

**BIOGRAPHICAL SKETCH**

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NAME Puck, Jennifer M.	POSITION TITLE Professor of Immunology, Dept. of Pediatrics, UCSF
eRA COMMONS USER NAME jenpuck	

EDUCATION/TRAINING (Begin with baccalaureate or other initial professional education, such as nursing, and include postdoctoral training.)

INSTITUTION AND LOCATION	DEGREE	YEAR(s)	FIELD OF STUDY
Harvard University, Cambridge, MA	B.A. cum laude	1971	Biochemistry
Harvard Medical School, Boston MA (Harvard-MIT Program in Health Sciences & Technology)	M.D.	1975	Medicine

**A. Positions and Honors.****Employment**

- 1975-78 Pediatric Resident, Infectious Diseases Fellow, St. Louis Children's Hosp., Wash. U, St. Louis, MO  
 1978-81 Pediatric Infectious Diseases & Immunology Fellowships: Influenza Research Center & Depts. of Pediatrics and Microbiology, Baylor College of Medicine, Houston, TX  
 1981-84 Instructor, Dept. of Pediatrics and Microbiology/Immunology, Baylor College of Medicine  
 1984-91 Assistant Professor, Dept. of Pediatrics, Univ. of Pennsylvania Sch. of Medicine, Philadelphia, PA  
 1991-93 Associate Professor, Depts. of Pediatrics and Human Genetics, Univ. of PA, Philadelphia, PA  
 1993-2006 Senior Staff Physician, W. G. Magnusen Clinical Center, NIH, Bethesda, MD  
 1993-2006 Head, Immunologic Disease Section, National Human Genome Research Inst (NHGRI), NIH  
 1994-97 Associate Chief, Laboratory for Gene Transfer, NHGRI, NIH  
 1997-2006 Chief, Genetics and Molecular Biology Branch, NHGRI, NIH  
 1998-2006 Director, CLIA Certified Molecular Diagnostic Laboratory in Immunologic Genetics, ID#21D0940018  
 2006- Professor of Immunology, Dept. of Pediatrics, UCSF, San Francisco  
 2006- Site Director, Pediatrics, Clinical Research Center, Clinical & Translational Sciences Inst., UCSF  
 2006- Investigator, Institute for Human Genetics, UCSF

**Selected Experience and Professional Service**

- NIH:** 1998-2001 Howard Hughes Medical Institute-NIH Research Scholars Program Advisory Committee  
 1998-2006 Advisory Board, Pharmacology Research Associate Training (PRAT) Program, NIH  
 1998-2006 Founder and Chair, NHGRI Institutional Review Board, NIH  
 1999-2006 NIH Human Subjects Research Advisory Committee (HSRAC)  
 2002-2006 NIH Clinical Research Training Program Advisory Committee  
**UCSF:** 2006- Advisory Committee, Pediatric Clinical Research Center, CTSI, UCSF  
 2006- Immunology Program Steering Committee, Biomedical Sciences Graduate Program, UCSF  
 2007- Course Director, Ethics and Responsible Conduct of Research, UCSF Biomedical Sciences  
**Nat'l:** 1996-99 Program Committee, American Society for Human Genetics  
 1998-2004 Basil O'Connor Advisory Committee, March of Dimes Foundation  
 2000-2007 Burroughs-Wellcome Fund Advisory Committee for Clinical Scientist Awards in Translational Research; Co-Chair, 2001, 2002, 2004, 2005  
 2002-04 DHHS Health Resources and Services Admin, Maternal & Child Health Genetic Services Workgroup on Uniform Criteria for Newborn Screening  
 2003- US Immunodeficiency Network Steering Committee; Grant Review Committee Co-chair  
 2004- Medical Advisory Committee, Immune Deficiency Foundation, U. S.  
 2007- Chair, NIH Clinical & Translational Sciences Award Nat'l Pediatrics Oversight Committee

**Selected Honors:**

- 1984-89 Physician Scientist Award, NIH  
 1991 Election to Society for Pediatric Research  
 1992 Election to American Society for Clinical Investigation  
 1997 Election to Association of American Physicians  
 1998 NHGRI Director's Award for Studies Defining Autoimmune Lymphoproliferative Syndrome  
 1995, 97, 98, 99 NHGRI Director's Awards for Scientific Achievement and Outstanding Service  
 2008 Election to American Pediatric Society

**B. Selected publications (in chronological order, from >190).**

- Puck J**, Glezen W, Frank A, Six H. Protection of infants from influenza A infection by transplacental antibody. *J Infect Dis* 142:844-849, 1980.
- Greenberg S, Tabor L, Septimus E, Kohl S, **Puck J**, Bryan R. Computerized tomography in brain biopsy proven herpes simplex herpes encephalitis. *Arch Neurol* 38:58-59, 1981.
- Parker P, **Puck J**, Fernandez F. Cerebral infarction associated with *M pneumoniae*. *Pediatr* 67:373-375, 1981.
- Abramson S, **Puck J**, Rich R. Antigen presentation to human T lymphocytes. I. Different requirements for stimulation of hapten-modified cells versus cell sonicates. *J Exp Med* 154:1005-1015, 1981.
- Puck JM**, Rich RR. Regulatory interactions governing the proliferation of T cell subsets stimulated with pokeweed mitogen. *J Immunol* 132:1106-1112, 1984.
- Croce C, Isobe M, Palumbo A, **Puck J**, Ming J et al. The gene for the alpha-chain of the human T cell receptor is on the region of chromosome 14 involved in translocations and in inversions in T cell neoplasms. *Science* 227:1044-1047, 1985.
- Gold D, **Puck J**, Petty C, Cho M, Coligan J, Woody J, Terhorst C. Isolation of cDNA clones encoding the 20kD non-glycosylated polypeptide chain (T3-Epsilon) of the human T cell receptor/T3 complex. *Nature* 321:431-434, 1986.
- Puck J**, Nussbaum R, Conley ME. Carrier detection in X-linked severe combined immunodeficiency based on patterns of X chromosome inactivation. *J Clin Invest* 79:1395-1400, 1987.
- Conley ME, Lavoie A, Briggs C, Guerra C, **Puck J**. Non-random X chromosome inactivation in B cells from carriers of X-linked severe combined immunodeficiency. *Proc Nat Acad Sci USA* 85:3090-3094, 1988.
- Conley ME, **Puck J**. Carrier detection in typical and atypical X-linked XLA. *J Pediatr* 112:688-694, 1988.
- Puck JM**, Nussbaum RL, Smead DL, Conley ME. X-linked severe combined immunodeficiency: Localization within the region Xq13.1-q21.1 by linkage and deletion analysis. *Am J Hum Genet* 44:724-730, 1989.
- Puck J**, Krauss C, Puck S, Buckley R, Conley M. Prenatal test for X-linked severe combined immunodeficiency by analysis of maternal X-chromosome inactivation and linkage. *New Engl J Med* 322:1063, 1990.
- Puck J**, Stewart C, Nussbaum, R. Maximum likelihood analysis of T-cell X chromosome inactivation: Normal women vs. carriers of X-linked SCID. *Am J Hum Genet* 50:742, 1992.
- Puck J**, Deschênes S, Porter J, Dutra A, Brown C, Willard H. The IL-2 receptor gamma chain maps to Xq13.1 and is mutated in X-linked severe combined immunodeficiency, SCIDX1. *Hum Molec Genet* 2:1099, 1993.
- Puck J**, Pepper A, Bédard P-M, Laframboise R. Female germ line mosaicism as the origin of a unique IL-2 receptor  $\gamma$ -chain mutation causing X-linked severe combined immunodeficiency. *J Clin Invest* 95:895, 1995.
- Fisher G, Rosenberg F, Straus S, Dale J, Middleton L, Lin A, Strober W, Lenardo M, **Puck J**. Dominant interfering Fas gene mutations impair apoptosis in human autoimmune lymphoproliferative syndrome. *Cell* 81:935, 1995.
- Pepper A, Buckley R, Small T, **Puck J**. Two CpG hot spots in IL2RG. *Am J Hum Genet* 57:564, 1995.
- Hirschhorn R, Yang D, **Puck J** et al. Spontaneous *in vivo* reversion to normal of an inherited mutation in a patient with adenosine deaminase deficiency. *Nature Genet* 13:290, 1996.
- Flake A, Porada G, **Puck J**, Roncorolo M-G, Evans M, Johnson M, Abella E, Harrison D, Zanjani E. Treatment of XSCID by in utero transplantation of CD34 enriched bone marrow. *New Engl J Med* 355:1806, 1996.
- Whitwam T, Haskins M, Henthorn P, Kraszewski J, Kleiman S, Seidel N, Bodine D, **Puck J**. Retroviral marking of canine bone marrow: Long term, high level expression of human IL-2 receptor common gamma chain in canine lymphocytes. *Blood* 92:1565, 1998.
- Puck J**, Pepper A, Henthorn P et al. Mutation analysis of *IL2RG* in human X-linked SCID. *Blood* 89:1968, 1997.
- Infante A, Britton H, DiNapoli T, Middleton L, Lenardo M, Jackson C, Wang J, Fleisher T, Straus S, **Puck J**. The clinical spectrum in a large kindred with ALPS due to a Fas mutation. *J Pediatr* 133:629, 1998.
- Grimbacher B, Holland S, Gallin J, Greenberg F, Hill S, Malech H, Miller J, O'Connell A, **Puck J**. Hyper-IgE recurrent infection syndrome. *New Engl J Med*, 340:692, 1999.
- Jackson C, Hsu A, Fischer R, Wang J, Lenardo M, Straus S, **Puck J**. Autoimmune lymphoproliferative syndrome with defective Fas: Genotype influences penetrance. *Am J Hum Genet* 64:1002, 1999.
- Choi Y, Ramnath V, Chen A, Eaton A, Kleiner D, Erikson J, **Puck J**. Expression in transgenic mice of dominant interfering mutations of Fas: A model for human ALPS. *Clin Immunol*, 93:34, 1999.
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- Puck J.** Genetic linkage of hyper-IgE syndrome to chromosome 4. *Am J Hum Genet* 65:735, 1999.
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- Niemela J, **Puck J,** Fischer R et al. Efficient detection of thirty-seven new *IL2RG* mutations in human X-linked severe combined immunodeficiency. *Clin Immunol* 95:33, 2000.
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- Fanos J, **Puck J.** Family pictures: Growing up with a brother with X-linked. *Am J Med Genet* 98:57, 2001.
- Avilés Mendoza G, Seidel N, Otsu M, Anderson S, Simon-Stoos K, Herrera A, Hoogstraten-Miller S, Malech H, Candotti F, **Puck J,** Bodine D. Comparison of five retrovirus vectors containing the human IL-2 receptor gamma chain gene for their ability to restore T and B lymphocytes in the X-linked severe combined immunodeficiency mouse model. *Molecular Therapy* 3:565, 2001.
- Straus S, Jaffe E, **Puck J** et al. The development of lymphomas in families with ALPS with germline Fas mutations. *Blood.* 98:194, 2001.
- Bleesing J, Brown M, Straus S, Dale J, Siegel R, Johnson M, Lenardo M, **Puck J,** Fleisher T. Immunophenotypic profiles in the autoimmune lymphoproliferative syndrome (ALPS). *Blood* 98:2466, 2001.
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- Licht T, Haskins M, Henthorn P, Kleiman S, Bodine D, Whitwam T, **Puck J,** Gottesman M, Melniczek J. Drug selection with paclitaxel restores expression of linked interleukin-2 receptor gamma chain and multidrug resistance (MDR1) transgenes in canine bone marrow. *Proc Natl Acad Sci USA* 99:3123, 2002.
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- Renner ED, **Puck J,** Holland S, Schmitt M, Weiss M, Frosch M, Bergmann M, Davis J, Belohradsky B, Grimbacher B. Autosomal recessive hyper-IgE syndrome, a distinct disease. *J. Pediatr* 144:93, 2004.
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- Kellermayer R, Hsu A, Stankovics J, Balogh P, Hadzsiev K, Wojczek Á, Maródi L, Kajtár P, Kosztolányi G, **Puck J.** Genetic Aspects of Severe Combined Immunodeficiency. *J Hum Genet*, 51:495, 2006.

Principal Investigator/Program Director (Last, First, Middle):

- Vacek M, Schäffer A, Davis J, Fischer R, Dale J, Straus S, **Puck J**. HLA B44 is associated with decreased severity of autoimmune lymphoproliferative syndrome in patients with CD95 mutations (ALPS Ia). *Clin Immunol* 118:59, 2006.
- Zhu S, Hsu A, Vacek M, Zheng L, Schäffer A, Dale J, Davis J, Fischer R, Straus S, Boruchov D, Saulsbury F, Lenardo M, **Puck J**. Genetic alterations in caspase-10 can be causative or protective in autoimmune lymphoproliferative syndrome. *Hum Genet* 119:284, 2006.
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- Freeman A, Kleiner D, Nadiminti H, Davis J, Quezado M, Anderson V, **Puck J**, Holland S. Causes of death in hyper-IgE syndrome. *J Allergy Clin Immunol* 119:1234, 2007.
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- Butte M, Haines C, Bonilla F, **Puck J**. IL-7 receptor deficient SCID with a unique intronic mutation and post-transplant autoimmunity. *Clin Immunol* 125:159, 2007.
- Chinen J, Davis J, DeRavin S, Hay B, Hsu A, Linton G, Naumann N, Nomicos E, Silvin C, Ulrick J, Whiting-Theobald N, Malech H, **Puck J**. Gene therapy improves immune function in pre-adolescents with X-linked severe combined immunodeficiency. *Blood*, 110:67, 2007.
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- Puck JM** on behalf of The Working Group for Newborn Screening for SCID. Development of population-based newborn screening for SCID. *J Allergy Clin Immunol* 120:760, 2007.
- Vogt G, Chappier A, Bustamante J, Feinberg J, Picard C, Gineau L, Alcaïs A, Lamaze C, Mahlaoui N, **Puck JM**, de Saint Basile G, Khayat C, Mikhael R, Casanova J-L. Complementation of human pathogenic in-frame mutations with modifiers of N-glycosylation. *J Exp Med* 205:1729-1737, 2008.
- Lebet T, Chiles R, Hsu AP, Mansfield E, Warrington JA, **Puck JM**. Mutations causing severe combined immunodeficiency: detection with a custom resequencing microarray. *Genet in Med* 10:575-585, 2008.
- Shiow LR, Roadcap DW, Paris K, Watson SR, Grigorova IL, Lebet T, An J, Xu Y, Jenne CN, Föger N, Sorensen RU, Goodnow CC, Bear JE, **Puck JM**, Cyster JG. Coronin-1A is mutated in a thymic egress defective mouse strain and a T-B+**NK**+ SCID patient. *Nature Immunol*, In Press, 2008.

### C. Support

NIH 1UL1 RR024131 (PI: McCune)	09/30/06 - 06/30/11
Clinical and Translational Sciences Institute (CTSI) at UCSF	2.4 calendar months
Associate Program Director, Pediatric Clinical Research Center	
NIH 1UL1 RR024131 (PI: McCune; Supplement Leader: Puck)	09/01/08 – 08/31/09
CTSA Administrative Supplement	1.8 calendar months
Distributed biobanks to Support Research on Rare Disorders in CTSA's	
NIH RO3 HD 060311-01 (PI: Puck)	08/01/08 – 07/31/10
itle: Newborn screening for SCID in a high-risk population	
NIH NIAID AI 030070USIDNET Subcontract (PI: Puck)	03/01/07 - 02/28/09
T Cell receptor Excision Circles (TRECs): a Newborn Screening Tool for SCID	1.2 calendar months
MCH U32 MC000148 to UTHSC San Antonio (PI: B. Therrell)	05/01/08-05/31/10
HRSA, DHHS/NIH/CIT/SDP Subcontract to Dr. Puck	0.24 calendar months
Title: National Newborn Screening and Genetic Resource Center	
Pilot Newborn SCID Screening on Navajo Reservation	