

CURRICULUM VITAE

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Education:

1982 – 1984 Human Sciences: Oxford University, Mansfield College, Oxford, England
1984 – 1987 B.A.: History, Philosophy and Social Studies of Science and Medicine,
University of Chicago, Chicago, IL
1990 – 1994 M.D.: University of Chicago, Pritzker School of Medicine, Chicago, IL
1994 – 1995 Internship, Naval Medical Center, San Diego CA
1995 - 1997 Residency, Pediatrics Naval Medical Center, San Diego CA
1996 – 1997 Chief Residency: Pediatrics, Naval Medical Center, San Diego CA
2001 – 2004 Fellowship: Dysmorphology and Medical Genetics with additional certificate
in Teratology, University of California, San Diego, CA. Mentor, Kenneth
Lyons Jones

Licensure and Certification:

California Medical License, Active
American Board of Pediatrics, Active
Drug Enforcement Administration, Controlled Substance Registration
Certification
Pediatric Advanced Life Support Certification
Basic Life Support Certification

Employment:

7/1997 – 6/1999 Naval Hospital, Guam, Attending Physician
6/1999 – 6/2001 Naval Medical Center, San Diego, Attending Physician
6/2004 – 7/2006 University of California, San Diego, Attending Physician
7/2006 – 8/2007 Children's Specialists of San Diego, Kaiser Permanente, Attending Physician
9/2007 – 3/2008 Kaiser Permanente, San Diego, Attending Physician
4/2008 – 9/2009 Children's Specialists of San Diego
9/2009 – 11/2010 University of California, San Diego, Associate Professor of Pediatrics.
Divisions of Dysmorphology and Hospital Medicine
11/2010- 2014 University of California, San Diego, Professor of Pediatrics. Divisions of
Dysmorphology and Hospital Medicine
07/2014-present University of California San Diego Professor of Medicine
07/2014-present Director and Clinical Chief, Division of Medical Genetics
03/2015-present University of California San Diego, Professor of Neurosciences
08/2012- 2016 Assistant Dean, Continuing Medical Education, Department of Medical
Education
7/2013-present Faculty Member, UCSD International CORE (Clinical Observership and
Research Education Program)
1/2014-present Medical Board of California, Expert Reviewer
1/2014-present Professor, UC San Diego Neurosciences and King Fahad Specialist Hospital
–Dammam University Kingdom of Saudi Arabia Faculty Exchange.
5/2014-present Medscape/E-Medicine Chief Editor, Genetics and Genomics

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Professional and University Service:

- 1997 – 1999 Naval Hospital Guam, Executive Committee of the Medical Staff, Elected Member
- 1998 – 2001 Secretary, Uniformed Services Pediatric Services Section, American Academy of Pediatrics
- 1999 – 2001 Faculty and Resident Advisor, Naval Medical Center San Diego
- 1999 – 2001 Information Management Executive Committee, Naval Medical Center San Diego
- 1999 – 2001 Faculty, UCSD/NMCS D Dyson Initiative for Community Pediatrics
- 1999 – 2001 Community Pediatrics Coordinator, NMCS D
- 1999 – 2001 Assistant Program Director, Transitional Intern Program Naval Medical Center San Diego
- 2001 – 2002 Planning Committee, Uniformed Services Pediatric Services Annual Meeting, San Diego CA 2002
- 2004 – 2008 Kaiser Regional Genetics Committee
- 2008 – 2009 Planning and Curriculum Committee, UCSD/RCHSD Pediatric Dermatology for the Practitioner, Apr 17-18, 2009
- 2008 – 2015 Executive Committee of the Medical Staff, Rady Children’s Hospital San Diego
- 2008 – 2015 RCHSD Quality Improvement Committee
- 2008 – 2015 RCHSD Continuing Medical Education Committee
- 2008 – 2015 Chair, Planning and Curriculum Committee, American Academy of Pediatrics and RCHSD 2009-2015 Advances in the Practice of Pediatrics Conference
- 2009 – 2014 Faculty Advisor, University of California, San Diego, Pediatric Residency Program
- 2009 – 2015 RCHSD/UCSD Pediatric Grand Rounds Planning Committee
- 2009 – 2015 Course Director, UCSD/RCHSD Summer Grand Rounds Series
- 2009 – 2015 Planning and Curriculum Committee, UCSD/RCHSD Evolving Concepts in the Management of Complex Congenital Heart Disease, 2010, 2013, 2015
- 2010 – 2015 Rady Children’s Hospital-San Diego Continuing Medical Education Curriculum and Strategic Planning Committee

Professional Memberships:

- 2001 – 2015 Western Society of Pediatric Research
- 2001 – American College of Medical Genetics
- 2002 – 2007 American College of Human Genetics
- 1998 – present American Academy of Pediatrics
- Section on Genetics and Birth Defects
 - Section on Hospital Medicine
 - Section on Complex Care

Honors and Awards:

- 1983 Secretary, Oxford University Basketball Team
- 1983 “Blue” Varsity Letter Basketball, Oxford University
- 1985, 1986 Varsity Letter, Basketball University of Chicago
- 1997, 1999 Navy Achievement Medal, 2 awards
- 1997 Meritorious Unit Commendation
- 1998 Humanitarian Service Award
- 2001 Navy Commendation Award
- 2002 Young Investigator Award, Teratology Society
- 2002 David W Smith Pediatric Trainee Award, Western Society for Pediatric Research
- 2002 Mead Johnson Fellowship Award, Western Society for Pediatric Research
- 2002 Navy Meritorious Unit Commendation
- 2004 Fellow Award, Teratology Society
- 2004 Fellow Award, DWS Workshop on Malformations and Morphogenesis

2005	Mead Johnson Young Investigator Award, Western Society for Pediatric Research
2010	Kaiser Excellence in Teach Award Nominee
2013	UC San Diego Academy of Clinical Scholars Nomination
2014	Saudi Arabian Cultural Mission-Department of Medical and Health Science Program

Contracts and Grants:

1. Principle Investigator: "Maternal Diabetes alters Sonic Hedgehog expression in Sonic Hedgehog knockout mice." University of California, San Diego Academic Senate S03057. (Completed)
2. Co-investigator: "A Prospective, multicenter study of bronchiolitis admissions: etiology and disposition (BABY BREATHE STUDY). NIH U01A 167693. CSSD Division of Pediatric Hospital Medicine, San Diego, CA. (Completed)
3. Producer and Principle Investigator: Immunization Practices among Military Providers, Practices and Education Program. Continuing Education Program for Armed Forces Pediatrics. \$20,000 Grant Merck. (Completed June 2000)
4. Site Principle Investigator: Efficacy of MMR compared to MMRV. (Completed 2001)
5. Site Principle Investigator: "Case Control Surveillance for Birth Defects in Relation to Environmental Exposure," Naval Medical Center, San Diego CIP #S-01-076. (No longer involved)
6. Site Principle Investigator: "Case Control Surveillance for Birth Defects in Relation to Environmental Exposure," Naval Hospital, Camp Pendleton CIP #S-01-076.
7. Member of Dysmorphology Core "OTIS Autoimmune Diseases in Pregnancy Project." University of California, San Diego FUNDING: Organization of Teratology Information Specialists (OTIS), Amgen. Study ID numbers: 20040246 Clinical Trials Identifier - NCT01017016. Active
8. Dysmorphology Core Member, "Ready Set Go NIH 5U24AA014815-10." National Institute of Alcohol Abuse and Alcoholism" and University of California, San Diego.
9. Principle Investigator: "Keith Vaux / Kelly Frazer Kabuki Syndrome study" RCHSD 12401. March 23, 2010. (Completed)
10. Co-investigator: West Coast Kawasaki Disease Epidemiology Consortium. UCSD 110306. (Completed)
11. Co-Principle Investigator: CIRM RFA 12-02 (CIRM Tissue Collection for Disease Modeling Awards) Grant IT1-06611, UCSD IRB Project 090293. December 2013-June 2016
12. Clinical Director NIH 7R01 MH076431-07 Sebat (PI) 9/30/05 - 11/30/2017. The role of germline mutation and parental age in Autism Spectrum Disorders.
 - a. The major goals of this project are: Identifying hotspots for germline mutation, examining intrinsic and extrinsic factors that influence mutation rates, and determining the role of hotspots in disease risk.
 - b. Role: Recruitment, phenotyping, clinical evaluation, results confirmation.
13. Clinical Director: SFARI 275724 Sebat (PI) 8/1/2013 – 7/30/2016. Simons Foundation Mutation Hotspots in Non-coding DNA and the Missing Heritability of ASD.
 - a. The major goal of this project is the identification of novel autism risk factors by targeted high throughput sequencing of non-coding DNA in autism families.
 - b. Role: Clinician, patient recruitment, enrollment, phenotyping and confirmation of research findings.
14. Principle Investigator: Autism and Neurogenetics Fund UCSD F. April 2015-ongoing. Support

External Professional Activities

2002 - 2004	President and CEO, Alpha Medical Associates (Imaging Care Center)
2004 – 2011	Advisor, Health Disk LLC (Health IT start-up)
2005 – 2011	Consultant, Proficio LLC (Health IT consulting)

2008 – present EPIC Systems, National Genomics Committee

Reviewer

2006 - present Reviewer, American Journal of Medical Genetics

2013, 2014 Reviewer, Society for Hospital Medicine – International Conference Abstracts

2016,2017 Reviewer – Cold Spring Harbor Molecular Case Studies

Presentations and Lectures:

1. "Overview of a Pediatric Transport Service," Pediatric Transport Medicine Conference, San Diego CA, 1996.
2. "Management of Pediatric Airway During Transport," Pediatric Transport Medicine Conference, San Diego, CA, 1996.
3. "Conotruncal Heart Lesions, History and Future Direction," Chief Resident Lecture, Naval Medical Center, San Diego Grand Rounds, 2007.
4. "Back to Sleep Program," Guam Department of Public Health, Guam, USA, 1998.
5. "Asthma Care," Guam Department of Public Health, Guam, USA, 1998.
6. "Multiple Circumferential Constriction Rings with Lineal Creases Over the Tibia, Mental Retardation and Autosomal Recessive Inheritance," DWS Workshop on Morphogenesis, San Diego, CA, August 2001.
7. "Cyclophosphamide, Methotrexate and Cytarabine Embryopathy: Is Apoptosis the Common Pathway?" Platform Presentation, Western Society for Pediatric Research, Carmel, CA, January 2002.
8. "Cyclophosphamide Embryopathy," Platform Presentation, Teratology Society Meeting, Scottsdale, AZ, Student Award Winner, February 2002.
9. "Dysmorphology and Genetics in the Community Nursery," 36th Annual Uniformed Services Pediatric Seminar, American Academy of Pediatrics, San Diego, CA 2002.
10. "The Fetal Alcohol Syndrome," San Diego County Public Health Nursing, Annual Meeting, San Diego, CA, 2003.
11. "CHARGE Association," Perinatal Care Continuing Education Conference, Naval Medical Center, San Diego, CA, 2003.
12. "Developmental Outcome in Kabuki Syndrome," Platform Presentation, DWS Workshop on Morphogenesis, Greenville, SC, August 2003.
13. "The Neonatal Phenotype of Kabuki Syndrome," Platform presentation, DWS Workshop on Morphogenesis, Greenville, SC, August 2003.
14. "Expanded Newborn Screening Program, Disparity Among Military Dependents," World Congress on Disabilities, Orlando, FL, September 19, 2003.
15. "Placental Abnormalities associated with Misoprostol Administration," Platform Presentation, Teratology Society Annual Meeting, Vancouver, British Columbia, Canada, Student Award Winner, June 30, 2004.
16. "Abnormal Pax1 expression is a factor in vertebral and rib anomalies seen in VATER association," Platform Presentation, DWS Meeting, Snowbird, Utah, August 18-21, 2004.
17. "Placental Anomalies in Artificial Reproductive Technology Conceived Pregnancies." Platform Presentation, DWS meeting. Snowbird, Utah. August 18-21, 2004.
18. "Diabetic Embryopathy," Pediatric Grand Rounds, Tripler Army Medical Center, HI, March 22, 2004.
19. "Genetic causes of IUGR," Pediatric Grand Rounds, Tripler Army Medical Center, HI, March 24, 2004.
20. "PAX 1 Expression is abnormal in VATER syndrome," Pediatric Pathology Grand Rounds, University of California, San Diego, May 19, 2004.
21. "Genetics in the Newborn Nursery," Southern California Association of Neonatal Nurses (SCANN), Annual Meeting, UCSD, March 2005.
22. "Ziprasidone Embryopathy: Are the defects caused by pharmacologically induced fetal arrhythmias?" Platform Presentation, Western Society for Pediatric Research, Carmel, CA. Young Investigator Award Winner, January 2005.

23. "Conjoined Twins: Fission or fusion, could the answer be so simple?" Platform Presentation, Judy Hall Symposium of Congenital Anomalies and Developmental Mechanisms, Carmel, CA, February 5, 2005.
24. Left-Right Axis Defects in Infants of Women with Diabetes: Further Evidence of the Link between Maternal Diabetes and Sonic Hedgehog," DWS Workshop on Morphogenesis, Vancouver, BC, August 2005.
25. "Genetic Conditions Incompatible with Life," Continuing Medical Education Conference, Southern California Association of Neonatal Nurses (SCANN), San Diego Hospice and Palliative Care, May 2006.
26. "Capillary Malformation, Arterial Venous Malformation Syndrome," Platform Presentation, DWS Workshop on Morphogenesis, September 2006.
27. "Development of the Vascular System," Cedar Sinai Genetics Grand Rounds, Los Angeles, CA, December 2006.
28. "Recognition, Management and Etiology of Pediatric Seizures," Pediatrics and Hospital Medicine Fellowship Core Curriculum, October 8, 2008.
29. "The Technology Dependent Child," Pediatrics and Hospital Medicine Fellowship Core Curriculum, April 15, 2009.
30. "Evidence Based Medicine and Leading a Health Care Team," UCSD Pediatric Fellowship Core Curriculum, San Diego, CA, October 10 2009.
31. "New Genetics and Personalized Medicine," World Congress on Disabilities, Jacksonville, FL, November 19, 2009.
32. "Genetics Primer," UCSD Pediatric Resident Core Curriculum, San Diego, CA, February 17, 2010.
33. "Common Syndromes and Genomic Sciences: How genetics will change our understanding of human disease," Southern California Association of Neonatal Nurses (SCANN), San Diego, CA, May 10, 2010.
34. "Trisomy 13 and 18, a Review and Discussion," RCHSD Ethics Forum, San Diego, CA, November 6, 2010.
35. "Genetic Disorders of Connective Diseases," UCSD Pediatric Resident Core Curriculum, San Diego, CA, November 19, 2010.
36. "Recognition, Management and Etiology of Pediatric Seizures," Pediatrics and Hospital Medicine Fellowship Core Curriculum, San Diego, CA, 2011.
37. "The Technology Dependent Child," Pediatrics and Hospital Medicine Fellowship Core Curriculum, San Diego, CA, 2011.
38. "Evidence Based Medicine," UCSD Pediatric Fellowship Core Curriculum, San Diego, CA, 2011.
39. "Metabolic Emergencies," Children's Hospital Emergency Transport Team Core Lecture Series, San Diego, CA, February 8, 2011.
40. "Metabolic Emergencies," Children's Hospital Emergency Transport Team Core Lecture Series, San Diego, CA, November 8, 2011.
41. "Middle East Siblings with Complex Genetic Defects," RCHSD Patient Care Operations Council, November 8, 2011.
42. "Evidence Based Medicine," Pediatrics and Hospital Medicine Fellowship Core Curriculum, November 9, 2011.
43. "Seizures" and "Caring for the Medically Complex Child," Pediatrics and Hospital Medicine Fellowship Core Curriculum, San Diego, CA, November 9, 2011.
44. "Update on Genetic Testing and its impact on Patient Care," Pediatric Grand Rounds at Scripps Mercy Hospital, March 15, 2012.
45. "Evidence Based Medicine," UCSD Pediatric Fellowship Core Curriculum, San Diego, CA, 2012.
46. "The Technology Dependent Child," Pediatrics and Hospital Medicine Fellowship Core Curriculum, San Diego, CA, 2012.
47. "Undiagnosed Neurogenetic Diseases," UCSD Pediatric Resident Core Curriculum, San Diego, CA, February 4, 2013.
48. "Undiagnosed Neurogenetic Diseases", Pediatric Resident Core Conference, San Diego, CA, February 4 2013.

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