
TAOSHENG HUANG, MD, Ph.D.

Professor (Tenure)

Director, Program of Mitochondrial Medicine
Associate Director, Molecular Diagnostic laboratory
Division of Human Genetics
Cincinnati Children's Hospital Medical Center
3333 Burnet Avenue
Building R, Room R1027, MLC 7016
Cincinnati, OH 45229-3039
Taosheng.huang@cchmc.org

EDUCATION

- 12/1987-12/1991** **Ph.D., Biomedical Science-**
Mount Sinai Medical School
New York, New York
- 07/1983-07/1986** **MS, Biochemistry**
The Third Military Medical College
Chongqing, China
- 10/1978-08/1983** **M.D. (Passed US Medical Board Exam step I, Step II and Step III)**
Fujian Medical College
Fuzhou, Fujian, China

ACADEMIC APPOINTMENTS:

- 09/12-** **Professor (Tenure)**
Division of Human Genetics
Cincinnati Children's Hospital Medical Center
Cincinnati, OH
- 09/12-** **Director**
Program of Mitochondrial Medicine
Division of Human Genetics
Cincinnati Children's Hospital Medical Center
Cincinnati, OH
- 09/12-** **Associate Director**
Molecular Diagnostic Laboratory
Division of Human Genetics
Cincinnati Children's Hospital Medical Center

Cincinnati, OH

- 09/11-** **Honorable Professor**
Peking Union Medical College
Beijing, China
- 2008-2012** **Associate professor (Tenure)**
Attending physician in Human Genetics/Metabolism
Department of Pediatrics/Genetics
College of Medicine
University of California, Irvine
Irvine, California
- 2008-2012** **Associate professor (joint appointment)**
Department of Developmental Biology
School of Biological Science
University of California, Irvine
Irvine, California
- 2008-2012** **Associate professor (joint appointment)**
Department of Pathology
College of Medicine
University of California, Irvine
Irvine, California
- 2008-2012** **Associate professor (joint appointment)**
Department of Ophthalmology
College of Medicine
University of California, Irvine
Irvine, California
- 09/2008- 12/2010** **Medical Director/Consultant**
Ambry Genetics
Aliso Viejo, California
- 2001-2012** **Attending physician**
Department of Pediatrics/Genetics
College of Medicine
University of California, Irvine
Irvine, California
- 2001-2012** **Director of Cardiovascular Genetic Clinic**
(National site of phenotype study for congenital cardiac defects)
Department of Pediatrics/Genetics
College of Medicine
University of California, Irvine

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- Irvine, California
- 2004-2012 Director**
MitoMed Molecular Diagnostic Lab
College of Medicine
University of California, Irvine
Irvine, California
- 2009-2012 Staff physician**
Department of Pediatrics/Genetics
Orange County Children's Hospital
Orange, CA
- 2007-2012 Consultant Physician**
Department of Pediatrics/Genetics
Long Beach Memorial Hospital
Long Beach, CA
- 07/01-07/08 Assistant Professor (Tenure track)**
Attending physician in Human Genetics/Metabolism
Department of Pediatrics/Genetics
College of Medicine
University of California, Irvine
Irvine, California
- 07/04-07/08 Assistant professor**
Department of Pathology
College of Medicine
University of California, Irvine
Irvine, California
- 07/2004- Medical Staff**
Long Beach Memorial Medical Center
2801 Atlantic Avenue
Long Beach, California
- 07/01-07/08 Assistant professor**
Department of Developmental Biology
School of Biological Science
University of California, Irvine
Irvine, California
- 07/99-07/01 Attending physician and Assistant in Medicine**
Children's Hospital
Harvard Medical School

Boston, Massachusetts

07/99-07/01 Instructor in Genetics
Harvard Medical School
Boston, Massachusetts

FELLOWSHIP & TRAINING:

12/97-07/99 Research Fellow
Seidman Laboratory
Howard Hughes Medical Institute
Harvard Medical School
Boston, Massachusetts

07/96-07/99 Clinical Fellow in Genetics and Metabolism
Children's Hospital
Harvard Medical School
Boston, Massachusetts

07/93-07/96 Intern and Resident in Pediatrics
Georgetown University Medical School
Children's Medical Center
Washington, DC

12/91-07/93 Postdoctoral Fellow
Jerome H. Holland Laboratory
American Red Cross
Rockville, Maryland

SELECT HONORS AND AWARDS:

1989 Predoctoral Travel Fellowship
American Society of Virology

1990 Third Place SCBA Poster Competition
Society of Chinese Biomedical Scientists in America

1996 Resident Research Award
Georgetown University Hospital
Children's Medical Center
Washington, DC

1996 Fellowship Training Grant
National Health Institute/Harvard Medical School
Boston, Massachusetts

1998 Farley Fellowship
Children's Hospital, Boston
Harvard medical School

2003	Boston, Massachusetts The Second Place of the Research Award College of Medicine University of California, Irvine Irvine, California
1999-2004	Clinical Associate Physician Award National Health Institute
2004	Junior Physician-Scientist Award Dean of College of Medicine University of California, Irvine Irvine, California

BOARD CERTIFICATION & LICENSING

1993	Educational Commission for Foreign Medical Graduates Certificate
1996	American Board of Pediatrics
1999	American Board of Medical Genetics Clinical Genetics
1999	American Board of Medical Genetics Clinical Molecular Genetics
2000-	California Medical License # A76405
2012-	Ohio Medical License # 35.121036
2002-	Clinical Genetics Molecular Biologist (CGMB) Lab Director, CA

MEMBERSHIPS:

2012-	Board Director, China California Heart Watch
2009-	Board Director, Association of Chinese Geneticist in America (ACGA)
2003-	Fellow, American College of Medical Genetics, 2003-
2005-	American Medical Association
2001-	Western Society for Pediatric Research
1999-	American Society of Human Genetics
1988-	American Association for Advancement of Science
1995-	American Academy of Pediatrics
1999-2001	Medical Director, Board Director, American Chinese Medical Association
1999-2001	Vice President of American Chinese Medical Association, Northwestern Chapter

Editorial Board

2013-	American Journal of Stem Cells (AJSC)
2015-	Journal of Ophthalmology and Visual Sciences (JOVS)
2015-	World Journal of Complex Medicine
2016-	Gavin Journal of Pediatrics
2016-	Henry Journal of Perinatology & Pediatrics
2016-	SOJ Ophthalmology
2016-	Ophthalmic Surgery

GRANTS AWARDED TO TAOSHENG HUANG AS PRINCIPAL INVESTIGATOR

ONGOING RESEARCH SUPPORT:

Cincinnati Children's Hospital Research Foundation Huang (PI) 09/01/2012-08/30/2017

Cincinnati Children's Hospital Research Foundation, CpG grant, 02/01/2015-01/30/2017

NIH-National Eye Institute (PI: T Huang) 1 R01EY026609-01

04/01/2016-

Molecular pathogenesis of SLC25A46 mutations in optic atrophy, axonal neuropathy, and cerebellar neurodegeneration

PENDING,

NIH-National Eye Institute (PI: T Huang)

04/01/2016-03/30/2021

iPS cell therapy of mitochondrial diseases

COMPLETED RESEARCH GRANTS

1. **NIH-National Eye Institute 1R01EY018876-01 (PI: T Huang)**
04/01/2008- 03/30/2014 Total direct cost: \$1,000,000
Title: Genetics studies of optic atrophy
2. **NIH-National Cancer Institute, 1R01CA121876 (PI: T Huang) 07/01/2007-06/30/2011, Total direct cost: \$750,000**
Title: Intracellular Network of TBX3 in breast cancer
3. **NIH-National Cancer Institute, 1R01CA121876 (PI: T Huang) 03/01/2008-06/30/2011**
Title: The role of TBX3 in Human Embryos Stem Cell Differentiation
Minority Supplement for Taraneh Esmailpour, \$250,000
4. **Genetic and iPS Cell studies (Private donation), \$50,000/year 2011-2013**
5. **NIH- National Cancer Institute RO3 07/012004-06/30/2006**
Title: Roles of TBX3 in Breast Cancer
Total direct cost: \$100,000

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6. **Howard Hughes Medical Research Program** 07/01/ 2001-06/30/2004
Title: Faculty Development
Total direct cost: \$180,000
 7. **Cancer Research Coordinating Committee** 07/01/2003-06/30/2004
Title: Molecular studies of TBX3 in Human Breast cancer
Total direct cost: \$50,000
 8. **Chao Family Comprehensive Cancer Center Seed grant 2004-2005,**
Title: Pilot study of TBX3 in human breast cancer
Total direct cost: **\$20,000**
 9. **Undergraduate Research Opportunities Program Award (PI: T Huang)**
Genetic study of cystinosis, \$500
Trainee: Stephenie Tse (summer, 2007)
 10. **Undergraduate Research Opportunities Program Award (PI: T Huang)**
Genetic study of optic atrophy, \$500
Trainee: Kimberly Nguyen (summer, 2007)
 11. **Susan Komen Breast Cancer Foundation BCTR-133006 (PI: T Huang),** 04/01/2006-03/30/2008
Title: Transgenic TBX3 Animal Model to study breast cancer
Total direct cost: \$240,000
 12. **Cystinosis Research Foundation CRF42443 (PI: T Huang)** 05/01/2007-04/30/2008
Title: Studies on mitochondrial function in cystinosis
Total direct cost: \$50,000
 13. **Cystinosis Research Foundation CRF42365 (PI: T Huang)** 07/01/2007-06/30/2010
Postdoctoral Fellowship (Postdoc: Sha Tang, Ph.D.; MD, Ph.D.)
Title: Molecular and Pathogenesis study of cystinosis
Total direct cost, \$87,000
 14. **Larry & Helen Hoag Foundation 5348-56608 (PI: T Huang)** 07/01/2005-12/30/2011
Title: Studies of the TBX5 targets
Total direct cost: \$300,000
 15. **Surber Foundation (PI: T Huang) 07-2011-09/12**
Title: Genetic study of mitochondrial diseases
Total direct cost: \$100,000

16. NIH-GCRC Seed Grant 03/01/2010-02/28/2011

Title: Genetic studies of optic atrophy

Total direct cost: \$15,000

17. Clinical Associate Physician Award (K23) M01RR0087 04/01/1999-03/30/2004

Title: The Molecular Basis of Phenotypic Variability in Holt-Oram Syndrome

Total direct cost \$650,000

INTRAMURAL RESEARCH SUPPORT

1. College of Medicine, UCI research award

07/01/2002-06/30/2/03

Title: Genetic study of birth defects

Total direct cost:

2. 2004-2005 Faculty Desktop Computing Initiative Allocation

College of Medicine, UCI

PROGRAM & TRAINING GRANT PARTICIPATION

NIH-National Cancer Institute (PI: Frank Meyskens)

Chao Family Comprehensive Cancer Center Grant (CA62203)

State of California- Institute of Degenerative Medicine

UCI Stem Cell Training Grant (PI: Peter Bryant)

CURRENT or PAST TRAINEES

NIH Training Grant (PI: L Marsh)

Developmental Biology training grant T32HD07029

Huang Lab Trainee supported by this grant: Tomasa Barrientos 07/01/2005-06/30/2006

K. Arima 03/01/2002-08/01/2002

NIH Training Grant (PI: Hung Fan)

Cancer Biology Training Grant (CA09054)

Huang Lab Trainee supported by this grant: Thin Aung Than 07/01/2005-06/30/2006

RESEARCH INTERESTS

The primary interest of the Huang lab is to study the molecular basis of genetic syndromes, to apply the discoveries from rare diseases to common conditions, and to develop treatments for genetic diseases. Currently, we are focusing on the following areas:

1. The Genetic basis of optic atrophy and inducible pluripotent stem cell (iPSC) therapy: We have worked with many families affected by autosomal dominant inherited optic atrophy. In collaboration with Dr. Arnold Star in the Department of Neurology, we find that the OPA1 gene mutation H445R causes loss of vision and hearing. OPA1 is encoded by the nuclear genome and functions in mitochondria. Using electrophysiological analysis, we find that this mutation causes asynchronous cochlear conduction, suggesting a novel mechanism of optic atrophy. To study the function of OPA1 and the molecular mechanisms of optic atrophy, we created a drosophila model of OPA1. We found that the dOpa1 somatic mutation caused an increase in reactive oxygen species (ROS) production and mitochondrial fragmentation. Our group shows that antioxidants can partially reverse the glossy eye phenotype, further suggesting that ROS plays an important role in cell death. Together, these results show that dOpa1 mutations cause cell loss by two distinct pathogenic pathways. This study provides novel insights into the pathogenesis of optic atrophy and demonstrates the promise of antioxidants as therapeutic agents for this condition. Recently, our lab is actively engaged in iPS cell therapy. We have successfully differentiated iPS cells into retinal ganglion cells.

2. Genetics of Mitochondrial Diseases: Mitochondria are the powerhouse of the cell. Over 90% of the energy required by the cell is produced in the mitochondria. MitoMed Molecular Diagnostics Lab is a CLIA-certified laboratory providing molecular diagnosis of mitochondria disease. Our researchers also work on the genetic causes of mitochondrial disorders. We have been using Exome sequencing, cell respiration assays, and mitochondrial functional assays to study the pathogenesis of mitochondria disease.

3. The role of TBX3 in breast cancer and human embryonic stem (hES) cells: TBX3 is a T-box transcription factor. Mutations of TBX3 cause Ulnar-Mammary syndrome, characterized by hypoplasia or absence of the mammary glands. Our lab is one of the first groups to show that overexpression of TBX3 plays an important role in breast cancer. Our study shows that TBX3 is overexpressed in primary breast cancer tissues. Mechanistically, we find that TBX3 interacts with HDACs to inhibit downstream target gene expression, such as p14ARF. In addition, we find that TBX3 regulates a large group of genes in breast cancer. Our current research aims to optimize the clinical relevance of this data working in parallel with animal and breast cancer tissues. Recently, we have also found that TBX3 plays a very important role in hES cell differentiation. This finding may further our understanding of TBX3 function.

4. Identifying the disease-causing gene associated with noncompaction of the ventricular myocardium (spongy heart): We are currently studying a family with balanced translocation with this condition, and are also performing a linkage study for a large pedigree with this disease. Currently, our lab is using next-generation sequencing technology to identify the new disease-causing gene.

5. Identification of the Disease-Causing Gene for Lenz Microphthalmia Syndrome (LMS) Using Whole Genome Exome Sequencing Technology: LMS is a rare condition characterized by small eyes/no eye and multiple congenital anomalies such as small brain and mental deficiency, abnormal ear, teeth,

digits, skeletal and/or genitourinary tract. In this study, we have used a very powerful technology, next generation sequencing, to search for the disease-causing gene in patients with LMS. Identification of disease-causing genes associated with LMS has significantly facilitated our understanding of this condition and can translate into clinical applications. Since LMS affects multiple organ systems, understanding the gene associated with LMS may open a window for the investigation of other common conditions and human development.

6. The intracellular pathway to study TBX5: TBX5 is a T-box transcription factor. Mutations of TBX5 cause Holt-Oram syndrome, characterized by congenital heart diseases and limb anomalies. By studying the intracellular network of TBX5, including the upstream transcription factors that control TBX5 expression and the cofactors that interact with TBX5 and its downstream targets, we anticipate identifying many genes associated with congenital heart disease, the most common congenital malformations in human which contribute significantly to the morbidity and mortality in the pediatric population.

TEACHING

<u>Date</u>	<u>Medical Student Course</u>	<u>Role</u>	<u>Enrollment</u>
2001	Medical Genetics	Lecturer	100
2002	Medical Genetics	Lecturer	100
2003	Medical Genetics	Lecturer	100
2004	Medical Genetics	Lecturer	100
2004	Medical Genetics	Course director	100
2005	Medical Genetics	Lecturer	100
2006	Medical Genetics	Lecturer	100
2007	Medical Genetics	Lecturer	100
2008	Medical Genetics	Lecturer	100
2009	Medical Genetics	Lecturer	100
2010	Medical Genetics	Lecturer	100

2003	Pedigree-taking exercise-Family Medicine Clerkship		10
2004	Pedigree-taking exercise-Family Medicine Clerkship		10
2005	Pedigree-taking exercise-Family Medicine Clerkship		10
2006	Pedigree-taking exercise-Family Medicine Clerkship		10

<u>Date</u>	<u>Ph.D. Graduate Course</u>	<u>Role</u>	<u>Enrollment</u>
2003	Pathogenesis of Disease PATH225	Lecturer	10
2004	Pathogenesis of Disease PATH225	Lecturer	10
2005	Pathogenesis of Disease PATH225	Lecturer	10
2006	Pathogenesis of Disease PATH225	Lecturer	10
2007	Pathogenesis of Disease PATH225	Lecturer	10
2008	Pathogenesis of Disease PATH225	Lecturer	10
2009	Pathogenesis of Disease PATH225	Lecturer	10
2010	Pathogenesis of Disease PATH225	Lecturer	10

2001	Patients-Doctor II Pediatrics Harvard Medical School		5
2000	Patients-Doctor II Pediatrics,Harvard Medical School		5
2000	Human Genetics Course, Harvard Medical School		7
2001	Human Genetics Course, Harvard Medical School		7
2006	Stem Cell Biology, Dev. Bio. 245	Lecturer	20
2008	Stem Cell Biology, Dev. Bio. 245	Lecturer	20
2007	Cancer Biology Journal Club 293A	Coordinator	30
2016	CCHMC Development & Disease course	Lecturer	35

<u>Date</u>	<u>Genetic Counseling Student Course</u>	<u>Role</u>	<u>Enrollment</u>
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2002	Human Molecular Genetics course 218	Lecturer	6
2003	Human Molecular Genetics course 218	Lecturer	6
2004	Human Molecular Genetics course 218	Lecturer	6
2005	Human Molecular Genetics course 218	Lecturer	6
2006	Human Molecular Genetics course 218	Lecturer	6
2007	Human Molecular Genetics course 218	Lecturer	6
2008	Human Molecular Genetics course 218	Lecturer	6
2009	Human Molecular Genetics course 218	Lecturer	6
2010	Human Molecular Genetics course 218	Lecturer	6
2002	Genetic Counseling Program, Metabolism	Lecturer	6
2003	Genetic Counseling Program, Metabolism	Lecturer	6
2004	Genetic Counseling Program, Metabolism	Lecturer	6
2005	Genetic Counseling Program, Metabolism	Lecturer	6
2006	Genetic Counseling Program, Metabolism	Lecturer	6
2007	Genetic Counseling Program, Metabolism	Lecturer	6
2008	Genetic Counseling Program, Metabolism	Lecturer	6
2009	Genetic Counseling Program, Metabolism	Lecturer	6
2003	Genetic Counseling Program Development malformation		6
2004	Genetic Counseling Program Development malformation		6
2005	Genetic Counseling Program Development malformation		6
2006	Genetic Counseling Program Development malformation		6
2007	Genetic Counseling Program Development malformation		6
2008	Genetic Counseling Program Development malformation		6
2009	Genetic Counseling Program Development malformation		5
2010	Genetic Counseling Program Development malformation		5
2011	Genetic Counseling Program Development malformation		6
2012	Genetic Counseling Program Development malformation		6
2014	Genetic Counseling Program at CCHMC		30

<u>Date</u>	<u>Resident and Fellowship Course</u>	<u>Role</u>	<u>Enrollment</u>
2002	Pediatrics resident lecture series		30
2003	Pediatrics resident lecture series		30
2004	Pediatrics resident lecture series		30
2005	Pediatrics resident lecture series		30
2006	Pediatrics resident lecture series		30
2005	Maternal Fetal Medicine and Neonatology Fellowship		15
2006	Maternal Fetal Medicine and Neonatology Fellowship		15
2007	Pediatric Grand Round		40
2010	Pediatric Grand Round (CHOC)		100

<u>Date</u>	<u>Undergraduate</u>	<u>Course</u>	<u>Role</u>	<u>Enrollment</u>
2002	Undergraduate research	Bio 2A	Lecturer	100
2005	Minority Research	course	Lecturer	100
2007	Minority Research	course	Lecturer	100

Other Teaching

Undergraduate student research Bio 199 2001-2012
 School of Biological Sciences
 University of California, Irvine
 Irvine, California

Ph.D. student rotation Bio201, 2001-2012
 School of Biological Sciences
 University of California, Irvine
 Irvine, California

School of biological Science, UCI, Bio 2A, 2002-2012
 School of Biological Sciences
 University of California, Irvine
 Irvine, California

Undergraduate student research, University of Cincinnati 2013-

PROFESSIONAL SERVICE

International

- 2005- Special Committee Member of Yusheng Youyu,
People's Republic of China,
- 2004- Peer Reviewer of National Natural Science Foundation of China (NSFC)
People's Republic of China
- 2005- Medical Research Council Grant Reviewer, England
- 2010- Canada Foundation for Innovation, Canada
- 2010- Grant Reviewer for Italian Ministry of Health
Republic of Italy
Ministry of Labour, Health and Social Policies
Department of Innovation
General Directorate for Health and Technologies Research

-
- 2011- Grant Reviewer for Wellcome Trust
Joint Research Office
Level 6 Leazes Wing
Royal Victoria Infirmary
Queen Victoria Road
Newcastle upon Tyne
NE1 4LP
Tyne and Wear
- 2010- Scientific advisory committee member to Chinese Ministry of Health on
Target Therapy.
- 2011 Grant Reviewer
South Africa's National Research Foundation (NRF)
- 2012- Grant Reviewer , Icelandic Research Fund
- 2013- Committee Chair, Advisory Committee
Beijing International Medical Center, Beijing, China
- 2014- Scientific Advisor to Chinese Ministry of Health on Birth Defect Control
- 2014 - Advisory committee member to Chinese Ministry of Health on Medical
Genetic program
- 2015 Steering committee member, standardized residency and fellowship
training, Chinese Medical Doctor Association

National

- 2005- 2012 Western Review Consortium
Peer Review Committee Member
American Heart Association
Western States Affiliate
- 2009 National Institute of Health
Study section
Emphasis Panel (SEP- ZRG1 OBT A 58)

University, Campus, School & Departmental Service

- 2006-2012 Scholarship Oversight Committee Neonatal-Perinatal Medicine
Fellowship Program, University of California, Irvine

-
- 2003-2006 School of Medicine Representative Assembly member
- 2001-2004 Member, Advisory committee
General Clinical Research Center, UCI
- 2004- Member, Data safety committee
General Clinical Research Center, UCI
- 2009- Mentoring committee
School of Medicine
University of California, Irvine
- 2010- The Research Committee, the Committee on Committees
School of Medicine
University of California, Irvine
- 2011- Council On Research, Computing And Libraries
University of California, Irvine

INVITED SPEAKER

International

- 2004 Newborn Screening in USA
2004 International Medical Genetics Conference
Beijing, China
- 2004 Molecular Basis of Cardiac Malformation
Beijing, China
2004
- 2004 Identification of a Novel Locus of Autosomal Dominant Isolated Noncompaction of the
Ventricular Myocardium
2004 IMG Conference
Beijing, China
July 2004
- 2004 Molecular Basis of Cardiac Malformations
Beijing Genome Center
Beijing, China
July 2004
- 2004 Cardiac Malformations

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- Grand Round
Beijing University Infant and Children's Hospital
Beijing, China
July 2004
- 2004 Molecular Basis of Congenital Heart Defects
Grand Round
Xiamen No. 1 Hospital
China
July 2004
- 2005 Course Director
2005 Sino-American Medical Genetics Training Course
Beijing
June 2005
- 2005 Newborn Screening in U.S.
ACGA-Fudan 2005
International Symposium on Genomic Medicine
June 2005
- 2005 Molecular Basis of Congenital Cardiac Defects
ACGA-Fudan 2005
International Symposium on Genomic Medicine
June 2005
- 2005 Human Mutation and Disease
First Hangzhou Annual Short Course on
Medical and Laboratory Applications of Genetics and Genomics
Hangzhou, China
October 2005
- 2005 Molecular Basis of Congenital Cardiac Defects
3rd meeting of pediatric physician-in-chief, Zhejiang Province, China
Hangzhou, China,
October 2005
- 2006 Mendelian Inheritance
The first Fujian province prenatal diagnostic conference
Fuzhou, Fujian, China
March 14th, 2006
- 2006 Genetic syndrome with congenital cardiac malformation
Annual Meeting of Chinese Society of Medical Genetics
Guangzhou, China

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- Dec 14th, 2006
- 2007 Prenatal screening in USA
Xiamen Women & Children's Hospital
Xiamen, China
July 27th, 2007
- 2007 TBX3 and Breast cancer
Symposium for graduate students on human genetics
Chinese Education Ministry, Changsha, China
August 2nd, 2007
- 2007 Molecular Basis of Congenital Cardiac Malformation
3rd Asian Pacific Congress in Maternal-Fetal Medicine, Nanjing, China
August 29, 2007
- 2007 Genetic studies of optic atrophy
Department of Ophthalmology and ENT
Huazhong University, China
September 4th, 2007
- 2008 International Symposium-Leber's Hereditary Optic Neuropathy
March 21-23, 2008, Wenzhou, China
- 2008 Cancer Genetics
2nd Hospital, Wenzhou Medical School
Wenzhou, China
- 2008 Genetics of Congenital Heart Defects
International Conference on Genetics and genomic Medicine
Hongkong
- 2009 Genetics of Optic Atrophy
United Mitochondrial Disease Foundation (UMDF)
Washington DC, July 31
- 2009 *Drosophila* Model of Optic Atrophy
Southern California *Drosophila* Symposium
September 11
- 2010 Current status of Prenatal Screening and Prenatal Diagnosis
International Conference on Genetics and genomic Medicine
Taiwan
- 2012 Application of novel genomic technologies to the birth defect control, The March of

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- Dimes, 2012 Conference
- 2012 Apply the novel genomic technologies to the CLIA certified laboratories, The SAPA 7th Annual Biomedical Forum
- 2013 CCHMC-Fudan Symposium: Fudan, Shanghai,
- 2014 Application of novel genomic technologies to the birth defect control
- 2015 1st International Conference on Women and Children Health in Nanning, China
Co-Organizer with over 700 attendees
- 2015 Application of novel genomic technologies to clinical genetics
- 2015 Potential of Stem Cell Therapy for Optic Atrophy
United Mitochondrial Disease Foundation (**UMDF**)
Washington DC,

Seminars

- 1998 Genotype-phenotype Correlation in Holt-Oram Syndrome
Clinical Genetic Conference
Harvard Medical School
Harvard Institute of Medicine
May 11, 1998
- 1998 Mutation Studies in Holt-Oram Syndrome
16th CANNEW (Canada-New York- New England) Clinical Genetic Conference
Portland, New Hampshire
May 1998
- 1998 Different TBX5 Interactions in heart and Limb defined by Mutation Studies in Holt-Oram Syndrome
98 Summer Greater Boston Biomed/Pharm Symposium
Harvard University, Boston
August 1998
- 2000 A path from clinic to lab: Clinical and Molecular Aspects of Holt-Oram syndrome
Fellow Seminar
Children's Hospital, Boston
Harvard Medical School
May 2000
- 2000 Clinical and Molecular Aspects of Holt-Oram syndrome
Departmental Seminar

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- Department of Pathology and Pediatrics
University of Virginia
May 2000
- 2000 Clinical and Molecular Aspects of Holt-Oram syndrome
Departmental Seminar
Department Pediatrics
University of Iowa
Aug 2000
- 2000 The Molecular Basis of Inter- and Intra-familial Phenotype Variations In Heart-Hand Syndrome. Department of Pediatrics /Department of Biophysics
University of California at Irvine
Nov, 2000
- 2001 A path from clinic to lab: Clinical and Molecular Aspects of Holt-Oram syndrome
National Children's Medical Center
George Washington University
Washington DC
Jan 24, 2001
- 2001 Clinical and Molecular Studies of Holt-Oram syndrome
Merck & Co. Inc. PA
March 6, 2001
- 2001 The Molecular Studies of Inter- and Intra-familial Phenotype Variations In Holt-Oram Syndrome. Department of Developmental Biology
University of California at Irvine
Feb 2001
- 2001 The Molecular Studies of Inter- and Intra-familial Phenotype Variations In Holt-Oram Syndrome. Department of Biological Chemistry
University of California at Irvine
Feb 2001
- 2002 A potential role of TBX3 in Human Breast Cancer, Signaling and Growth Factor Seminar.
Chao Family Cancer Center
October 7, 2002
- 2003 TBX3 and Breast Cancer
Chao Family Cancer Center Retreat
Palm Springs
September 2003

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- 2003 T-box Genes and Human Diseases
Department of Microbiology and Molecular Genetics
University of California, Irvine
October 2003
- 2004 Molecular Studies of TBX3 in Breast Cancer
Developmental Group Retreat
Chao Family Cancer Center
November 2003
- 2004 T-box Transcription Factors, TBX3, and Breast Cancer
2004 UCI Campuswide Symposium on Basic Cancer Research
University of California, Irvine
May 2004
- 2008 Mitochondrial disorder
Ambry Genetics, 2008
- 2008 Personalized medicine
2nd Hospital, Wenzhou Medical School
Wenzhou, China
- 2009 Personalized medicine
VA Hospital, Long Beach
- 2010 Target Therapy
2nd Hospital, Wenzhou Medical School
Wenzhou, China
- 2011 Clinical Application of Next Generation Sequencing
UCI Personalized medicine symposium
- 2011 Mitochondrial disorder
Systems Biology Lecture Series—Translational Medicine
- 2011 TBX3 and Breast Cancer
OIS/Breast Translational Working Group meeting
UC Irvine
- 2011 Stem cell therapy
Biopharmagen
Suzhou, China
- 2011 Are you ready for the new wave of genomic medicine?
Orange County Children's Hospital Grand Round

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- 2011 Genetics of CHD
Neonatal Fellow Lecture Series
- 2011 Orange County Children's Hospital Intensive Care Lecture Series
New wave of genomic medicine-MitoMed diagnostic lab
- 2011 TBX3 & Breast Cancer
Western University, CA
- 2011 Genetics of Optic Atrophy
Department of Ophthalmology, UC Irvine
- 2012 Applying novel technologies to solve puzzles of mitochondrial diseases, MitoClub
seminar
- 2012 Genetics of mitochondrial-related retinal degenerative diseases and potential of iPS cell-
based therapy, Cincinnati Children's Hospital
- 2013 Genetics of Mitochondrial diseases, Grand rounds, Cincinnati Children's Hospital
- 2013 Genetics of Mitochondrial diseases, Grand rounds, Joseph M. Sanzari Children's
Hospital, New Jersey
- 2014 Genetics of Mitochondrial-related Retinal Degenerative Diseases & Potential of iPS Cell-
based Therapy, Department of Molecular Biology, Cincinnati University
- 2014 Genetics of mitochondrial-related retinal degenerative diseases and potential of iPS cell-
based therapy. Keynote Speaker

The 6th Chinese Congress of Research in Vision and Ophthalmology (CCRVO2014),
Beijing
- 2014 Genetics of Mitochondrial-related Retinal Degenerative Diseases & Potential of iPS Cell-
based Therapy, Shanghai Children's Hospital-CCHMC symposium
- 2014 CHALLENGES OF DIAGNOSIS & MANAGEMENT OF MITOCHONDRIOPATHY, DIVISION OF
GASTROENTEROLOGY, CCHMC
- 2014 CHALLENGES OF DIAGNOSIS & MANAGEMENT OF MITOCHONDRIOPATHY, DIVISION OF
SURGERY, CCHMC
- 2014 Common Technologies for Mitochondrial Diseases, Metabolic Club, CCHMC
- 2014 Challenges of Diagnosis & Management of Mitochondriopathy, 2nd Rare disease
conference, Beijing

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- 2014 Challenges of Diagnosis & Management of Mitochondriopathy, Beijing Children's Hospital
- 2014 Challenges of Diagnosis & Management of Mitochondriopathy, Beijing Children's Hospital, Capital Pediatric Research Institute.
- 2014 Current Status of Application Genomic Technologies in Medical Genetics in USA, 9th Molecular and prenatal diagnostic symposium, Lanzhou, China
- 2014 Genetics of Mitochondrial Diseases, Genetic Counseling Program, CCHMC
- 2015 CHALLENGES OF DIAGNOSIS & MANAGEMENT OF MITOCHONDRIOPATHY, DAYTON CHILDREN'S HOSPITAL
- 2015 Current Status of Application Genomic Technologies in Medical Genetics in USA, Beijing Children's Hospital,
- 2015 Clinical Fellowship training in USA, National Standardized Training for Residency and Fellowship in China, Beijing

TEACHING & TRAINING

FACULTY

Jack Zhao	2009-2010
Chengkang Zhang, Ph.D., assistant project scientist	2009-2010
Jungwoo Kim, Ph.D., Professor, Pai Chai University, Korea	07/2010-
Jade Tran, Instructor	07/2011-07/2012
Guoli Sun, MD, Ph.D. visiting scientist	03/2013-09-2014

Research Postdoctoral Fellows

Xiaoying Peng	2015-
Shiyu Luo	2015-
Jerry Zhuo	2014-
Robert Hufnagel MD, Ph.D	2013-
Xiaolei Lui, Ph.D,	2014-
Xiaotao Zhao, MD	2014-
Jing Chen, Ph.D,	2012-
Shadi Khademi, Ph.D.	2013-2014
Feixia Dong	2014-2015
Jin Wang	2013-2014
Xin Fan	2013-2014
Taraneh Esmailpour, Ph.D.	2008-2012
Yan Wan, MD	2011-2012
Yongjun Luo, Ph.D, Postdoc Fellow	2011-2012

Sha Tang, Ph.D., Postdoc Fellow (Recipient of Cystinosis Research Foundation Postdoc Fellowship)	2007-2010
Jing Liu, MD	2007-2009
Yulin Zhou, Ph.D.	2008-2008
Michael Zaragoza, M.D., Ph.D.	2003-2003
Limin Lin, MD, Ph.D.	2005-2006
Xu Huang, Ph.D.	2003-2005
Joe Dae Whang, M.D., Ph.D. (supported by Korean Government, current attending and director in South Korea)	2004-2005
Xianhe Xie, M.D., Ph.D. (Currently Chairmen, Department of Hematology, Haikou Hospital)	2004-2005
Guifeng Sun, M.D., Ph.D.	2003-2005
Hongfei Pan, MD (Supported by Chinese Government Currently chairman, Department of Pediatrics, Youjian Medical college)	2005-2006
Tomasa Barrientos, Ph.D. (NIH Developmental Biology training grant)	2005-2006
Thin Aung Than, M.D., Ph.D. (NIH cancer biology training grant)	2005-2006

Residents:

Hanke Legere, M.D., Pediatrics resident	2004-2004
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Clinical Fellows (Harvard Medical School(1999-2001):

Jonathan Picker, M.D., Ph.D. (Clinical Genetics Fellow)	
Simon Albers-Bremer, M.D. (Clinical Genetics Fellow)	
Michele Hibbard, Ph.D. (Clinical Genetics Fellow)	
Can H. Ficioglu, M.D., Ph.D. (Clinical Genetics Fellow)	
Mike Murray, M.D. (Clinical Genetics Fellow)	
Juan Sebastian Saldivar, M.D. (Clinical Genetics Fellow)	
Mei Peng, M.D., Ph.D. (Clinical Molecular Genetics Fellow)	
Helen Lyons, M.D. (Clinical Genetics Fellow)	
Deyanira Corzo, M.D. (Clinical Genetics Fellow)	
Ana Cristina, M.D. (Clinical Genetics Fellow)	
Maria Judith Peterschmit, M.D. (Clinical Genetics Fellow)	
June-Anne Gold, MD, 2007-2009	
Esther Friedrich, MD, 2007-2010	
Shamin Jivabhai, MD, 2009-2010	
Robert Hufnagel, M.D., Ph.D. (Clinical Genetics Fellow) 2012-	

Ph.D. Students

Yanyan Peng	02/01/2013-
William Yaroshi,	07/01/2005-2008
Taraneh Esmailpour	07/01/2006-2012

Mariella Simmons 07/01/2010-

MD. Students

Catherine Nguyen (The Dean's Summer Research Grant), 2012-2013

Master Students:

Daryl Martinez. 2001-2002
Master Degree in Biotechnology

Ilham Salman 2004-2005
Master Degree in Biotechnology

Bonnie Chen, M.D. 2001-2006
Master Degree in Biotechnology

Linbo Yu 2007-2009
Master Degree in Genetic Counseling (Committee Chair)

Rotation Graduate Student

Ling Li 09/01/2001-01/15/2002
Weiwei Fan 07/01/2002-01/15/2003
Eric Wang 07/01/2003-09/15/2003

Thesis Committee & Advancement

2010 John Yang, Ph.D. student advancement & thesis committee member
2009 Linbo Yu, Chair, Genetic Counseling student, thesis committee member
2005 Weiwei Fan, Ph.D. student advancement, thesis committee member
2005 Malia Rumbaugh, Genetic Counseling student, thesis committee member
2004 Lianne Hasegawa, Genetic Counseling student, thesis committee member
2003 Kimberly Ann Kopita, Genetic Counseling student, thesis committee member
2006 Heidi Schoenh, Genetic Counseling student, thesis committee member
2003 Erin Nicholas, Genetic Counseling student, thesis committee member

Undergraduate Students:

Hai Dong 07/01/2013-
Vi A. Ngo 07/01/10-2012
Adnan Hussaini 07/01/10-2012
Brittany Chow 07/01/10-2012
Shreshtha Madaan 09/01/07-07/01/10
Zi xiao Liu 09/01/07-07/01/10
Jessica Orellana 09/01/07-07/01/10
Andy Chen 01/15/07-2009

Stephane Tse, (UROP Award recipient)	09/15/05-2008
Kimerly Nguyen (UROP Award recipient)	09/15/05-2008
Jerry Nguyen	09/15/05-2008
Daniel Ferrall	06/15/07-2008
Phung Khanh Le	09/01/04-2008
Laura Feucht	09/01/02-07/01/2004
Shelby Padua	09/01/02-07/01/2004
Andy Zinelis	09/01/02-07/01/2003
Chris Price (Honorable mentioned by Science)	09/01/03-07/01/2005
John Chen	09/01/02-07/01/2005
Maria Sarsiki,	09/01/03-07/01/2005
Delphine Larrrieu,	09/01/03-07/01/2004
University of Saoy in Bouret Du Lac, France	
Nathan Miu	09/01/03-07/01/2006
Thu Ha	09/01/04-07/01/2005

High School Students:

David Chen	07/07
Lydia Liang	07/09
Vincent Huang	07/09-

Qualifying/Preliminary Exam Committee

I have served on the qualifying and preliminary exam committee for the interdisciplinary Graduate Program in Molecular Biology, Genetics & Biochemistry (MBGB) for 15 students

PUBLICATIONS (in chronological order,*=co-first author) Complete List of Published Work in MyBibliography (79-76 publications):

<http://www.ncbi.nlm.nih.gov/myncbi/browse/collection/48064921/?sort=date&direction=ascending>

1. **Taosheng Huang**, and Yanlin Dong (1987) Changes in protein metabolism in the soleus muscles of the scalded rats and the therapeutic effect of leucine and insulin (II). *Journal of PLA Medical Colleges* 4:1-9.
2. **Taosheng Huang**, and Yanlin Dong (1987) Changes in protein metabolism in the soleus muscle of the scalded rats and therapeutic effect of leucine and insulin (I). *Acta Academia Medicine (Chinese)* 9:16-20
3. **Taosheng Huang**, and Yanlin Dong (1987) Changes in protein and nucleic acid metabolism in the liver of the scalded rats. *Acta Academia Medicine (Chinese)* 10:41-45. 1987
4. **Taosheng Huang**, and Yanlin Dong. (1987) Assay of the protein turnover rate for skeletal muscle. *Biochem Biophys Acta (Chinese)* 11:43-48
5. **Taosheng Huang**, Peter Palese and Mark Krystal (1990) Determination of influenza virus proteins required for genome replication. *Journal of Virology* 64:5669-5673
6. Yi Chu, **Taosheng Huang** and Ming-Ta Hsu (1990) P1 nuclease defines a subpopulation of active SV40 chromatin-- a new nuclease hypersensitivity assay. *Nucleic Acid Research* 18:3705-3711.
7. **Taosheng Huang** and Ming-Ta Hsu (1991) Inhibition of DNA replication of adenovirus type 5 and Simian virus 40 by tunicamycin. *Virology*, 182:889-893.
8. Kyeon H. Park, **Taosheng Huang**, Fred F. Correia and Mark Krystal (1991) Rescue of a synthetic gene into Sendai virus. *Proceedings of the National Academy Sciences of the USA*, 88:5537-5541.
9. **Taosheng Huang**, Jovan Pavloic, Peter Staeheli and Mark Krystal (1992) Overexpression of the influenza polymerase protein can titrate out inhibition by the murine Mx1 protein. *Journal of Virology*, 66:4154-4160.
10. Huang, T., Elias, E. R., Mulliken, J. B., Kirse, D. J. & Holmes, L. B. (1999) A new syndrome: heart defects, laryngeal anomalies, preaxial polydactyly, and colonic aganglionosis in sibs, *Genetics in medicine : official journal of the American College of Medical Genetics*. **1**, 104-8.
11. Craig T Basson, **Taosheng Huang**, Robert Lin, David R Bachinsky, Stanislaw Weremowicz, Alicia Vaglio, Rina Bruzzone, Roberto Quadrelli, Margherita Lerone, Giovanni Romeo, Margherita Silengo, Cynthia C Morton, Christoph W Muller, JG Seidman, Christine E Seidman (1999) Different TBX5 interactions in heart and limb defined by Holt-Oram syndrome. *Proceedings of the National Academy Sciences of the USA*, 96:2919-2924
12. **Huang, T.**, Yang, W., Pereira, A. C., Craigen, W. J. & Shih, V. E. (2000) Cloning and characterization of a putative human d-2-hydroxyacid dehydrogenase in chromosome 9q, *Biochemical and biophysical research communications*. **268**, 298-301.
13. **Huang, T.**, Lin, A. E., Cox, G. F., Golden, W. L., Feldman, G. L., Ute, M., Schrandt-Stumpel, C., Kamisago, M. & Vermeulen, S. J. (2002) Cardiac phenotypes in

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- chromosome 4q- syndrome with and without a deletion of the dHAND gene, *Genetics in medicine : official journal of the American College of Medical Genetics*. **4**, 464-7.
14. **Huang, T.**, Korson, M. S., Krauss, C. & Holmes, L. B. (2002) Four cases with hypoplastic thumbs and encephaloceles, *American journal of medical genetics*. **111**, 178-81. (Collected by London Dysmorphology Database)
 15. Lyon, H. M., Holmes, L. B. & **Huang, T.** (2003) Multiple congenital anomalies associated with in utero exposure of phenytoin: possible hypoxic ischemic mechanism?, *Birth defects research Part A, Clinical and molecular teratology*. **67**, 993-6.
 16. Drapkin, R. I., Genest, D. R., Holmes, L. B., **Huang, T.** & Vargas, S. O. (2003) Unilateral transverse arm defect with subterminal digital nubbins, *Pediatric and developmental pathology : the official journal of the Society for Pediatric Pathology and the Paediatric Pathology Society*. **6**, 348-54.
 17. Sun, G., Lewis, L. E., Huang, X., Nguyen, Q., Price, C. & **Huang, T.** (2004) TBX5, a gene mutated in Holt-Oram syndrome, is regulated through a GC box and T-box binding elements (TBEs), *Journal of cellular biochemistry*. **92**, 189-99.
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 20. **Huang, T.**, Whang, J. D. & Kimonis, V. (2006) Sex-influenced autosomal dominant optic atrophy is caused by mutations of IVS9 +2A>G in the OPA1 gene, *Genetics in medicine : official journal of the American College of Medical Genetics*. **8**, 59.
 21. Quadrelli, R., Strehle, E. M., Vaglio, A., Larrandaburu, M., Mechoso, B., Quadrelli, A., Fan, Y. S. & **Huang, T.** (2007) A girl with del(4)(q33) and occipital encephalocele: clinical description and molecular genetic characterization of a rare patient, *Genetic testing*. **11**, 4-10.
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 23. Quadrelli, A., Vaglio, A., Quadrelli, R., Mechoso, B., Fan, Y. S. & **Huang, T.** (2007) High-density array comparative genomic hybridization analysis and follow-up of a child with a de novo complex chromosome rearrangement detected prenatally, *Prenatal diagnosis*. **27**, 982-3.
 24. Tang S, Xu Q, Xu X, Du J, Yang X, Jiang Y, Wang X, Speck N, **Huang T.** (2007) A novel RUNX2 missense mutation predicted to disrupt DNA binding causes cleidocranial dysplasia in a large Chinese family with hyperplastic nails. *BMC Med Genet*, 8:82.
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 26. Yarosh, W.,* Barrientos, T.,* Esmailpour, T., Lin, L., Carpenter, P. M., Osann, K., Anton-Culver, H. & **Huang, T.** (2008) TBX3 is overexpressed in breast cancer and

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- represses p14 ARF by interacting with histone deacetylases, *Cancer research*. **68**, 693-9.*These authors contribute equally
27. Luo, Y., Gao, W., Gao, Y., Tang, S., Huang, Q., Tan, X., Chen, J. & **Huang, T.** (2008) Mitochondrial genome analysis of *Ochotona curzoniae* and implication of cytochrome c oxidase in hypoxic adaptation, *Mitochondrion*. **8**, 352-7.
 28. Tang, S., Le, P. K., Tse, S., Wallace, D. C. & **Huang, T.** (2009) Heterozygous mutation of Opa1 in *Drosophila* shortens lifespan mediated through increased reactive oxygen species production, *PLoS one*. **4**, e4492.
 29. Tang, S., Danda, S., Zoleikhaeian, M., Simon, M. & **Huang, T.** (2009) An Indian boy with nephropathic cystinosis: a case report and molecular analysis of CTNS mutation, *Genetic testing and molecular biomarkers*. **13**, 435-8.
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 33. Tang, S. & **Huang, T.** (2010) Characterization of mitochondrial DNA heteroplasmy using a parallel sequencing system, *BioTechniques*. **48**, 287-96.
 34. Tang, S., Batra, A., Zhang, Y., Ebenroth, E. S. & **Huang, T.** (2010) Left ventricular noncompaction is associated with mutations in the mitochondrial genome, *Mitochondrion*. **10**, 350-7.
 35. Liu, J., Esmailpour, T., Shang, X., Gulsen, G., Liu, A. & **Huang, T.** (2011) TBX3 over-expression causes mammary gland hyperplasia and increases mammary stem-like cells in an inducible transgenic mouse model, *BMC developmental biology*. **11**, 65.
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 38. Joshua Park, Debbie Liang, Hoda Anton-Culver, Jung Woo Kim, Yongjun Luo, **Taesheng Huang**, Soo-Young Kim, Seong-Sil Chang. (2012) Nail DNA and Possible Biomarkers: A Pilot Study, *J Prev Med Public Health* 45;235-243
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- A., McGillivray, B. C., Masser-Frye, D. & **Huang, T.** (2012) Genotype-phenotype analysis of 4q deletion syndrome: proposal of a critical region, *American journal of medical genetics Part A*. **158A**, 2139-51.
40. Strehle, E. M., Gruszfeld, D., Schenk, D., Mehta, S. G., Simonic, I. & **Huang, T.** (2012) The spectrum of 4q- syndrome illustrated by a case series, *Gene*. **506**, 387-91.
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42. Esmailpour, T. & **Huang, T.** (2012) TBX3 promotes human embryonic stem cell proliferation and neuroepithelial differentiation in a differentiation stage-dependent manner, *Stem cells*. **30**, 2152-63.PMID: 22865636, PMCID: PMC3517731
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44. Brodehl A, Dieding M, Klauke B, Dec E, Madaan S, **Huang T**, Gargus J, Fatima A, Saric T, Cakar H, Walhorn V, Tönsing K, Skrzypczyk T, Cebulla R, Gerdes D, Schulz U, Gummert J, Svendsen JH, Olesen MS, Anselmetti D, Christensen AH, Kimonis V, Milting H. (2013) The Novel Desmin Mutant p.A120D Impairs Filament Formation, Prevents Intercalated Disk Localization and Causes Sudden Cardiac Death. *Circ Cardiovasc Genet*. **6**(6):615-23 PMID: 2420090
45. Fan Y, Steller J, Gonzalez IL, Kulik W, Fox M, Chang R, Westerfield BA, Batra AS, Wang RY, Gallant NM, Pena LS, Wang H, **Huang T**, Bhuta S, Penny DJ, McCabe ER & Kimonis VE. (2013) A Novel Exonic Splicing Mutation in the TAZ (G4.5) Gene in a Case with Atypical Barth Syndrome, *JIMD reports*. **11**, 99-106 PMID:23606313
46. Riazifar H, Jia Y, Chen J, Lynch G. & **Huang T.** (2014) Chemically induced specification of retinal ganglion cells from human embryonic and induced pluripotent stem cells, *Stem cells translational medicine*. **3**, 424-32.PMID: 24493857
47. Esmailpour T, Riazifar H, Liu L, Donkervoort S, Huang VH, Madaan S, Shoucri BM, Busch A, Wu J, Towbin A, Chadwick RB, Sequeira A, Vawter MP, Sun G, Johnston JJ, Biesecker LG, Kawaguchi R, Sun H, Kimonis V & **Huang T.** (2014) A splice donor mutation in NAA10 results in the dysregulation of the retinoic acid signalling pathway and causes Lenz microphthalmia syndrome. *Journal of medical genetics*. **51**, 185-96. PMID: 24431331
48. Yang L, Tan Z, Wang D, Xue L, Guan MX, **Huang T** & Li R. (2014) Species identification through mitochondrial rRNA genetic analysis. *Scientific reports*. **4**, 4089.
49. Kwong JQ, Davis J, Baines CP, Sargent MA, Karch J, Wang X, **Huang T** & Molkenin JD. (2014) Genetic deletion of the mitochondrial phosphate carrier desensitizes the mitochondrial permeability transition pore and causes cardiomyopathy. *Cell death and differentiation*. **21**, 1209-17. PMID:24522485
50. Gong S, Peng Y, Jiang P, Wang M, Fan M, Wang X, Zhou H, Li H, Yan Q, **Huang T** & Guan MX. (2014) A deafness-associated tRNA^{His} mutation alters the mitochondrial function, ROS production and membrane potential. *Nucleic acids research*. **42**, 8039-48.

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51. Barber JC, Rosenfeld JA, Graham JM, Kramer N, Lachlan KL, Bateman MS, Collinson MN, Stadheim BF, Turner CL, Gauthier JN, Reimschisel TE, Qureshi AM, Dabir TA, Humphreys MW, Marble M, **Huang T**, Beal SJ, Massiah J, Taylor EJ & Wynn SL. (2015) Inside the 8p23.1 duplication syndrome; eight microduplications of likely or uncertain clinical significance. *American journal of medical genetics A*. **167A**, 2052-64.
52. Jiang P, Liang M, Zhang J, Gao Y, He Z, Yu H, Zhao F, Ji Y, Liu X, Zhang M, Fu Q, Tong Y, Sun Y, Zhou X, **Huang T**, Qu J & Guan MX. (2015) Prevalence of Mitochondrial ND4 Mutations in 1281 Han Chinese Subjects With Leber's Hereditary Optic Neuropathy. *Investigative ophthalmology & visual science*. **56**, 4778-88.
53. Riazifar H, Sun G, Wang X, Rupp A, Vemaraju S, Ross-Cisneros FN, Lang RA, Sadun AA, Hattar S, Guan MX & **Huang T**. (2015) Phenotypic and functional characterization of Bst+/- mouse retina, *Disease models & mechanisms*. **8**, 969-76.
54. Hufnagel RB, Arno G, Hein ND, Hersheson J, Prasad M, Anderson Y, Krueger LA, Gregory LC, Stoetzel C, Jaworek TJ, Hull S, Li A, Plagnol V, Willen CM, Morgan TM, Prows CA, Hegde RS, Riazuddin S, Grabowski GA, Richardson RJ, Dieterich K, **Huang T**, Revesz T, Martinez-Barbera JP, Sisk RA, Jefferies C, Houlden H, Dattani MT, Fink JK, Dollfus H, Moore AT, Ahmed ZM. (2015) Neuropathy target esterase impairments cause Oliver-McFarlane and Laurence-Moon syndromes, *Journal of medical genetics*. **52** (2), 85-94.
55. Ma H, Folmes CD, Wu J, Morey R, Mora-Castilla S, Ocampo A, Ma L, Poulton J, Wang X, Ahmed R, Kang E, Lee Y, Hayama T, Li Y, Van Dyken C, Gutierrez NM, Tippner-Hedges R, Koski A, Mitalipov N, Amato P, Wolf DP, **Huang T**, Terzic A, Laurent LC, Izpisua Belmonte JC & Mitalipov S. (2015) Metabolic rescue in pluripotent cells from patients with mtDNA disease, *Nature*. **524**, 234-8.
56. Abrams AJ, Hufnagel RB, Rebelo A, Zanna C, Patel N, Gonzalez MA, Campeanu IJ, Griffin LB, Groenewald S, Strickland AV, Tao F, Speziani F, Abreu L, Schule R, Caporali L, La Morgia C, Maresca A, Liguori R, Lodi R, Ahmed ZM, Sund KL, Wang X, Krueger LA, Peng Y, Prada CE, Prows CA, Schorry EK, Antonellis A, Zimmerman HH, Abdul-Rahman OA, Yang Y, Downes SM, Prince J, Fontanesi F, Barrientos A, Nemeth AH, Carelli V#, **Huang T**#\$, Zuchner S#\$ & Dallman JE.# (2015) Mutations in SLC25A46, encoding a UGO1-like protein, cause an optic atrophy spectrum disorder, *Nature genetics*. **47**, 926-32. #These authors contributed equally, \$These are the corresponding authors.
57. Simon M, Richard EM, Wang X, Shahzad M, Huang VH, Qaiser TA, Potluri P, Mahl SE, Davila A, Nazli S, Hancock S, Yu M, Gargus J, Chang R, Al-Sheqaih N, Newman WG, Abdenur J, Starr A, Hegde R, Dorn T, Busch A, Park E, Wu J, Schwenzler H, Flierl A, Florentz C, Sissler M, Khan SN, Li R, Guan MX, Friedman TB, Wu DK, Procaccio V, Riazuddin S, Wallace DC, Ahmed ZM, **Huang T**# & Riazuddin S#. (2015) Mutations of human NARS2, encoding the mitochondrial asparaginyl-tRNA synthetase, cause nonsyndromic deafness and Leigh syndrome, *PLoS genetics*. **11**, e1005097. #Corresponding author.
58. Leslie N, Wang X, Peng Y, Valencia CA, Khuchua Z, Hata J, Witte D, **Huang T**, Bove KE. (2016) Neonatal multiorgan failure due to ACAD9 mutation and complex I deficiency with mitochondrial hyperplasia in liver, cardiac myocytes, skeletal muscle, and renal tubules. *Hum Pathol*, **49**:27-32.

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59. Chen J, Riazifar H, Guan MX, **Huang T**. (2016) Modeling autosomal dominant optic atrophy using induced pluripotent stem cells and identifying potential therapeutic targets. *Stem Cell Res Ther*, **7**(1):2.
60. Jiang P, Jin X, Peng Y, Wang M, Liu H, Liu X, Zhang Z, Ji Y, Zhang J, Liang M, Zhao F, Sun YH, Zhang M, Zhou X, Chen Y, Mo JQ, **Huang T**, Qu J, Guan MX. (2016) The exome sequencing identified the mutation in YARS2 encoding the mitochondrial tyrosyl-tRNA synthetase as a nuclear modifier for the phenotypic manifestation of Leber's hereditary optic neuropathy-associated mitochondrial DNA mutation. *Hum Mol Genet*. **25**(3):584-96.
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