# CURRICULUM VITAE

## 1. PERSONAL DATA

**Name**: (Anne) Chun-Hui Tsai, M.D., MSc, FAAP, FACMG

**Present position: Visiting Associate Research Fellow**
**IBMS, Academia Sinica, Taipei**

**ACADEMIC**

1. 2006- Associate Professor (tenure track), Department of Pediatrics, University of Colorado School of Medicine
2. 2000- Faculty member, Graduate School, University of Colorado Health Sciences Center
3. 2002- Faculty member, Human Medical Genetic Program, UCHSC

**CLINICAL**

1. 1999- Attending physician/ Clinical Geneticist, Section of Clinical Genetics and Metabolism, Department of Pediatrics, The Children's Hospital, Denver
2. 2001- Clinical Privilege, Denver Health, Denver, CO
3. 2005- Clinical Privilege, Memorial Hospital, Colorado Spring, CO

**Address**: (Office) Section of Clinical Genetics and metabolism
Department of Pediatrics
The Children's Hospital, B300
1056 19th Ave
Denver, CO 80218
Tel: 303-861-6395
Fax: 303-861-3921
Email:tsai.chun-hui@tchden.org

**Nationality**: US/Taiwan Citizen

**Marital status:** Married

## 2. EDUCATION

**Degrees:**

1983-1990 China Medical College-- M.D. Granted 1990
1994-1996 Institute of Medical Science University of Toronto-- MSc Granted 1996

**Internship:**

1988-1989 Clerkship, Mackay Memorial Hospital, Taiwan
1989-1990 Internship, Mackay Memorial Hospital, Taiwan

**Residency:**

1990-1993 Residency, Department of Pediatrics, Cathay General Hospital, Taiwan
1996-1997 Senior Residency, PL-3 level, Henry Ford Hospital, Detroit, Michigan

**Fellowship:**

1993-1995 Clinical fellowship, Division of Clinical Genetics, The Hospital for Sick Children, Toronto, Canada
1995- 1996 Research Fellowship, Division of Endocrinology, The Toronto Hospital, Toronto, Canada
1997-1999 Clinical fellowship, Department of Medical Genetics, Henry Ford Hospital, Detroit Michigan
ABMG fellowship program

## 3. ACADEMIC APPOINTMENTS:

1. 1999-2006 Assistant Professor ( tenure track) , Department of Pediatrics, University of Colorado School of medicine
2. 1999- Attending physician/ Clinical Geneticist
   Section of Clinical Genetics and metabolism Department of Pediatrics
3. 2000- Faculty member, Graduate School, UCHSC
4. 2002- Faculty member, Human Medical Genetic Program, UCHSC
5. 2006 Associate Professor, Department of Pediatrics, University of Colorado School of medicine

## 4. HOSPITAL AND OTHER PROFESSIONAL POSITIONS:

1. 1999- Attending physician/ Clinical Geneticist
   Section of Clinical Genetics and metabolism
   Department of Pediatrics
   The Children's Hospital, Denver, CO
2. 2001- Genetic consultant, Honorary, Cathay General Hospital, Taipei, Taiwan
3. 2001- Clinical Privilege, Denver Health, Denver, CO
4. 2002, 2005- Clinical Privilege, Memorial Hospital, Colorado Spring, CO

## 5. HONORS/SCHOLARSHIP:

1983 First Prize of English Speech Contest, China Medical College
1986 Excellent Prize of English Speech Contest, National Youth Society
1986-88 Annual Scholarship of Shing-Kuang and Cathay Health
1994-95 University of Toronto, Open Scholarship
1995 Travel Award of 2nd joint Clinical Genetics Meeting, ACMG & NIH
2005 Excellent abstract, 1st ASPR, Taiwan Pediatric Association
2005 Travel Award, 1st Asia Pediatric Research conference, Nov 2005, Tokyo

## 6. MEMBERSHIP OF PROFESSIONAL SOCIETY: (Year starts, -till present)

| | |
|---|---|
| 1991 | Member, Taiwan Pediatric Association |
| 1991 | Member, Asia-Pacific Critical Care Society |
| 1992 | Member, American Society of Human Genetics |
| 1994 | Member, Taiwan Human Genetic Society |
| 1996 | Member, American Academy of Pediatrics |
| 1998 | Fellow, American Academy of Pediatrics |
| 1999 | Fellow, American College of Medical Genetics |

## 7. MAJOR COMMITTEE AND COMMUNITY SERVICE

1. Committee member for the NIH-National Human Genome Research Institute (NHGRI) haplotype mapping project involving the Chinese community in Denver (National)
2. Clinical Service Committee-Mountain State Genetics Network (Regional, national)
3. Education Committee-Mountain State Genetics Network (Regional, national)
4. Resident recruitment interviewer (TCH)
5. Medical Education committee (UCHSC)
6. National board examination question writer-Book of Biochemical Genetics (National)

## 8. LICENSURE AND BOARD CERTIFICATION

Licensure

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| 1991 | Medical License, Taiwan |
| 1997-2004 | Michigan General Medical License |
| 1999 | Colorado General Medical License |
| 1999 | Wyoming General Medical License |

Board certification

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| 1993 | Board Certified, Taiwanese Board of Pediatrics |
| 1993 | Board Certified, Taiwanese Board of Pediatric Emergency Medicine |
| 1996 | Board Certified, Taiwanese Board of Clinical Genetics |
| 1996 | Board Certified, Taiwanese Board of Biochemical Genetics |
| 1998 | Board Certified, American Board of Pediatrics |
| 1999 | Board Certified, American Board of Medical Genetics, Clinical Genetics |
| 2002 | Board Certified, American Board of Medical Genetics, Clinical Biochemical Genetics |
| 2005 | Recertified, American Board of Pediatrics |

## 9. INTELLECTUAL PROPERTY:

1. Genetic jeopardy for medical students, residents and graduate students (exported)
2. Hook-on Biochemical Genetics (computerized learning tool, in progress)

## 10. REVIEW AND REFEREE WORK:

1. Program/guest speaker list review and invitation: 2006 Asia Pacific Pediatrics, March 2006 (ad hoc)
2. Reviewer, American Journal of Medical Genetics, Mosby 2005
3. Reviewer, Pediatric International, Blackwell, 2005
4. GCRC guest reviewer, UCHSC/TCH, 2005

## 11. INVITED EXTRAMURAL LECTURES, PRESENTATIONS AND VISITING PROFESSORSHIPS

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| 1994 | Mirror-image and same-side craniofacial defects in MZ twins. Genetic Round, Oct. 6, 1994, The Hospital for Sick Children, Toronto |
| 1994 | Genetics Update. Genetics Round, Nov. 15, 1994, National Taiwan University Hospital. |
| 2000 | Annual workshop for Speech therapists in South-East Colorado. Solo speaker for a 3-hour lecture and 2-hour discussion. |
| 2000 | Clinical Genetics update National Taiwan University April 2000 |
| 2001 | Enzyme replacement therapy for Pompe disease, HMGP, Colorado |
| 2002 | Genetic counseling, what the parents need to know? 23rd Annual Perinatal Clinical Update. TCH, Denver, April 2002 |
| 2002 | Clinical Genetics update Chung-Shan Hospital, Taipei November 2002 |
| 2003 | Advance in Medical Genetics. Wangfan Municipal Hospital, Taipei April |
| 2003 | "One fun day in a genetic Clinic" Veteran General Hospital, Taipei, April |
| 2003 | Genetics of Psychiatric disorders, Sept 2003, Centennial, CO |
| 2004 | Genetic Counseling Veteran General Hospital, Combined conference Pediatrics/OBGYN Grand Round, Taipei, November 2004 |
| 2005 | Genetic counseling, what the primary doctors need to know? Louisiana Academy Of Family Practice, Winter CME course Feb 5, 2005 |
| 2005 | Molecular mechanism of Genetic Syndromes, genetic short course, Human genetic Society, Tokyo, Japan. April 8-11, 2005 |
| 2005 | Molecular Cytogenetics, Human genetic Society, genetic short course, Tokyo, Japan. April 8-11, 2005 |
| 2005 | Genetics of parry-Romberg Syndrome. Romberg Association, Lake Geneva, June 2005 |
| 2006 | Microdeletion syndrome, Short course in medical genetics, Human genetic Society, Taipei, March 2006 |
| 2007 | Molecular Diagnosis of Dysmorphic syndrome, Short course in medical genetics, Human genetic Society, Taipei, March 2007 |

## 12 TEACHING ACTIVITIES

**Graduate program of genetic Counseling**

BMGN 6618 Applied Medical Genetics Clinic (8 hours/week)
BMGN 6620-1 Applied Medical Genetics Clinic – Metabolic (6 hours/week)
BMGN 6620-2 Applied Medical Genetics Clinic - Regional/Specialty (8 hours/Month)
Advisory and examination committee member of

Jennifer Sollenberger, Class of 2000
Tamra Gibby and Katarynna Bloch, Class of 2001
Cary Armstrong and Rebecca Pollack, Class of 2002
Mercy Laurino, Kristen Zoller, Class of 2003
Gwendolyn Steffen and Charlotte Norman, Class of 2004
Agnes Kwan, Shannon Scrivner and Vivian Oliva, Class of 2005

**Medical School**
Didactic lecturer: BMG 5354 Medical Genetics, First year medical student (1 hour)
Didactic lecturer: Junior cord lectures, Third year medical student (once every six weeks)
Course Director: Pediatrics 8008, Elective rotation for 3rd or 4th medical students
Course Director: Pediatrics 7000-Clinical Genetics and inpatient
Section Coordinator: Metabolic block, first year medical student

**Resident Teaching**
a. Supervisor, Continuous clinic for genetic residents and fellows (2 patient/week/trainee; teaching 8 hours/week)
b. Annual floor attending for pediatric services, bedside teaching, didactic lectures (2 weeks/year; 8 hours a week)
c. Noon lectures for pediatric resident (1-2 annually)
d. Director: Elective rotations fro pediatric residents and pediatric subspecialty fellows.

**Innovative teaching material**
Genetic jeopardy
Rotation handbook Genetic and metabolic program

## 13. CLINCIAL DUTY

1. Director, Denver Clinic: one whole day clinic, weekly
2. Greeley out reach clinic: once every two month
3. Cheyenne outreach clinic: once every two month
4. Subspecialty inpatient services, 5 month a year
5. Pediatric inpatient services: 2 weeks a year

## 14. GRANT SUPPORT AND RESEARCH

**Funded Grants**
1. House officer research grant: 1999 Henry Ford Health System: Characterization of mitogen induced fragile sites
2. A Prospective, Multinational, Multicenter Clinical Trial of the Safety and Efficacy of Recombinant Human Acid (α-Glucosidase (rhGAA) in Cross Reacting Immunologic Material Positive (CRIM (+)) Patients with Classic Infantile Pompe Disease
   Pl: Chun-Hui (Anne) Tsai, MD
   Sponsor: Genzvme Corporation-AGLU-001-00 ($42,572/patient/year)

--Study closed, paper submitted to J of Pediatrics

3. Natural History of Recombinant 8 Syndrome
   PI: Chun-Hui Tsai, MD
   Mountain States Genetic Network
   $ 30,000, 2005-2006 active

4. Dysmorphology and Autism. Georgia CADDRE Project, Centers for Disease Control and Prevention, National Center on Birth Defects and Developmental Disabilities Division of Birth Defects and Developmental Disabilities
   $ 5400 annually, active

5. Etiological characterization of congenital malformation syndromes
   NSC-18,000 US, active

6. Autism treatment Center, Denver, Autism society USA.
   $ 7000 annually, active

**Research**

1999- Characterization of chromosome 8p23.1 breakpoints, Co-PI, Closed (published)
2000- Biochemical and clinical investigations of inborn errors of metabolism Co-PI, Active
2000**-** Investigation of hyperphenylalaninemia and phenylketonuria Co-PI, active
2001**-** A prospective multinational, multicenter, clinical trial of the safety and efficacy of recombinant human acid a-glucosidase (rhgaa) in cross-reacting immunologic material-positive (crim(+)) patients with classical infantile pompe disease, PI, Closed (submitted)
2001**-** Clinical experience in the dietary management of amino acid inborn errors of metabolism, Co-PI, active
2001- PVR genes in cleft lip with or without cleft palate, Co-PI, Closed
2003- Chromosomal markers and their genotype/phenotype correlation, Co-PI, Closed
2003- Genetic and metabolic characterization of PDD and Autism
2003- Genetic and metabolic characterization of psychiatric disorders
2004- VATER association and and early embryogenesis anomalies. (Paper submitted)
2004- Genetic thrombophilic predisposition and malformation s secondary to vascular accidents.
2005- Natural History of Recombinant 8 Syndrome, PI, Active
2005- Genetic and natural course characterization of Parry-Romberg syndrome
2006-CADDRE Autism study
2007-Etiological Characterization of congenital malformation syndrome.

## 15. PUBLICATIONS:

***Peer reviewed papers:***

1. Tsai CH, Hung KL. Vein of Galen aneurysm-- A Case Report.

Acta Paed Sin Suppl A 1992, 33:14-15.

2. Tsai CH, Hung KL. Ketogenic diet in treating Children with intractable Epilepsy. Medicine Today 1992, 19:197-199.

3. Chiang PW, Wang SQ, Song WJ, Crombez E, Smithivas P, Akhtar A, Im R, Greenfield J, Ramamoorthy S, Tsai CH, Blackburn C, Van Keuren M, Kurnit DM. Isolation and characterization of the human homologue of the yeast SPT4 gene. Genomics 1996, 34:328-333.

4. Chiang P-W, Wang S-Q, Smithivas P, Song W-J, Ramamoorthy S, Hillman J, Puett S, Van Keuren ML, Crombez E, Kumar A, Glover TW, Miller DE, Tsai CH, Blackburn CC, Chen X-N, Sun Z, Cheng J-F, Korenberg JR, Kurnit DM. Identification and analysis of the human and murine putative chromatin structure regulator, SUPT6H and *Supt6h*. Genomics 1996, 34:368-375.

5. Chiang PW, Baldacci PA, Babinet C, Camper SA, Watkins-Chow D, Baker DD, Tsai CH, Ramamoorthy S, King E, Slack AC, Fogel E, Morahan G, Ashworth A, Blackburn CC, Kurnit DM. Linkage mapping of murine homologue of the yeast SPT6 gene to MMU11B1, Mammalian Genome 1996, 7:459-460.

6. Tsai CH, Hill M, Drucker DJ. Biological Determinants of intestinotrophic properties of GLP-2 in vivo. Am. J. Physiol. 1997, 273:E77-84.
7. Tsai C-H, Hill, M, Asa SL, Brubaker PL, Drucker DJ. Intestinal growth-promoting properties of glucagon-like peptide-2 in mice. Am. J. Physiol. 1997, 272:G662-668.

8. Brubaker PL, Crivici A, Izzo A, Ehrlich P, Tsai C-H, Drucker DJ. Circulating and tissue forms of the intestinal growth factor, glucagon-like peptide-2. Endocrinology, 1997, 138:4837-4843.

9. Gupta IR, Tsai CH, Siegel-Bartelt J, Thorner P, Balfe JW. "Cutaneous telangiectasias, sparse hair, and type I membranoproliferative glomerulonephritis: Proesmans syndrome revised?" Pediatric Nephrology, Feb; 13(2):129-31, 1999

10. Tsai CH, Van Dyke DL, Feldman GL. A child with Velo-Cardio-Facial Syndrome and del (4) (q34.2): another critical region associated with a Velocardiofacial Syndrome-like phenotype. American J of Medical Genetics Vol 82, N0.4, p336-339 Feb 12, 1999

11. Tsai CH, Graw SL, McGavran L. 8p23 duplication reconsidered: is it a true euchromatic variant with no clinical manifestation? J Med Genet 2002 Oct; 39(10):769-74

12. Tsai AC, Robertson JR, Teebi AS. Teebi hypertelorism syndrome: Report of a family with previously unrecognized findings. Am J Med Genet. 2002 Dec 1;113(3):302-6.

13. Tsai AC , Gibby T, Beischel L , McGavran L, Johnson JP. A Child with Angelman syndrome and Trisomy 13 findings associated with Paternal UPD 15 and Segmental UPD 13 2004 Am J Med Genet Apr 15;126(2):208-12.

14. Toriello HV, Carey JC, Addor M , Allen W , Burke L, Chun N , Dobyns W , Elais, Gallagher R , Hordijk R , Hoyme G , Irons M, Jewett T , LeMerrer M , Lubinsky M , Martin R, McDonald-McGinn R, Neumann L, Newman W, Pauli R, Seaver L, Tsai A, Wargowsky D , Williams M, Zackai E. Toriello-Carey syndrome: Delineation and review Am J Med Genet. 2003 Nov 15;123(1):84-90. Review.

15. Salviati L, Freehauf C Sacconi S, DiMauro S, Thomas J, Tsai AC A Novel SURF1 Mutation in a Child with Subacute Encephalopathy and without the Radiological Features of Leigh Syndrome. Am J Med Genet. 2004 Jul 15;128A(2):195-8.

16. Tsai AC, Digiovanni M, Walton C, Cotter PD. De novo Duplication of the Short Arm of Chromosome 12:dup(12)(p13.1p13.3) Am J Med Genet A. 2005 Jan 4;134A(2):229-230

17. Struys EA, Verhoeven NM, Salomons GS, Berthelot J, Vianay-Saban C, Chabrier S, Thomas JA, Tsai AC, Gibson KM, Jakobs C.d-2-Hydroxyglutaric aciduria in three patients with proven SSADH deficiency: Genetic coincidence or a related biochemical epiphenomenon? Mol Genet Metab. 2006 Jan 24

18. Tsai, AC, Rawlinson CA, Yang M, Walton CS, Johnson JP De novo isodicentric X chromosome: 46,X,idic(X)(q24), and summary of literature. Am J Med Genet A. 2006 Apr 15;140(8):923-30

19. Kishnani PS, Nicolino M, Voit T, Rogers RC, Tsai AC, Waterson J, Herman GE, Amalfitano A, Thurberg BL, Richards S, Davison M, Corzo D, Chen YT. Chinese hamster ovary cell-derived recombinant human acid alpha-glucosidase in infantile-onset Pompe disease. J Pediatr 2006; 149(1):89-97.

20. Tsai, AC. SALL4 mutation: from Holt-Oram to VACTERL?--A hypothesis of the pathogenesis of VACTERL association. AJMG-MS-05-0401 (Revision 1)

21. Thurberg BL, Lynch CM, Vaccaro C, Afonso K, Tsai AC, Bossen E, **Kishnani PS**, O'Callaghan M. Characterization of pre- and post-treatment pathology after enzyme replacement therapy for pompe disease.Lab Invest. 2006 Dec;86(12):1208-20. Epub 2006 Oct 30.

22. Anne Chun-Hui Tsai, Chantal F. Morel, Gunter Scharer, Michael Yang, , Jordan P. Lerner-Ellis, David S. Rosenblatt, and Janet A. Thomas Late-onset Combined Homocystinuria and Methylmalonic Aciduria (cblC) and Neuropsychiatric Disturbance. Am J Med Genet A. 2007;143A:2430-34

22. Chiang PW, Aliaga S, Travers S, Spector E, Tsai AC. Case report: WT1 exon 6 truncation mutation and ambiguous genitalia in a patient with Denys-Drash syndrome. Curr Opin Pediatr. 2008 Feb;20(1):103-106.

23. Pei-Wen Chiang · Elaine Spector · Anne Chun-Hui Tsai. Evidence Suggesting the Inheritance Mode of the Human P Gene in Skin Complexion Is Not Strictly Recessive. In press, AJMG- 2008

**Letter to the editor:**

1. Tsai CH, Van Dyke DL, Feldman GL. Letter to the editor. Reply to the letter to the editor by Zackai-"deletion 4q34.2". Am J Med Genet. 1999 Sep 10; 86(2):198.

2. ACH Tsai and JP Johnson. Response to "How Exhaustive Are Reviews in Research Review Articles?" AJMG MS06-1001

**Book chapters:**

1. Tsai AC, Stool S "Phylogenic Aspects and Embryology" of the Bluestone et al. Pediatric Otolaryngology, 4th edition. 2003 Saunders p1-21

2. Tsai AC, Vallee S "Genetics, Syndromology and Craniofacial Anomalies" of the Bluestone et al. Pediatric Otolaryngology, 4th edition. 2003 Saunders p37-58

3. Elias E, Tsai CH, Manchester DK "Genetics and Dysmorphology" chapter 33, Pediatric diagnosis and treatment. William W. Hay et al. Ed 16, 2003 McGraw Hill, p1001-1050

4. Elias E, Tsai CH, Manchester DK "Genetics and Dysmorphology" chapter 33, Pediatric diagnosis and treatment. William W. Hay et al. Ed 17, 2005 McGraw Hill, p 1039-1079

5. Tsai AC, Greene C. Approach for child with congenital anomalies and dysmorphic features, Steve Berman, Pediatric decision making, 4th edition. 2003 Mosby p490-497

6. Tsai AC, Walton C. Approach for child with cleft lip and palate, Steve Berman, Pediatric decision making, 4th edition. 2003 Mosby p164-169

7. Thomas JT, Tsai ACH and Bernstein LW Nutrition Therapies for Inborn Errors of Metabolism. Neonatal nutrition and Metabolism. 2nd Edition. Cambridge University Press. Patti J. Thureen. Edited by William W. Hay April 2006

8. Tsai, CH Intervention program, Chapter 11 of Metabolic diseases, Taiwan experience. 1st Ed, Lee 2004 Yee-Shuan Publisher. p110-115

9. Text book of pediatric development. Tsai AC, Pickler L, Hagerman R. Chapter: chromosomal anomalies and X-link mental retardation

***Abstracts and Presentations***

**Oral presentation and Competitive abstracts:**

1. Tsai CH, Chen WC, Wang YC. High Dose Frequently Administered Nebulized Terbutaline in Children with Moderate to severe Acute Asthma.--Abstract of **Oral Presentation** On Biannual Convention of the Chinese Taipei Pediatric Association. Acta Paed Sin Suppl B. 1992, 33:79-80.

2. Tsai CH, Lin LH. The Effects Of Cisapride in infant with Gastroesophageal Reflux.--Abstract of **Oral Presentation** on Biannual conference of Chinese Taipei Pediatric Association. Acta Paed Sin Suppl D. 1992, 33: 47.

3. Tsai ACH, Siegel-Bartelt J. Mirror image Saethre-Chotzen syndrome in MZ Twins: Presumed evidence of early gene expression-- Abstract of **Platform Presentation**. XV David W. Smith Workshop on Malformations and Morphogenesis. 1994, p72. (paper in preparation).-- **Highly competitive!!**

4. Tsai C.H, Van Dyke DL, Roberson JR. A child with GCMS and (9)(q22.1q22.3) deletion. **Platform presentation.** 6th Joint clinical Genetics Meeting. American College of Medical Genetics, March 1999 (Paper in preparation)--**Highly competitive!!**

5. Tsai C.H, Bawle EV, Van Dyke DL, Roberson JR. Two patients with del (9)(q22.1q22.3) syndrome. --Abstract of **Oral Presentation** On Biannual Convention of the Chinese Taipei Pediatric Association. Acta Paed Sin Suppl B. 1999

6. Tsai CH, A woman with Sex Chromosome Abnormality: 47,XXX[4] **/** 45,X[3] **/** 48,XXXX[1] **/** 49,XXXX,+8[1] **/** 46,XX[39] and Mullerian Aplasia—Title changed to Two patients with Mullerian Aplasia -**Platform presentation** at the Asia pediatric congress 2000 Taipei

7. Anne Chun-Hui Tsai, M.D., Matthew Taylor, M.D., Luaren Shermann, M.S**.** Is Klippel-Feil deformity, conductive deafness, and absent vagina syndrome in the spectrum of MURCS association. Abstract to XXI David W. Smith Workshop on Malformations and Morphogenesis. Aug 2000. Abstract published on Proceedings of the Greenwood Genetic Center 2001, Volume 20 p16--**Highly completive!!**

8. Anne Chun-Hui Tsai, Gary Bellus, M.D., Severe manifestation of Goltz syndrome-- a presumed evidence of defect in cholesterol biosynthesis. Abstract to XXI David W. Smith Workshop on Malformations and Morphogenesis. Aug

2000. Abstract published on Proceedings of the Greenwood Genetic Center 2001, Volume 20 p166-- **Highly competitive!!**

9. CH Tsai, T Gibby, L Beischel, L. McGavran and John P. Johnson. A Child with Angelman syndrome and facial findings of Trisomy 13 due to paternal UPD 15 and Segmental UPD 13: evidence of post-zygotic somatic recombination between maternal and paternal #13 homologues Poster at XXII David W. Smith Workshop on Malformations and Morphogenesis. Sept 2001. (paper published #14)--**Highly competitive!!**

10. Tsai CH. Non-classic cocaine syndrome with documented DNA repair defect-Broaden the screen in patient with lacking of physiological growth for DNA repair defect. **Oral presentation** 172 biannual pediatric convention 2002 Acta Paed Sin Suppl. Vol. 43 2002

11. J Levine, S Colan, P Kishnani, A Amalfitano, CH Tsai, G Herman, J Waterson, R Rogers, F Yong, and YT Chen. Cardiac response in patients with classical infantile Pompe disease (CIPD) receiving recombinant human acid alpha glucosidase (rh-GAA): Preliminary results from a Phase 2 study. American Heart Association annual meeting, poster presentation 2002

12. . P Kishnani, T Voit, M Nicolino, A Amalfitano, CH Tsai, G Herman, J Waterson, RC Rogers, H Landy, D Corzo, B Thurberg, S Richards, and YT Chen. Treatment of classical infantile pompe disease (cipd) with recombinant human acid alpha glucosidase (rhgaa): preliminary data from a phase 2 study Poster presentation # 2417, ASHG, 2002

13. P Kishnani, T Voit, M Nicolino, A Amalfitano, CH Tsai, G Herman, J Waterson, A Rogers, H Landy, G Cox, T Braakman, D Corzo, B Thurberg, S Richards, and YT Chen. Recombinant human acid alpha glucosidase (rh-GAA) for treatment of classical infantile pompe disease (cipd): preliminary data from a phase 2 study. **Oral presentation** at SSIEM meeting in Dublin, Sept, 2002.

14. P Kishnani, T Voit, M Nicolino, CH Tsai, G Herman, J Waterson, RC Rogers, J. Levine, A Amalfitano, H Landy, D Corzo, B Thurberg, S Richards, and YT Chen. Enzyme Replacement Therapy with Recombinant Human Acid Alpha Glucosidase (rhGAA) in Classical Infantile Pompe Disease (CIPD): Results from a Phase 2 Study. **Oral presentation**. Society for Pediatric Research Meeting in 2003

15. Tsai AC. SAL-4 mutation: from Holt-Oram to VATER. Abstract of **Oral Presentation** at Biannual Convention of the Taiwan Pediatric Association. 2003

16. Tsai AC. SAL-4 mutation: from Holt-Oram to VATER. Abstract of **Oral Presentation** at 25$^{th}$ David W. Smith Workshop on Malformations and Morphogenesis snowbird, Utah, Aug 2004. Abstract published on Proceedings of the Greenwood Genetic Center 2004 **Highly competitive!**

17. V. Cunningham, C Tsai. Pfeiffer syndrome type 3 associated with a G>T mutation in exon 7 of the FGFR2 previously reported associated with Pfeiffer type 2. Abstract of Poster Presentation 25th David W. Smith Workshop on Malformations and Morphogenesis Abstract published on Proceedings of the Greenwood Genetic Center 2004--**Highly competitive!!**

18. Chun-Hui Tsai and Virginia Proud, Malformation syndromes secondary to vascular accidents- 5 case reports as evidence that thrombophilic factors may be causal. Abstract of **Oral Presentation at** 26th David W. Smith Workshop on Malformations and Morphogenesis Iowa City, Aug 2005 –**Highly competitive!!**

19. Arlene Drack, MD and Anne Chun-Hui Tsai, MD Chromosome 9 Inversion Associated with Juvenile Cataracts and Ambiguous Genitalia. Abstract accepted for **Rapid Fire poster session.** Ophthalmology Conference September 2005.

20. Chun-Hui Tsai Malformation syndromes secondary to vascular accidents- 6 case reports as evidence that thrombophilic factors may be causal. Abstract of **Oral Presentation** at Biannual Convention of the Taiwan Pediatric Association. Nov 2005

21. Anne Chun-Hui Tsai Thrombophilic predispositions in malformation syndromes secondary to vascular accidents. Abstract accepted for ***Oral presentation*** at the 1st Congress of the Asian Society for Pediatric Research (ASPR). November 2005 Tokyo, Japan.—travel award winner

22. Anne Chun-Hui Tsai 2 familial cases of Parry-Romberg Syndrome--evidence of Mendelian inheritance and vascular involvement. Abstract of **Oral Presentation** at 28th David W. Smith Workshop on Malformations and Morphogenesis Williamsburg,VA, Aug 2007. Abstract published on Proceedings of the Greenwood Genetic Center 2007 **Highly competitive!**

**Poster presentations: (numbers continued from above)**

23. Tsai CH, Costa MT, Chen WC, Feigenbaum A, Teshima I. Terminal chromosome 11q delesion (Jacobsen Syndrome)-Report of three cases.-- Abstract of Oral Presentation. Acta Paed Sin Suppl D. 1993, 34:67.

24. Tsai ACH, Allingham-Hawkins, DJ, et al. Becker Muscular Dystrophy Caused by duplication of exon 3-6 of the dystrophin gene presenting as dilated cardiomyopathy. ASHG suppl 1994 #1833.

25. Tsai ACH, Teshima I, Cytrynbaum C, Siegel-Bartelt J. F.I.S.H. Analysis of Rare Maternal Translocation (15;16) (q13;q13) in a boy with duplication 16p and Angelman syndrome --Poster #44, 2nd Joint Clinical Genetics Meeting, 26th March of Dimes Clinical Genetics Conference, American College of Medical Genetics, March 1995

26. Tsai ACH, Siegel-Bartelt J. Respective Heterotaxia and VATER Association in Monoaminiotic Monozygotic Twins-- Poster #155, 2nd Joint clinical Genetics Meeting, 26th March of Dimes Clinical Genetics Conference, American College of Medical Genetics, March 1995 ( Paper in preparation)

27. Tsai CH, Van Dyke DL, Feldman GL. A child with Velo-Cardio-Facial Syndrome and a terminal deletion of chromosome 4 long arm, del (4) (q34.2): another critical region associated with a DiGeorge-like phenotype-- Poster #118. 5nd Joint clinical Genetics Meeting. American College of Medical Genetics, March 1998 (Paper published #10 and #11)

28. Tsai CH, Conard JV, Van Dyke DL, Feldman GL. Unusual phenotype of inverted duplication of 8p, dup(8)(p23p22), in a mother and a daughter -- Poster #119, 5th Joint clinical Genetics Meeting. American College of Medical Genetics, March 1998

29. Lazebnik N, Tsai CH. Abnormal multiple marker screen and ultrasound findings of fetal edema secondary to Congenital Erythropoietic Porphyria. ASHG suppl 1999 # 987.

30. C-H Tsai, J, Yu, T. Gilfillian, L Meltesen, B Hirsch, L McGavran. Complex chromosomal rearrangements in a 1p36 deletion syndrome child with a cryptic interstitial deletion. ASHG poster 858 2000

31. C.H. Tsai, M. Taylor, J. Siegel-Bartelt. Early clinical features of Angelman Syndrome in infants with chromosomal deletion of 15q11-q13 (Poster presentation # 90 at 7th Joint clinical Genetics Meeting. American College of Medical Genetics, March 2000)

32. K.H. Greenly, R.W. Tyson, C.H. Tsai. The extreme Spectrum of Pallister-Hall syndrome. (Poster presentation # 166 at 7th Joint clinical Genetics Meeting. American College of Medical Genetics, March 2000)

33. Familial hypertelorism, a G syndrome variant or a Teebi-like syndrome C.-H. Tsai, KG Monaghan, J. R. Roberson -Poster presentation, accepted for the 8th Joint clinical Genetics Meeting. American College of Medical Genetics, March 2001 (paper published #13)

34. Inverted duplication 16 q 24.1-22 syndrome. T. Datkhaeva, L. McGavran, A. Boswell, and C.-H. Tsai. (Poster presentation accepted for the 8th Joint clinical Genetics Meeting. American College of Medical Genetics, March 2001)

35. S.L. Graw, C.-H. Tsai, L. McGavran Duplication of 8p23.1: Characterization of 8 new cases. Poster presentation ASHG 2001 (Paper published. # 12)

36. TI Datkhaeva, E Sujansky, CH Tsai. The clinical significance of heteromorphism of chromosome 4p10. Poster # 728 ASHG 2001

37. K. A. Washington, E. J. Hoffenberg, C.-H. Tsai, E. R Elias. Unusual GI Pathology in a Patient With Rett Syndrome (RS) Poster presentation ASHG 2001

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