENDOTHELIALIZATION: A FEASIBILITY STUDY IN PIGLETS DEMONSTRATING RE-STENTING WITH SIMULTANEOUS INTENTIONAL FRACTURE OF PREVIOUSLY PLACED STENTS*

**(43) JESSE LEE**

*TRANSCATHETER DUCTUS ARTERIOSUS STENTING WITH EXPECTED REDILATION AS A SURGICAL ALTERNATIVE*

**(44) SULAIMAN LUBEGA**

*UPDATE ON UGANDA HEART INSTITUTE 5 YEAR PROSPECTIVE PLAN TOWARD INDEPENDENT PEDIATRIC CARDIAC CATHETERIZATION*

**(45) RAGHAV MURTHY**

*REFRAMING THE STS STAR RATING SYSTEM: A HEART CENTER PERSPECTIVE*

**(47) KANISHKA RATNAYAKA**

*RADIATION-FREE MRI HEART CATHETERIZATION IN CHILDREN*

**(48) RODRIGO RIOS**

*3D ROTATIONAL ANGIOGRAPHY (3DRA) GUIDANCE OF NON-TRADITIONAL ASCENDING AORTA STENTING*

## CARDIOLOGY/RADIOLOGY

**(40) SANJEET HEGDE**

*FONTAN REVISION: PRE-SURGICAL PLANNING USING FOUR DIMENSIONAL (4D) FLOW AND THREE DIMENSION-*

**(41) SANJEET HEDGE**

*EXTERNALLY IMPLANTABLE MICRO SENSOR TO MONITOR INTRACARDIAC PRESSURE IN CHILDREN WITH COMPLEX CONGENITAL HEART DEFECTS*

**(42) SANJEET HEGDE**

*PATIENT-SPECIFIC MODELING OF CARDIAC BIOMECHANICS IN REPAIRED TETRALOGY OF FALLOT*

## CELLULAR & MOLECULAR MEDICINE

**(26) PAULINA ORDONEZ**

*AN INDUCED PLURIPOTENT STEM CELL (IPSC) MODEL TO STUDY MECHANISMS OF NON-ALCOHOLIC FATTY LIVER DISEASE (NAFLD) ASSOCIATED WITH PNPLA3 POLYMORPHISMS IN HUMAN HEPATOCYTES*

**(103) EMMA WU**

*TARGETING MITOCHONDRIA DYSFUNCTION AS A PROSPECTIVE AVENUE FOR DRUG DISCOVERY IN NIEMANN PICK DISEASE TYPE C*



## CHILD AND ADOLESCENT SERVICES RESEARCH CENTER

### (67) MARY BAKER-ERICZÉN

*SUCCESS PROGRAM FOR ADULTS WITH AUTISM: TEACHING VOCATIONAL SOFT SKILLS, AN INTEGRATION OF COGNITIVE ENHANCEMENT AND SOCIAL SKILLS TRAINING*

### (74) ERIN DELKER

*THE EFFECTS OF TIMING AND INTENSITY OF WEIGHT-GAIN ON IRON STATUS AMONG A COHORT OF CHILEAN INFANTS*

### (77) MEGHAN FITCH

*AN EXECUTIVE FUNCTIONING INTERVENTION FOR TEENS AND YOUNG ADULTS WITH ASD: COMMAND & CONTROL COGNITIVE TRAINING PILOT STUDY*

### (82) MAYA KUMAR

*A BRIEF TRAINING VIDEO IMPROVES PROVIDER RECOMMENDATION OF THE HUMAN PAPILLOMAVIRUS VACCINE*

### (83) VIVIAN LI

*IDENTIFYING ACTIVE INGREDIENTS: EXAMINING THE RELATIONSHIP BETWEEN TEACHER FIDELITY OF IMPLEMENTATION OF CLASSROOM PIVOTAL RESPONSE TEACHING AND STUDENT ENGAGEMENT*

### (86) OLGA ROSE

*ASSOCIATION BETWEEN TELEVISION VIEWING AND COMPUTER USAGE AND COGNITIVE SCORES IN YOUNG ADULTS*

### (88) ENRIQUE VELAZQUEZ

*MENTAL HEALTH BENEFITS OF AN INNOVATIVE INTERVENTION PROGRAM IN ADULTS WITH ASD*

### (90) VICTORIA WU

*ASSOCIATIONS BETWEEN MATERNAL DEPRESSIVE SYMPTOMS, MATERNAL PROVISION OF SUPPORT, AND CHILD VERBAL COGNITION OVER A 16-YEAR PERIOD*

## DERMATOLOGY

### (70) JENNA BOROK

*PROSPECTIVE STUDY ON THE EFFICACY AND SYSTEMIC ABSORPTION OF TOPICAL TIMOLOL FOR INFANTILE HEMANGIOMAS*

### (7) ANDREA WALDMAN

*CHARACTERIZATION OF GENOTYPE, VIRULENCE, AND MICROFLORAL BALANCE OF S. AUREUS STRAINS CAUSING INFECTION IN CHILDREN WITH ATOPIC DERMATITIS VERSUS CONTROLS*

## DYSMORPHOLOGY & TERATOLOGY

### (15) MARIA ACUERO

*AN EDUCATIONAL FEEDBACK INTERVENTION TO PREVENT ALCOHOL-EXPOSED PREGNANCIES AMONG NON-PREGNANT LOW-INCOME LATINAS*

### (2) GRETCHEN BANDOLI

*AUTOIMMUNE CONDITIONS AND COMORBID DEPRESSION IN PREGNANCY: EXAMINING THE RISK OF PRETERM BIRTH AND PREECLAMPSIA*

### (16) KERRI BERSTRAND

*UNTARGETED MASS SPECTROMETRY ANALYSIS IN HUMAN MILK SAMPLES: A COMPARISON TO MATERNAL REPORT DATA IN THE UCSD HUMAN MILK BIOREPOSITORY COHORT*

### (17) DAVE HIRAL

*APPLICATION OF A SCREENING TOOL IN IDENTIFYING FETAL ALCOHOL SYNDROME FACIAL PHENOTYPES IN A LARGE POPULATION BASED PEDIATRIC SAMPLE – HIT OF MISS?*

### (18) KRISTI PALMSTEN

*PATTERNS OF PREDNISON USE DURING PREGNANCY: DAILY AND CUMULATIVE DOSE*

## EMERGENCY MEDICINE

**(68) TIFFANI BARHAM**

*UNDERSTANDING REVISITS TO A PEDIATRIC EMERGENCY DEPARTMENT: A PILOT STUDY IN THE PATIENT EXPERIENCE*

**(72) HEATHER CONRAD**

*A SPECIALIZED PEDIATRIC EMERGENCY MEDICINE TRACK DECREASES COMPUTED TOMOGRAPHY IN HEAD INJURED PATIENTS*

**(73) HEATHER CONRAD**

*THE IMPACT OF BEHAVIORAL HEALTH PATIENTS ON A PEDIATRIC EMERGENCY DEPARTMENT'S LENGTH OF STAY AND LEFT WITHOUT BEING SEEN*

**(75) JOELLE DONOFRIO**

*COMPARING THE CRITERIA OUTCOMES TOOL TO VARIOUS MASS CASUALTY INCIDENT TRIAGE ALGORITHMS IN CHILDREN < 15 YEARS OF AGE USING THE NATIONAL TRAUMA DATABASE*

**(81) JOHN KANEGAYE**

*THE ASSOCIATION OF URINE QUANTITATIVE METHAMPHETAMINE LEVELS WITH CLINICAL AND CHILD PROTECTION OUTCOMES IN ACUTE PEDIATRIC METHAMPHETAMINE INGESTION*

**(84) MICHAEL LONG**

*SEATED POSITION FOR INFANT LUMBAR PUNCTURE DOES NOT IMPROVE SONOGRAPHICALLY MEASURED SPINAL DIMENSIONS COMPARED TO LATERAL DECUBITUS POSITION*

**(89) SHANNON WAI**

*THE EFFECTIVENESS OF A PEDIATRIC EMERGENCY MEDICINE BLOCK EDUCATION SESSION FOR PEDIATRIC RESIDENTS*

## ENDOCRINOLOGY

**(8) WENXIAN FU**

*REGULATION OF ISLET AUTOIMMUNITY IN TYPE 1 DIABETES BY MODULATING INTERLEKIN-2 RECEPTOR SIGNALING*

**(9) JANE KIM**

*URINE METABOLOMIC PROFILING REVEALS A UNIQUE SIGNATURE FOR TYPE 2 DIABETES IN YOUTH*

**(10) JANE KIM**

*DISTINCT GENE SIGNATURES CAN PREDICT INSULIN RESISTANCE IN PUBERTAL MICE PRIOR TO THE DEVELOPMENT OF DIET-INDUCED OBESITY IN EARLY ADULTHOOD*

**(13) SEJAL KADAKIA**

*EFFECTS OF TYPE 1 DIABETES MELLITUS AND INCREASED WEIGHT ON METABOLOMIC AND MICROBIOME PROFILES IN THE PEDIATRIC POPULATION*

**(14) SEJAL KADAKIA**

*MUTATIONAL ANALYSIS IN 46 PEDIATRIC THYROID CANCER SUBJECTS AND CORRELATIONS WITH AGE, ETHNICITY, AND CLINICAL PRESENTATION*

**(85) MAJA MARINKOVIC**

*EARLY SOCIAL TRANSITION IN PATIENTS WITH CHILDHOOD ONSET OF GENDER DYSPHORIA IS COMMONLY SEEN IN SAN DIEGO'S GENDER MANAGEMENT (GEM) CLINIC*

**(12) CHUN ZENG**

*PSEUDOTEMPORAL ORDERING OF SINGLE CELLS REVEALS METABOLIC CONTROL OF POSTNATAL BETA-CELL PROLIFERATION*

## GASTROENTEROLOGY, HEPATOLOGY & NUTRITION

**(19) ARIEL FELDSTEIN**

*HEPATOCTE-DERIVED, CIRCULATING EXTRACELLULAR VESICLES CORRELATE WITH FIBROSIS STAGE AND PORTAL HYPERTENSION IN PATIENTS WITH NONALCOHOLIC STEATOHEPATITIS*

**(20) ARMEN GHARIBANS**

*IMPROVING THE CLINICAL UTILITY OF ELECTROGASTROGRAPHY BY CONDUCTING SIMULTANEOUS HIGH-RESOLUTION ELECTROGASTROGRAM AND ANTRoduODENAL MANOMETRY IN CHILDREN*

**(21) KATHRYN HARLOW**

*APPLICATION OF THE NHLBI DYSLIPIDEMIA GUIDELINES IN CHILDREN WITH NONALCOHOLIC FATTY LIVER DISEASE*

**(22) MAHEEN HASSAN**

*ESOPHAGEAL DISTENSIBILITY AS A PREDICTOR OF CLINICAL PHENOTYPE IN PEDIATRIC PATIENTS WITH EOSINOPHILIC ESOPHAGITIS*

**(24) CASEY JOHNSON**

*HEPATIC STELLATE CELL SPECIFIC NLRP3 INFLAMMASOME ACTIVATION RESULTS IN SPONTANEOUS FIBROSIS*

**(25) KIMBERLY NEWTON**

*BIRTH WEIGHT IS AN IMPORTANT LIFE STAGE FACTOR FOR CHILDREN WITH NONALCOHOLIC FATTY LIVER DISEASE*

**(27) SUSANNE SCHUSTER**

*OXIDIZED LINOLEIC ACID LEADS TO OXIDATIVE STRESS-INDUCED MITOCHONDRIAL DYSFUNCTION AND NLRP3 INFLAMMASOME ACTIVATION IN THE LIVER*

## GENOMIC MEDICINE

**(29) YAN DING**

*DEVELOPMENT OF A CLINIC GENOMIC LABORATORY AT RADY CHILDREN'S INSTITUTE FOR GENOMIC MEDICINE*

**(23) AMBER HILDRETH**

*UTILITY OF RAPID WHOLE-GENOME SEQUENCING IN PATIENTS WITH NEONATAL CHOLESTASIS*

**(31) LAURA PUCKETT**

*VALIDATION OF CUSTOM VARIANT ANALYSIS ASSAY USING SANGER SEQUENCING FOR RADY CHILDREN'S HOSPITAL MOLECULAR GENETICS LABORATORY*

**(33) NATHALY SWEENEY**

*RAPID WHOLE GENOME SEQUENCING (WGS) IMPROVES MANAGEMENT OF CRITICALLY ILL INFANTS WITH CONGENITAL HEART DISEASE*

**(34) LUCA VAN DER KRAAN**

*VALIDATION OF WHOLE GENOME SEQUENCING ASSAY USING NEXT GENERATION SEQUENCING FOR RADY CHILDREN'S INSTITUTE FOR GENOMIC MEDICINE*

## HOSPITAL MEDICINE

**(69) JULIA BEAUCHAMP-WALTERS**

*RADY'S HOMECARE ORDERSET: NO MORE GUESSES, NO MORE ERRORS! LET EMR WORK FOR YOU!*

**(80) MARIA HUANG**

*"IT'S NOT ME, IT'S YOU!" PHYSICIAN PERCEPTIONS ABOUT VIRAL TESTING IN BRONCHIOLITIS*

**(78) JEANNIE HUNG**

*PEDIATRIC OPIATE PRESCRIBING PRACTICES AT A LARGE METROPOLITAN CHILDREN'S HOSPITAL*

**(79) JEANNIE HUNG**

*USING AUTOMATED FACIAL RECOGNITION TO DISTINGUISH PAIN STATES IN YOUTH*

**(87) ERIN STUCKY-FISHER**

*BRONCHIOLITIS: DON'T SOUND THE ALARM!*

## HOST MICROBE SYSTEMS & THERAPEUTICS

**(91) AUSTIN CHIANG**

*GENOME-WIDE ANALYSIS OF THE TRANSCRIPTIONAL RESPONSE TO VIRUS INFECTION IN CHO CELLS*

## INFECTIOUS DISEASES

**(95) GRANT CAMPBELL**

*INHIBITION OF IAPs INDUCES AUTOPHAGOSOME DEPENDENT APOPTOSIS OF RESTING MEMORY CD4+ T CELLS LATENTLY INFECTED WITH HIV*

**(96) SARAH DABYDEEN**

*HIV GENE VARIANTS ALTERING CORECEPTOR BINDING & DRUG RESISTANCE PREDICT CNS PHENOTYPES*

**(98) PRATIMA RAWAT**

*HIV RNA INDUCED INFLAMMASOME ACTIVATION IMPAIRS AUTOPHAGIC CLEARANCE OF DAMAGED MITOCHONDRIA*

**(97) CARMEN TEODOROF**

*HIV-1 VIRAL PROTEINS INDUCE MITOCHONDRIAL FRAGMENTATION AND ALTER NEURONAL MITOPHAGY IN HIV-1 INFECTED BRAIN*

**(100) GANG ZHANG**

*PREFERENTIAL KILLING OF HIV-1 INFECTED CELLS THROUGH THE INDUCTION OF Na<sup>+</sup>/K<sup>+</sup>-ATPASE DEPENDENT AUTOPHAGY*

## INTENSIVE CARE

**(99) JOE TREISTER**

*CLINICAL COMPARISON OF KAWASAKI DISEASE SHOCK SYNDROME AND SEPTIC SHOCK IN THE PEDIATRIC INTENSIVE CARE UNIT*

## NEONATOLOGY

**(28) LAILA AKHMETOVA**

*FISHING IN TROUBLED WATERS: IN VIVO VALIDATION OF DISEASE-CAUSING GENES*

**(104) HAWRA ALSHAKHORI**

*UNTARGETED, LARGE-SCALE METABOLOMICS FOR NEONATAL DISEASE BIOMARKER DISCOVERY*

**(113) JEANNE CAROLL**

*THE ROLE OF HEDGEHOG INTERACTING PROTEIN IN HUMAN EMBRYONIC STEM CELL DIFFERENTIATION TO LUNG PROGENITORS*

**(105) AMY CHONG**

*CATCH YOUR BREATH: PILOTING AN INNOVATIVE NEONATAL RESUSCITATION APP THROUGH SIMULATION IN PERU*

**(114) FRAUKE DREES**

*NOVEL MECHANISMS OF HYPOXIA AND HEAT-STRESS PROTECTION IDENTIFIED THROUGH GENOME-WIDE ANALYSES IN*

**(118) JENNIFER FROHLICH**

*HYPOXIA-INDUCED EARLY HATCHING IN ZEBRAFISH EMBRYOS—A HIF-INDEPENDENT ENVIRONMENTAL TRIGGER?*

**(106) SANDRA LEIBEL**

*DERIVATION OF PATIENT-SPECIFIC ALVEOLAR TYPE II CELLS FROM SURFACTANT PROTEIN B DEFICIENT INDUCED PLURIPOTENT STEM CELLS*

**(107) AUDRA WISE**

*HELIOX ADJUNCT THERAPY FOR NEONATES WITH CONGENITAL DIAPHRAGMATIC HERNIA*

## NEPHROLOGY

**(108) ROBERT MAK**

*CLINICAL OUTCOMES AND RISK FACTORS IN ANTENATAL HYDRONEPHROSIS*

**(109) PETER YORGIN**

*EFFECTIVE UTILIZATION OF THE PHQ-9 FOR THE DETECTION OF DEPRESSION IN ADOLESCENTS AND YOUNG ADULTS WITH LUPUS NEPHRITIS: A QUALITY IMPROVEMENT STUDY*

## NEURO-ONCOLOGY

**(52) VALENTIN BARSAN**

*THE APPLICATION OF TUMOR MOLECULAR PROFILING IN PEDIATRIC NEURO-ONCOLOGY PATIENTS*

## NEUROSURGERY

**(53) MARK CALAYAG**

*POSTERIORLY DISPLACED AND LATERALIZED COMPONENTS OF CRANIO-PHARYNGIOMAS IN CHILDREN: CONSIDERATIONS IN MODIFYING CURRENT CLASSIFICATIONS*

**(54) MARK CALAYAG**

*CONSIDERATIONS IN RELATIONSHIP TO THE APPROACH FOR THE TREATMENT OF LATERALIZED POSTERIOR FOSSA TUMORS IN CHILDREN*

**(55) MARK CALAYAG**

*MIDDLE FOSSA APPROACH TO LATERALIZED PONTINE CAVERNOMAS IN CHILDREN*



**(56) MARK CALAYAG**

*ENDOSCOPIC MANAGEMENT OF THIRD VENTRICULAR CYSTS IN CHILDREN*

**(140) REID HOSHIDE**

*HYPOTHALAMIC INJURY DURING CRANIOPHARYNGIOMA RESECTIONS IN CHILDREN: A LEARNING CURVE*

**(141) REID HOSHIDE**

*ROBOT-ASSISTED ENDOSCOPIC THIRD VENTRICULOSTOMY*

**(142) JAVAN NATION**

*CSF LEAK RATE AFTER ENDOSCOPIC SKULL BASE TUMOR RESECTIONS IN CHILDREN: A SINGLE INSTITUTION EXPERIENCE*

## NURSING

**(76) MARY FAGAN**

*TEAM TRAINING IN THE PEDIATRIC LIFE SUPPORT PROGRAM: EFFECTS ON TEAMWORK AND QUALITY OF RESUSCITATIONS*

## ORTHOPEDICS

**(148) RAGHAV BADRINATH**

*INCREASED HIP INTRA-CAPSULAR PRESSURE DECREASES PERFUSION OF THE CAPITAL FEMORAL EPIPHYSIS IN A SKELETALLY IMMATURE PORCINE MODEL*

**(149) AILEEN CHANG**

*3D KNEE TROCHLEAR MORPHOLOGY BY MAGNETIC RESONANCE IMAGING: NORMAL VERSUS PATELLA INSTABILITY IN PEDIATRIC PATIENTS*

**(150) JERICHO HALLARE**

*TORSIONAL GROWTH MODULATION OF LONG BONES VIA OBLIQUE PLATING IN A RABBIT MODEL*

**(151) PETER NEWTON**

*CLINICAL OUTCOMES OF ANTERIOR SPINAL GROWTH TETHERING AND 3-DIMENSIONAL ANALYSIS OF THE*

## OTOLARYNGOLOGY

**(112) MATTHEW BRIGGER**

*LARYNGEAL STENOSIS IN CHILDREN: EPIDEMIOLOGY USING A POPULATION BASED APPROACH*

**(143) DANIELA CARVALHO**

*THE EFFECT OF POST OPERATIVE USE OF IBUPROFEN ON POST-TONSILLECTOMY HEMORRHAGE RATE OVER A 6-YEAR PERIOD*

**(144) WEN JIANG**

*OBESITY AND TONSILLECTOMY: CORRELATION BETWEEN BODY MASS INDEX AND INDICATIONS FOR SURGERY*

**(145) ALEXANDER MANTEGHI**

*THE DIFFICULT DUCT: A NOVEL APPROACH FOR INTRAOPERATIVE DUCTAL LOCALIZATION DURING SIALOENDOSCOPY*

**(146) ALEXANDER MANTEGHI**

*PEDIATRIC SEPTOPLASTY AND FUNCTIONAL SEPTORHINOPLASTY: A QUALITY OF LIFE OUTCOME STUDY*

**(65) JUSTIN WILSON**

*RARE EXTRAMEDULLARY MANIFESTATION OF ACUTE LYMPHOBLASTIC LEUKEMIA AS A SINONASAL MASS*

## PHARMACOLOGY AND DRUG DISCOVERY

**(3) ALISON COADY**

*SIGLEC RECEPTORS MODULATE THE INNATE IMMUNE RESPONSE TO FUNGAL PATHOGENS*

**(93) KATY PATRAS**

*TAMM-HORSFALL GLYCOPROTEIN REGULATES NEUTROPHIL RESPONSES IN THE URINARY TRACT*

**(6) ANGELICA Riestra**

*INVESTIGATION OF THE INNATE IMMUNE RESPONSE TO THE SEXUALLY TRANSMITTED PARASITE TRICHOMONAS VAGINALIS*

**(94) JOSH SUN**

*P2Y12 INHIBITION ENHANCES PLATELET-MEDIATED IMMUNE KILLING AGAINST METHICILLIN-RESISTANT STAPHYLOCOCCUS AUREUS*



## PHARMACY

(92) WILLIAM MURRAY

*CALCULATION OF VANCOMYCIN CLEARANCE AND AREA UNDER THE CURVE USING NON-STEADY STATE SERUM CONCENTRATIONS: A COMPARISON OF THE RELATIONSHIP BETWEEN*

## PLASTIC SURGERY

(130) SAMUEL LANCE

*SURGEON SPECIALTY AND RECONSTRUCTIVE OUTCOMES IN ALVEOLAR BONE GRAFTING FOR PATIENTS WITH ORAL CLEFTS*

(135) VIRIDIANA TAPIA

*QUALITATIVE METHODS IN THE DEVELOPMENT OF A BILINGUAL AND BICULTURAL QUALITY OF LIFE OUTCOMES MEASURE FOR PEDIATRIC PATIENTS WITH CRANIOFACIAL CONDITIONS*

## PULMONOLOGY

(111) PRITI AZAD

*MOLECULAR BASIS OF HYPOXIA INDUCED EXCESSIVE ERYTHROCYTOSIS*

(116) CELIA ESPINOZA

*DYNAMIC CHANGES OF DNA METHYLATION DURING DIFFERENT STAGES OF NORMAL MOUSE LUNG DEVELOPMENT*

(115) THU DUONG

*A HIGH-THROUGHPUT SINGLE-CELL CHROMATIN ACCESSIBILITY ASSAY AND ITS APPLICATION TO LUNG DEVELOPMENT*

(117) GUILLERMO FLORES-DELGADO

*DISTINCT EXPRESSION OF ENAC SUBUNITS IN CILIATED CELLS OF CONDUCTIVE AIRWAYS*

(119) MALLAREDY MADIREDDI

*DIFFERENTIAL EFFECTS OF NEW DRUGS ON  $Cl^-$  /  $HCO_3^-$  TRANSPORT FUNCTIONS OF CFTR IN NORMAL AND CF SWEAT DUCTS*

(120) AKM SHAMSUDDIN

*BICARBONATE SECRETION IN HUMAN NATIVE SMALL AIRWAYS CONTRIBUTE TO THE PATHOPHYSIOLOGY OF CYSTIC FIBROSIS*

(121) TZU-PIN SHENTU

*MESENCHYMAL STEM CELL-DERIVED EXTRACELLULAR VESICLES PROMOTE RESOLUTION OF PULMONARY FIBROSIS*

(122) TSERING STOB DAN

*YOCYTE-SPECIFIC ENDOTHELIN RECEPTOR TYPE B KNOCKOUT MICE CONFERS CARDIAC RESISTANCE TO EXTREME HYPOXIA*

(123) SIMON WONG

*THY-1 INTERACTION WITH FAS IN LIPID RAFTS REGULATES FIBROBLAST APOPTOSIS AND LUNG INJURY RESOLUTION*

(124) SIMON WONG

*LOSS OF FIBROBLAST THY-1 IN A MODEL OF PROGRESSIVE PULMONARY FIBROSIS*

(125) HANG YAO

*PHI REGULATION IN ASTROCYTES DERIVED FROM HUMAN SUBJECTS WITH MONGE'S DISEASE*

(126) YAN ZHANG

*E3 UBIQUITIN LIGASE FBXW7 CONTROLS AIRWAY CELL FATE*

(110) HUIWEN ZHAO

*ALTERED MITOCHONDRIAL STRUCTURE AND FUNCTION IN IPSC-DERIVED NEURONS IN SUBJECTS WITH CHRONIC MOUNTAIN SICKNESS*

## SURGERY

(127) PETER ABRAHAM

*POSTOPERATIVE COMPLICATIONS OF CRANIOSYNOSTOSIS REPAIR: IMPACT OF TIMING AND CONCURRENT FRONTO-ORBITAL ADVANCEMENT*

(128) MICHAEL BRANDEL

*QUANTIFYING INTRACRANIAL VOLUME CHANGE ACHIEVED BY DISTRACTION OSTEOGENESIS FOR CRANIOSYNOSTOSIS*

(71) LILLIA CHERKASSKIY

*FACTORS INFLUENCING PEDIATRIC ADHERENCE TO OUTPATIENT ANTIBIOTICS: INSIGHTS FROM RESEARCH IN MEDICINE AND PSYCHOLOGY*

(134) CHRISTOPHER REID

*A NATIONAL STUDY OF PROVIDER CHARACTERISTICS IMPACTING PRIMARY CLEFT PALATE REPAIR*

**(136) ANDREW WANG**

*APPENDICOLITHS CONTAIN CALCIUM STEARATE CRYSTALS AND BOTH HUMAN AND MICROBIAL PROTEINS*

**(137) ANDREW WANG**

*GASTROSTOMY TUBE OUTCOMES IN PEDIATRIC PATIENTS WITH CONCOMITANT INTRA-ABDOMINAL CATHETERS*

**(138) ERIN WARD**

*PILONIDAL DISEASE MANAGEMENT AT CHILDREN'S HOSPITALS: CHARACTERIZING DEMOGRAPHICS, TREATMENT, AND RETURNS TO THE SYSTEM*

**(139) ERIN WARD**

*SKATEBOARDING RELATED ACCIDENTS HAVE AN UNDERAPPRECIATED RATE OF TRAUMATIC BRAIN INJURIES*

## UROLOGY

**(129) DIANA CARDONA-GRAU**

*PATIENT PORTAL USAGE IN PEDIATRIC UROLOGY: IS IT MEANINGFUL USE TO EVERYONE?*

**(131) KELLY NAST**

*SCISSOR URETEROCELE UNROOFING ENDOSCOPIC TECHNIQUE: A NOVEL TECHNIQUE FOR MINIMALLY INVASIVE URETEROCELE TREATMENT*

**(132) KELLY NAST**

*DECREASING OPERATING ROOM COSTS VIA REDUCTION OF SURGICAL INSTRUMENTS*

**(133) KELLY NAST**

*PATIENT'S PERCEPTION OF QUALITY OF LIFE WHILE USING THE VESICOSTOMY BUTTON*

## HEMATOLOGY/

**(51) PAULA ARISTIZABAL**

*ADDRESSING PEDIATRIC CANCER DISPARITIES IN THE U.S-MEXICO BORDER REGION: THE RADY CHILDREN'S HOSPITAL SAN DIEGO CROSS-BORDER NEURO-ONCOLOGY PROGRAM*

**(57) JENNIFER ELSTER**

*PILOT TOLERABILITY STUDY COMBINING POLY-ICLC ADJUVANT WITH A CANCER GERMLINE ANTIGEN DENDRITIC CELL VACCINE TO TREAT RELAPSED PEDIATRIC SOLID TUMORS*

**(58) ANUSHA GANESAN**

*TISSUE-RESIDENT MEMORY FEATURES ARE LINKED TO THE MAGNITUDE OF CYTOTOXIC T CELL RESPONSES IN HUMAN CANCER*

**(59) SARA HAKIM**

*EFFICACY OF THE PAN-FGFR INHIBITOR AZD4547 IN NEUROBLASTOMA AND RHABDOMYOSARCOMA TUMOR CELLS THROUGH LEVELS AND AUC CALCULATIONS*

**(60) KSENYA SHILAKHSTISAVA**

*IMPROVING THE APPROACH FOR VITAMIN D DIAGNOSTIC TESTING AND SUPPLEMENTATION IN NEWLY DIAGNOSED PEDIATRIC CANCER PATIENTS AT RADY CHILDREN'S HOSPITAL - SAN*

**(62) KAVITHA SUBRAMANIAM**

*CONFIDENCE IN SURVIVORSHIP CARE IN PARENTS OF YOUNG CANCER SURVIVORS: DOES HEALTH LITERACY MATTER?*

**(63) DIVYA SUBRAMONIAM**

*EFFECTS OF THE MULTI KINASE INHIBITOR REGORAFENIB IN NEUROBLASTOMA IN VITRO*

**(66) VICTOR WONG**

*BRINGING PRECISION MEDICINE TO PEDIATRIC ONCOLOGY IN COLLABORATION WITH UCSD MOLECULAR TUMOR BOARD VAGINALIS*

**(11) MATTHEW WORTHAM**

*CONTROL OF BETA CELL METABOLISM AND INSULIN SECRETION BY THE HISTONE DEMETHYLASE LSD1*

# HEMATOLOGY/

**(51) PAULA ARISTIZABAL**

*ADDRESSING PEDIATRIC CANCER DISPARITIES IN THE U.S-MEXICO BORDER REGION: THE RADY CHILDREN'S HOSPITAL SAN DIEGO CROSS-BORDER NEURO-ONCOLOGY PROGRAM*

**(57) JENNIFER ELSTER**

*PILOT TOLERABILITY STUDY COMBINING POLY-ICLC ADJUVANT WITH A CANCER GERMLINE ANTIGEN DENDRITIC CELL VACCINE TO TREAT RELAPSED PEDIATRIC SOLID TUMORS*

**(58) ANUSHA GANESAN**

*TISSUE-RESIDENT MEMORY FEATURES ARE LINKED TO THE MAGNITUDE OF CYTOTOXIC T CELL RESPONSES IN HUMAN CANCER*

**(59) SARA HAKIM**

*EFFICACY OF THE PAN-FGFR INHIBITOR AZD4547 IN NEUROBLASTOMA AND RHABDOMYOSARCOMA TUMOR CELLS THROUGH LEVELS AND AUC CALCULATIONS*

**(60) KSENYA SHILAKHSTISAVA**

*IMPROVING THE APPROACH FOR VITAMIN D DIAGNOSTIC TESTING AND SUPPLEMENTATION IN NEWLY DIAGNOSED PEDIATRIC CANCER PATIENTS AT RADY CHILDREN'S HOSPITAL - SAN*

**(62) KAVITHA SUBRAMANIAM**

*CONFIDENCE IN SURVIVORSHIP CARE IN PARENTS OF YOUNG CANCER SURVIVORS: DOES HEALTH LITERACY MATTER?*

**(63) DIVYA SUBRAMONIAM**

*EFFECTS OF THE MULTI KINASE INHIBITOR REGORAFENIB IN NEUROBLASTOMA IN VITRO*

**(66) VICTOR WONG**

*BRINGING PRECISION MEDICINE TO PEDIATRIC ONCOLOGY IN COLLABORATION WITH UCSD MOLECULAR TUMOR BOARD VAGINALIS*

**(11) MATTHEW WORTHAM**

*CONTROL OF BETA CELL METABOLISM AND INSULIN SECRETION BY THE HISTONE DEMETHYLASE LSD1*