1. NAME AND PERSONAL DATA

Samantha A. Brugmann, Ph.D.
samantha.brugmann@cchmc.org
(o) 513-636-1599
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2. EDUCATION

Stanford University 2004-2010
Postdoctoral Research Fellow
Department of Surgery
(mentor: Jill Helms) 2004-2010
Department of Chemical and Systems Biology
(mentor: Joanna Wysocka) 2010-2011

The George Washington University 1998-2004
Graduate Student (Ph.D.), Genetics
Department of Anatomy and Cell Biology
(mentor: Sally Moody)
Columbian College for Arts and Sciences
Institute for Biomedical Sciences
Dissertation: The induction and function of *Xenopus* Six1 in cranial placode development

Tulane University 1994-1998
B.S., Cell and Molecular Biology
Newcomb School of Arts and Science
Minor in Spanish

3. ACADEMIC APPOINTMENT

Assistant Professor, Cincinnati Children's Hospital Medical Center (CCHMC)
Department of Pediatrics (primary) 2011-2016
Division of Developmental Biology
Department of Surgery (secondary)
Division of Plastic Surgery

Associate Professor, CCHMC 2016-2019
Department of Pediatrics
Division of Developmental Biology
Department of Surgery
Division of Plastic Surgery

Scientific Staff, Shriners Hospital for Children, Cincinnati 2017-present

Associate Professor (with Tenure), CCHMC 2019-present
Department of Pediatrics
Division of Developmental Biology

4. AWARDS AND HONORS

Research Achievement Award (Faculty Awards Committee CCHMC)	2018
Cincinnati Children's Research Foundation Endowed Scholar	2017
Presidential Early Career Award for Scientists and Engineers (received at White House)	2016
AAA Young Faculty Travel Award	2015
Pediatric Research Fund-Child Health Research Program Grant	2009
Helena Anna Henzl-Gabor Young Women in Science Fund Travel Fellowship.	2008
Development Traveling Fellowship from the journal *Development*	2007
Loan Repayment Grant from the Loan Repayment Program (NIH).	2007
3rd Place in Society for Experimental Biology Graduate Student Symposium	2004
Honorable Mention at SDB/ISDB international meeting poster competition	2003
Honorable Mention at Mid-Atlantic Regional SDB meeting	2002
Cosmos Club Foundation Young Scholars Award	2001

5. MEMBERSHIPS IN PROFESSIONAL ORGANIZATIONS

Society for Integrative and Comparative Biology
- Attended meeting in 2007
- Published paper based on data presented at meeting

American Society of Human Genetics
- Poster presentation in 2006

Orthopaedic Research Society
- Podium presentation 2006

Society for Craniofacial Genetics & Developmental Biology
- Member since 2005
- Assisted in the organization of the meeting in 2007
  - Formulated program
  - Organized poster session
- Podium presentation in 2007, 2010, 2014
- Poster presentation 2013, 2015, 2017
- Chaired session on Genomic Approaches to Uncover Diseases Mechanisms 2015
- Chaired session on Variation and Patterning the Craniofacial Complex 2017
- Elected Secretary of Society 2020

Society for Developmental Biology
- Member since 2000
- Presented posters at 2000, 2001, 2003, 2009, 2013 national/international meetings

Presented posters at 2002, 2006, 2009, 2012 regional meetings
Oral presentation and session chair at 2016 regional meeting
Oral presentation at Satellite Symposium on Neural Crest and Ectodermal Placodes, 2009
Attended New Faculty Boot Camp 2010
Judged student and postdoc posters 2010, 2011, 2013
Podium presentation at Hilde Mangold Postdoctoral Symposium 2010

American Association for Anatomy
Member since 2010
Invited podium speaker 2011, 2015, 2019

6. SERVICE

a. Committee Involvement:

Admissions Committee member: Graduate Program in Molecular & Developmental Biology 2011-Present
- Review and score applications
- Interview candidates

Seminar Committee Chair: Cincinnati Children's Hospital Research Foundation/Molecular and Developmental Biology 2017-Present
- Chair Committee to select annual seminar speakers
- Survey current literature and select high quality speakers
- Host selected speakers

Executive Committee Member for Master's Program in Biomedical Research Technologies 2015-Present
- Assisted in curriculum design
- Review and score applications
- Interview candidates

Associate Director of Graduate Studies 2018-2021
- Serve as a point of contact for graduate students
- Act as the liaison among the graduate students and program faculty

Basic Science Research Committee (BSRC)
- Elected member 2018-Present
- Active participation in key institutional research initiatives
- Facilitate understanding of CCHMC leadership to academic issues
- Recommend new strategic research opportunities for investment

Integrated Research Advisory Committee (IRAC)
- Elected member 2018-Present
- Develop recommendations for an institutional research strategy
- Convey research issues from BSRC to CCRF leadership

Surgical Services Research Committee 2018-Present
Invited member
Discuss and take action on issues related to the improvement of Basic Science Research and Research associated infrastructure in the Department of Surgery

Faculty Awards Committee 2019-Present
Invited member
Review nominations for each of the faculty awards at CCHMC

b. Professional activities:

Early Career Reviewer NIH, SBDD Study Section, CSR 10/2012
Ad-hoc reviewer NIH, NIDCR Study Section, (DSR Panel) 2/2014
Back up member of BTS Study Section (CCHMC) 1/2014, 7/2014, 10/2014
Ad-hoc reviewer for NIH, NIEHS ONES Study Section 7/2014
Ad-hoc reviewer NIH, ZRG1 MOSS V-02 Special Emphasis Panel 10/2014
Ad-hoc reviewer NIH, ZDE1 VH (04)1 Study Section 6/2015
Member DSR Study Section NIH, (NIDCR) 7/2016-6/2022
Ad-hoc reviewer NIH, ZHD1 DRG-D (50) 4/2017

c. National/international distinguished activities:

Chair of first Gordon Research Seminar for Craniofacial Morphogenesis and Tissue Regeneration 4/2010

Invited Panelist for Career Mentorship Session at Gordon Research Seminar for Craniofacial Morphogenesis & Tissue Regeneration 3/2014

Invited Discussion Leader for Gordon Research Conference on Craniofacial and Tissue Regeneration 3/2014

Invited Panelist for Meet the Experts session at FASEB Biology of Cilia and Flagella meeting 7/2015

Invited Session Chair for 38th annual Society for Craniofacial Genetics and Developmental Biology meeting 10/2015

Invited Session Chair for Midwest regional meeting of the Society for Developmental Biology 10/2016

Invited Panelist for Career Mentorship Session at Gordon Research Seminar on Neural Crest and Placode Development 2/2017

Invited Session Chair for 40th annual Society for Craniofacial Genetics and Developmental Biology meeting 7/2017

Invited Discussion Leader ‘Clinical Genetics of Craniofacial Disorders’

Gordon Research Conference on Craniofacial Morphogenesis and Tissue Regeneration 2/2018

d. Editorial Service:

Frontiers in Craniofacial Biology: Member of Editorial Board 2013-present
AIMS Genetics: Member of Editorial Board 2014-2017
Genesis: Guest Editor Special Issue for Craniofacial Development 2018

e. Manuscripts review:

1. Development
2. Developmental Biology
3. *PLoS* Genetics
4. Tissue Engineering
5. Cellular and Molecular Life Sciences
6. Genesis
7. Developmental Dynamics
8. Disease Models & Mechanisms
9. eLife

f. Participation in department recruitment activities including resident staff interviewing committee:

1. Interviewed and evaluated faculty candidates for: 2011-present
   i. Developmental Biology
   ii. Human Genetics
   iii. Urology
   iv. Neuroscience
   v. Endocrinology
   vi. Reproductive Sciences
   vii. Plastic Surgery
   viii. Mind, Brain, Behavior
   viiii. CuSTOM
   x. Molecular Cardiovascular Biology

2. Interviewed and evaluated resident and graduate student candidates 2012-present
   Presented review of programmatic research for MD/PhD Resident applicants

3. Member of search committee for Division Director of Developmental Biology
   Attended committee meetings to select candidates 2012-2013
   Attended candidate lectures and gave feedback
   Met with candidates as a part of the Craniofacial group

4. Member of search committee for Financial Analyst Position for Surgical Services
   Attended committee meetings to define position 2013
   Attended committee meetings to review and interview candidates

5. Member of search committee for Director, Molecular Cardiovascular Biology & Executive Co-Director of Heart Institute of Developmental Biology 2017
   Attended committee meetings to select candidates
   Attended candidate lectures and gave feedback
   Met with candidates as a representative of the Craniofacial group

6. Member of search committee for Division Director, Plastic Surgery 2018
   Participated in candidate selection
   Met with candidates as a representative of the Research group
   Evaluated and ranked candidates

g. Participation in local activities that benefit the institution:
   Volunteer for Cincinnati Walks: September 2013
   Faculty Participant, Cell Motility Poster Session, Molecular and Cellular Biology core curriculum course: December 2013
   Go the Distance 5K participant: September 2018

h. Community activities:
   Member of Mt. Adams Civic Association: January 2011-Present
   Member of Mt. Adams Movie Night Committee: 2011
   Volunteer for Mt. Adams Movie Night: June-August 2011
   Volunteer for Labor Day street fair in Mt. Adams: September 2011
   Participant in annual Thanksgiving Day Race: November 2011-2013, 2017
   Participant in Flying Pig Half Marathon: May 2013, 2014, 2018
   Participant in AHA Mini Marathon: March 2013, 2014
   Volunteer for Cincinnati Walks: September 2013
   Volunteer for Bloom Africa: December 2013
   Volunteer for The Cincinnati Zoo: 2013-2016
   Member of Eastern Hills Tennis Club: 2012-2018
   Member of Cincinnati Tennis Club: 2016

i. Service related grants and contracts: N/A

7. TEACHING

a. Percent time per year spent in teaching of medical students/graduate students-10%

1. MDB/MSTP students hosted for 4-12 week laboratory rotation
   a. Bliss Magella-11/2011-12/2011
   b. Katie Bezold- 5/2012-6/2012
   c. Elizabeth Schock-7/2012-8/2012
   d. Grethel Millington-2/2014-3/2014
   e. Rebecca Rice-8/2014-9/2014
   f. Kelsey Elliot-1/2016-2/2016
   g. Lauren Falkenberg-6/2017-7/2017

h. Evan Brooks-7/2018-8/2018
i. Regan Bales- 9/2018-12/2018
j. Andrea Holderbaum- 6/2019-7/2019
k. Simon Han-7/2019-8/2019
l. Kieran Phelan-7/2020-8/2020

2. Current Post-doctoral fellows
   a. Ching-Fang Chang 2012-present (promoted to Research Scientist)
   b. Christian Louis Bonatto Paese 2019-present

3. Former Post-doctoral fellows
   a. Ya-Ting Chang 2014-2017
   b. Megan Aarnio-Peterson 2017-2018
   c. Sai Balchand 2017-2020

4. Current Ph.D. Students
   a. Evan Brooks 2019-present

5. Former students
   a. Grethel Millington, M.S. 2014-2016
   b. Elizabeth Schock, Ph.D. 2012-2017
   c. Kelsey Elliott, Ph.D. 2016-2020

6. Participation in Mentorship programs
   a. Host for exchange program between CCHMC and Aix Marseille.
      Hosted student Dilara Tomtiyeva July-August 2012
   b. Host for McAuley High School Student
      Hosted student Annamarie Helpling April 2013
   c. Host for Women in Science and Engineering (WISE) Student
      Hosted Aarti Kumar Summer 2014
   d. Host for Summer Undergraduate Research Fellowship (SURF) Students
      Hosted Julie Chen Summer 2014
      Hosted Hoa Pham Summer 2018
   e. Selection Committee member for Schmidlapp Young Women Scholar Award 2016-Present
   f. Host for UC Co-OP and CapStone program 2019-2020
      Hosted Preston Schultz

b. Dates of academic courses, lectures, grand rounds, professor rounds, and participation in firms

Class: Introduction to Developmental Biology
Lecture: Neural crest and axon specificity
Dates: 11/9/12, 11/14/12, 11/8/13, 11/13/13, 11/5/14, 11/7/14, 11/6/15, 11/9/15, 11/4/16, 11/7/16, 11/8/17, 11/13/17

Class: Development and Disease

Lecture: Craniofacial
Dates: 2/9/12, 2/28/14, 3/28/15, 3/26/18

Class: Advanced Developmental Biology
Lecture: Primary cilia
Dates: 3/20/17, 3/22/17

Journal Clubs for Graduate Students
Date: 11/8/11, 10/15/13, 9/29/15, 11/15/16

Lecture for Physician Scientist Training Program Students
Date: 10/6/11

Lecture for Molecular and Developmental Biology Program Graduate Students
Dates: 7/22/11, 7/10/12, 7/9/13

Lecture for Heart Institute students and postdocs about obtaining a faculty position
Date: 12/2/13

Lecture for OAACD Career Development Seminar: Strategies for success in basic research for junior faculty
Date: 7/19/18

Organizer for judging talks at Molecular and Developmental Biology Program Graduate Symposium
Date: 8/22/13, 8/21/14, 9/1/16, 9/21/17

Lecture for Molecular and Cellular Biology Core Course for Graduate Students
Lecture: Molecular motors, intracellular transport, spindle dynamics
Dates: 11/10/14, 11/12/14

Course Director for Practical Laboratory skills course for Master's Program in Biomedical Sciences
Formulated syllabus, participated in lab classroom design, sat on admissions committee
Dates: 2015-Present

Lecture for Practical Laboratory skills course for Master's Program in Biomedical Sciences
Lecture and hands on laboratory training in: Histology, Immunostaining and In Situ Hybridization
Dates: 11/17/15- 12/3/15

c. Percent time spent in preceptorships: N/A

d. Time spent in participation on graduate or post graduate student thesis committee
    1. 5%
        a. PhD thesis advisor: Elizabeth Schock
        b. MS thesis advisor: Grethel Millington
        c. PhD thesis advisor: Kelsey Elliot
        d. Member of thesis committee for Abigail Bower: Potter Lab
        e. Member of thesis committee for Zheng Zhang, Zorn Lab
        f. Member of qualifier and thesis committee for Lu Han, Zorn Lab
        g. Member of pre-qualifier committee for Ming Fang, Yutzey Lab
        h. Member of pre-qualifier committee for Bliss Magella, Potter Lab
        i. Member of qualifier and thesis committee for Aria Attia, Stottmann Lab
        j. Member of qualifier and thesis committee for John Snedeker, Stottmann Lab
        k. Member of qualifier and thesis committee for Andrew DiStasio, Stottmann Lab
        l. Member of qualifier committee for Marshal Luckas (MSTP), Stottmann Lab
        m. Member of qualifier and thesis committee for Alexandra Eicher, Wells Lab
        n. Member of thesis committee for Talia Nasr (MSTP), Zorn Lab
        o. Member of thesis committee for Jacqueline Ehrman (MSTP), Campbell Lab

e. Teaching awards
    N/A

f. Teaching material
    Upon request

8. RESEARCH

a. Grants and Contracts
    Current Funding

i. 543938 Effort: 3.72 mos.
"The role of the ciliary protein C2CD3 during craniofacial skeletogenesis"
Shriner's Hospital for Children
Funded from: 01/01/2018-12/31/2022
Role: PI
Total costs: $1,320,000

ii. R35 DE027557 Effort: 7.8 cal mos.
"Harnessing the therapeutic potential of neural crest cells by manipulating the primary cilium"
NIH-NIDCR
Funded from: 09/14/2017 – 06/30/2025
Role: PI
Total Direct costs: $5,200,000

iii. Endowed Scholars Award Effort: 0 cal mos.

Children's Research Foundation (CRF) CCHMC
Role: PI
Total Direct costs: $150,000

v. Center for Pediatric Genomics Pilot Project Grant Effort: 0 cal mos.
"Forward Genetic Analysis of Craniofacial Development"
Children's Research Foundation (CRF) CCHMC
Role: member of Craniofacial Anomalies Research Consortium (Bender, Brugmann Gordon, Hopkin, Jiang, Lan, Saal, Stottmann, Sund, van Aalst)
Total Direct costs: $100,000

Completed Funding

i. R01 GM112744 Effort: 0.24 cal. mos.
"A genetic approach to defining the Ttc21b interactome in mammalian ciliopathies."
NIH-NIGMS
Funded from: 2/1/15-1/31/19
Role: co-I (with R. Stottmann)
Total Direct costs: $192, 500

ii. U18 NS080815-01 Effort: 1.8 cal. mos.
"Generating Human Intestinal Organoids with an ENS"
NIH- NCATS and NINDS
Funded from: 7/24/2012 - 6/30/2014
Role: co-I (with J. Wells)
Total Direct costs: $444,939 ($192,151 to Brugmann)

iii. Pathway to Independence Award (K99/R00) Effort: 9 cal. mos
5R00DE019853-04
"The role of primary cilia in craniofacial development"
NIH, NIDCR
Funded from 2/1/10-1/31/14
Role: PI
Total Direct costs: $710,957

iv. Trustees Award Effort: 0 cal. mos
"The role of Kif3a in craniofacial development"
Children's Hospital Research Foundation, Cincinnati Children's Hospital
Funded from 1/1/12-12/31/13
Role: PI
Total Direct costs: $120,000

v. Research Innovation and Pilot Funding Effort: 0 cal. mos
"A translational approach towards the identification of causative genetic elements for ciliopathies"
Children's Hospital Research Foundation, Cincinnati Children's Hospital
Funded from 6/1/12-5/31/13
Role: co-PI (with R. Stottmann)
Total Direct costs: $75,000

vi. Research in Progress Award Effort: 0 cal mos.
"Using the Developing Embryo to Identify Factors that Influence the Primary Ciliome"
Funded from: 8/13/15-8/12/16
Role: PI
Children's Research Foundation (CRF) CCHMC
Total Direct costs: $75,000

vii. R01 DE023804 Effort: 3.6 cal. mos.
"The role of primary cilia in murine craniofacial development"
NIH-NIDCR
Funded from: 12/13/13-9/14/17 (terminated early because award of R35)
Role: PI
Total Direct costs: $1,250,000

b. Peer reviewed articles
(*41 publications; Total citations= 5966; h-index=23; as of 9/2020 (SCOPUS))*

C.L. Bonatto Paese, E.C. Brooks, M. Aarnio-Peterson, **S.A. Brugmann**. (2020). Ciliopathic micrognathia is caused by aberrant skeletal differentiation and remodeling. (*In Revision, Development*)

K.H. Elliott, X. Chen, J. Salomone, P. Chaturvedi, P.A. Schultz, S.K. Balchand, J.D. Servetas, A. Zuniga, R. Zeller, B. Gebelein, M.T. Weirauch, K.A. Peterson, **S. A. Brugmann**. (2020). Gli3 utilizes Hand2 to synergistically regulate tissue-specific transcriptional networks. (*In Revision, eLife*)

M.A. Metzler, S. Raja, K.H. Elliott, R.M. Friedl, N.Q.H. Tran, **S.A. Brugmann**, M Larsen, L.L. Sandell. (2019). RDH10-mediated retinol metabolism and RARα-mediated retinoic acid signaling are required for submandibular salivary gland initiation. *Development.* **145**(15) PMID:29986869.

K.H. Elliott, G. Millington, **S.A. Brugmann.** (2019). A novel role for cilia-dependent Sonic hedgehog signaling during submandibular gland development. *Dev Dyn.* **247**(6):818-831. PMID:29532549. (*Highlighted in Faculty of 1000*) (*Cover photo*)

K.H. Elliott and **S.A. Brugmann.** (2018). Sending mixed signals: Cilia-dependent signaling during development and disease. *Dev Bio.* PMID:29548942. Epub ahead of print.

E.N. Schock and **S.A. Brugmann.** (2017). Neural crest cells utilize primary cilia to regulate ventral forebrain morphogenesis via Hedgehog-dependent regulation of oriented cell division. *Dev Bio.* **431**(2):168-178. PMID: 28941984.

E.N. Schock, J.N. Struve, C.F. Chang, T.J. Williams, J. Snedeker, A.C. Attia, R.W. Stottmann, **S.A. Brugmann**. (2017). A tissue-specific role for intraflagellar transport genes during craniofacial development. *PLoS One.* **12**(3):e0174206. PMID:28346501

J Snedeker, E.N. Schock, J.N. Struve, C.F. Chang, M. Cionni, P.V. Tran, **S.A. Brugmann**, R.W. Stottmann. (2017). Unique spatiotemporal requirements for intraflagellar transport genes during forebrain development. *PLoS One*. **12**(3):e0173258. PMID:28291836

G. Millington, K.H. Elliott, Y.T. Chang, C.F. Chang, A. Dlugosz, **S.A. Brugmann**. (2017). Cilia-dependent GLI processing in neural crest cells is required for tongue development. *Dev Biol.* pii: S0012-1606(16)30879-X. PMID:28286175

M.J. Workman, M.M. Mahe, S. Trisno, H.M. Poling, C.L. Watson, N. Sundaram, C.F. Chang, J. Schiesser, P. Aubert, E.G. Stanley, A.G. Elefanty, Y. Miyaoka, M.A. Mandegar, B.R. Conklin, M. Neunlist, **S.A. Brugmann**, M.A. Helmrath, J.M. Wells. (2016). Engineered human pluripotent-stem-cell-derived intestinal tissues with a functional enteric nervous system. *Nature Medicine.* **23**(1):49-59. PMID:27869805

Y.T. Chang, P. Chaturvedi, E.N. Schock and **S.A. Brugmann**. (2016). Understanding mechanisms of GLI-mediated transcription during craniofacial development and disease using the ciliopathic mutant, *talpid*^{2}. *Frontiers in Physiology.* **7**(468). PMID:27799912

C.F. Chang, Y.T. Chang, G. Millington and **S.A. Brugmann**. (2016). Craniofacial ciliopathies reveal specific requirements for GLI proteins during development of the facial midline. *PLoS Genetics.***12**(11):1-22. PMID:27802276

E.N. Schock and **S.A. Brugmann**. (2016). Discovery, diagnosis and etiology of craniofacial ciliopathies. *Cold Spring Harbor Perspectives.* pii: a028258. doi: 10.1101/cshperspect.a028258. PMID: 28213462

L.E. Romick-Rosendale, E.E. Hoskins, L.M. Privette Vinnedge, G.D. Foglesong, M.G. Brusadelli, S.S. Potter, K. Komurov, **S.A. Brugmann**, P. Lambert, R.J. Kimple, E.L. Virts, H. Hanenberg, M.L. Gillison, S.I. Wells. (2016). Defects in the Fanconi anemia pathway in head and neck cancer cells stimulate tumor cell invasion through DNA-PK and Rac1 signaling. *Clin Cancer Res.* **22**(8):2062-73. PMID:26603260

E.N. Schock, C.F. Chang, I.A. Youngworth, M.G. Davey, M.E. Delany and **S.A. Brugmann**. (2016). Utilizing the chicken as an animal model for human craniofacial ciliopathies. *Developmental Biology.* **415**(2):326-37. PMID:26597494

E.N. Schock, C.F. Chang, J.N. Struve, Y.T. Chang, J. Chang, M.E. Delany and **S.A. Brugmann**. (2015). Using the avian mutant *talpid*^{2} as a disease model for understanding the oral-facial phenotypes of Oral-facial-digital syndrome. *Disease Models and Mechanisms.* **8**(8):855-66. PMID: 26044959

H.M. Saal, C.A. Prows, I. Guerreiro, M. Donlin, L. Knudson, K.L. Sund, C.F. Chang, **S.A. Brugmann**, R.W. Stottmann. (2015). A mutation in FRIZZLED2 impairs Wnt signaling and causes autosomal dominant omodysplasia. *Human Molecular Genetics.* **24**(12):3399-409. PMID:25759469

C.F. Chang, E.N. Schock, A.C. Attia, R.W. Stottmann and **S.A. Brugmann**. (2015). The Ciliary Baton: Orchestrating Neural Crest Cell Development*. Current Topics in Developmental Biology. Vol. **111.*** Paul A. Trainor, editor*,* Burlington: Academic Press, pp. 97-134. ISBN: 978-0-12-407759-1

C.F. Chang, E.N. Schock, E.A. O'Hare, J. Dodgson, H.H. Cheng, W.M. Muir, R.E. Edelmann, M.E. Delany, **S.A. Brugmann**. (2014). The cellular and molecular etiology of the craniofacial defects in the avian ciliopathic mutant, *talpid*^{2}. *Development.* **114**(15):3003-3012. PMID:25053433. (*Highlighted in Faculty of 1000*)

S.A. Brugmann and J.M. Wells. (2013). Building additional complexity to *in vitro* derived intestinal tissues. *Stem Cell Research & Strategy.* **4**(Suppl 1): S1.

A.L. Chang, G. Atzmon, A. Bergman, **S. Brugmann**, S.X. Atwood, H.Y. Chang, N. Barzilai. (2013). Identification of Genes Promoting Skin Youthfulness by Genome-Wide Association Study. *J Invest Dermatol.* **134**(3):651-7. PMID: 24037343

H. Liu, Y. Lan, J. Xu, C.F. Chang, **S.A. Brugmann**, R. Jiang. (2013). Odd-skipped related-1 controls neural crest chondrogenesis during tongue development. *Proc Natl Acad Sci.* **110**(46):18555-60. PMID:24167250

A. Rada-Iglesias, R. Bajpai, S. Prescott, **S.A. Brugmann**, T. Swigut, J. Wysocka. (2012). Epigenomic annotation of enhancer elements predicts transcriptional regulators of human neural crest. *Cell Stem Cell.* **11**(5):633-48. PMID:22981823

K.E. Powder, Y-C Ku, **S.A. Brugmann**, R.A. Veile, N.A. Renaud, J.A. Helms, M. Lovett. (2012). A Cross-Species Analysis of MicroRNAs in the Developing Avian Face. *PLoS ONE* **7(**4): e35111. PMID:22523571

K. Lenton, A.W. James, A. Manu, **S.A Brugmann**, D. Birker, E.R. Nelson, P. Leucht, J.A. Helms, M.T. Longaker. (2011). Indian hedgehog positively regulates calvarial ossification and

modulates bone morphogenetic protein signaling. *Genesis*. **49**(10):784-96. PMID: 21557453

B. Levi, A.W. James, E.R. Nelson, **S.A. Brugmann**, M. Sorkin, A. Manu, M.T. Longaker. (2011). Role of Indian hedgehog signaling in palatal osteogenesis. *Plast Reconstr Surg*. **127**(3):1182-90. PMID: 21364421

N.A. Zaghloul and **S.A. Brugmann.** (2011). The emerging face of primary cilia. *Genesis.* **49**(4):231-46. PMID: 21305689

A. Rada-Iglesias, R. Bajpai, T. Swigut, **S.A. Brugmann**, R.A. Flynn, J. Wysocka. (2011). A unique chromatin signature uncovers early developmental enhancers in humans. *Nature.* **470**(7333):279-83. PMID: 21160473

D.R. Cordero, **S. Brugmann**, Y. Chu, R. Bajpai, M. Jame, J.A. Helms. (2011). Cranial neural crest cells on the move: Their roles in craniofacial development. *Am J Med Genet A*. **155**(2):270-9. PMID:21271641

B. Levi**, S. Brugmann**, M.T. Longaker. (2010). Discussion: Hes1 is required for the development of craniofacial structures derived from ectomesenchymal neural crest cells. J Craniofac Surg. **21(**5):1450-1. PMID:20818250

S.A. Brugmann, D.R. Cordero, J.A. Helms. (2010). Craniofacial ciliopathies: A new classification for craniofacial disorders. *Am J Med Genet Part A*. **152A**:2995–3006. PMID: 21108387

S.A. Brugmann, N. C. Allen, Z. Mekonnen, E. Madan, J.A. Helms. (2010). A primary cilia-dependent etiology for midline facial disorders**.** *Human Molecular Genetics.* **19**(8):1577-92. PMID:20106874

S.A. Brugmann*, K.A. Powder*, N. Young, L.H. Goodnough, S.M. Hahn, A.W. James, J.A. Helms, M. Lovett. (2010). Comparative gene expression analysis of avian embryonic facial structures reveals new candidates for human craniofacial disorders. *Human Molecular Genetics.* **19**(5):920-30. PMID:20015954

A.W. James, A.A. Theologis, **S.A. Brugmann**, Y. Xu, A.L. Carre, P. Leucht, K. Hamilton, K.S. Korach, M.T. Longaker. (2009). Estrogen/estrogen receptor alpha signaling in mouse posterofrontal cranial suture fusion. *PLoS One.* **4**(9):e7120. PMID:19771170

D. ten Berge, **S. A. Brugmann**, J.A. Helms, R. Nusse. (2008). Wnt and FGF signals interact to coordinate growth with cell fate specification during limb development. *Development*. **135**:3247-3257. PMID:18776145

C.T. Anderson, A.B. Castillo, **S.A. Brugmann**, J.A. Helms, C.R. Jacobs, T. Sterns. (2008). Primary Cilia: Cellular Sensors for the Skeleton. *Anatomical Record*. **291**(9):1074-8. PMID:18727074

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J.L. Rinn, J.K. Wang, N. Allen, **S.A. Brugmann**, A. Mikels, H. Liu, T. Ridky, H.S. Stadler, R. Nusse, J.A. Helms, H.Y. Chang. (2008). A dermal HOX transcriptional program regulates site-specific epidermal fate. *Genes and Development*. **22**(3):303-71. PMID:18245445

J.A. Helms and **S.A. Brugmann**. (2007). The origins of species-specific facial morphology: The proof is in the pigeon. Integrative and Comparative Biology. 51(1):1-5. URL http://icb.oxfordjournals.org/cgi/ content/full/icm051v1

J. L. Rinn, M. Kertesz, J. K. Wang, S. L. Squazzo, X. Xu, **S.A. Brugmann**, L. H. Goodnough, J. A. Helms, P. J. Farnham, E. Segal, and H. Y. Chang (2007). Functional Demarcation of Active and Silent Chromatin Domains in Human HOX Loci by Noncoding RNAs. *Cell*. **129**:1311-1323. PMID: 18245445

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S.A. Brugmann, J. Kim and J.A. Helms. (2006). Looking different: towards an understanding diversity in facial form. *American Journal of Medical Genetics*. **140**(23):2521-9. PMID:16838331

S.A. Brugmann, M.D. Tapadia and J.A. Helms. (2006). The Molecular Origins of Species-Specific Facial Pattern. *Current Topics in Developmental Biology*. **73**:1-42. PMID:16782454

S.A. Brugmann and S.A. Moody. (2005). Induction and specification of the vertebrate ectodermal placodes: precursors of the cranial sensory organs. *Biology of the Cell.* **97**(5):303-19. (Cover). PMID:15836430

S.A. Brugmann, P.D. Pandur, K.L. Kenyon, F. Pignoni and S.A. Moody. (2004). Six1 promotes a placodal fate within the lateral neurogenic ectoderm by functioning as both a transcriptional activator and repressor. *Development*. **131**(23):5871-81. PMID:15525662

c. Quality review of publications

1. **S.A. Brugmann**, N. C. Allen, Z. Mekonnen, E. Madan, J.A. Helms. (2010). A primary cilia-dependent etiology for midline facial disorders**.**

a. Significance: This manuscript was the first to propose that loss of primary cilia can result in a gain of Hedgehog function. It also proposed a novel class of craniofacial disorders called craniofacial ciliopathies.
b. Number of citations: **90**

2. C.F. Chang, E.N. Schock, E.A. O'Hare, J. Dodgson, H.H. Cheng, W.M. Muir, R.E. Edelmann, M.E. Delany, **S.A. Brugmann**. (2014). The cellular and molecular etiology of the craniofacial defects in the avian ciliopathic mutant, *talpid*^{2}.
   a. Significance: This manuscript identified the causal mutation of the long utilized developmental mutant *talpid*^{2}.
   b. Number of citations: **30**

3. **S.A. Brugmann**, L.H. Goodnough, A. Gregorieff, P. Leucht, D. ten Berge, C. Fuerer, H. Clevers, R. Nusse, and J.A. Helms. (2007). Wnt signaling mediates regional specification in the vertebrate face.
   a. Significance: This manuscript addressed the molecular nature of craniofacial morphology. We found that levels of Wnt activity play a major role in determining outgrowth of facial prominences.
   b. Number of citations: **155**

4. A. Rada-Iglesias, R. Bajpai, T. Swigut, **S.A. Brugmann**, R.A. Flynn, J. Wysocka. (2011). A unique chromatin signature uncovers early developmental enhancers in humans.
   a. Significance: This manuscript addressed the epigenetic signature of enhancers. We found that the histone modification profile of the enhancer region of active genes is distinct from those that are poised to be active.
   b. Number of citations: **1234**

5. **S.A. Brugmann**, P.D. Pandur, K.L. Kenyon, F. Pignoni and S.A. Moody. (2004). Six1 promotes a placodal fate within the lateral neurogenic ectoderm by functioning as both a transcriptional activator and repressor.
   a. Significance: This manuscript identified the transcription factor, Six1 as an essential gene in establishing the pre-placodal domain. We determined that Six1 functions both as an activator and repressor to form tissue boundaries in the developing embryo.
   b. Number of citations: **154**

d. Abstracts (Selected)

Title: **Understanding the Etiology of Craniofacial Ciliopathies: Past, Present and Future**
Author(s): Ching-Fang Chang, Kelsey H. Elliott, Sai Balchand, Yanfen Yang, Samantha A. Brugmann
Conference: David W. Smith Workshop on Malformations and Morphogenesis
Location: Snowmass, UT

Date: AUGUST 23 - 28, 2019

Title: **A novel human homozygous IGF1R mutation is causal of Congenital Malformations, Severe GH, IGF-I and Insulin Insensitivities**
Author(s): Fujimoto, M., Maystadt, I., Zhang, D., Andrew, S.F, Chang, C.F.,Wauters, N., Joset, P., Benoît, V., Oneda B., Brugmann, S., Dauber, A., Rosenfeld, R.G., Sanchez-Gurmaches, J., Rauch, A., De Schepper, J., Yakar, S. and Hwa, V.
Conference: FASEB
Location: West Palm Beach, FL
Date: JULY 7 - 12, 2019

Title: **Co-factors and motif sequence features diversify transcriptional outputs in mandibular neural crest cells**
Author(s): Elliott, K.E., Chen, X., Salomone, J., Chaturvedi, P., Gebelein B., Weirauch, M.T., Peterson, K.A, Brugmann, S.A.
Conference: Gordon Conference for Neural Crest and Placode Development
Location: Barga, Italy (INTERNATIONAL)
Date: APRIL, 2019

Title: **GLI-dependent Etiology of Craniofacial Ciliopathies**
Author(s): Chang, Ching-Fang; Chang, Ya-Ting; Millington, Grethel; **Brugmann, S.A.**
Conference: Hedgehog 2015
Location: Puerto Varras, Chile (INTERNATIONAL)
Date: OCTOBER 24, 2015

Title: **Understanding the cellular and molecular etiology of craniofacial ciliopathies**
Author(s): Chang, Ching-Fang; Chang, Ya-ting; Millington, Grethel; **Brugmann, S.A.**
Conference: Craniofacial Genetics
Location: Baltimore, MD (NATIONAL)
Date: OCTOBER 6, 2015

Title: **Understanding the cellular and molecular etiology of craniofacial ciliopathies**
Author(s): Chang, Ching-Fang **and Brugmann, S.A.**
Conference: FASEB SRC: The Biology of Cilia and Flagella
Location: Snowmass, CO (NATIONAL)
Date: JULY 19, 2015

Title: **GLI-dependent Etiology of Craniofacial Ciliopathies**
Author(s): Chang, Ching-Fang and **Brugmann, S.A.**
Conference: AAA 2015
Location: Boston, MA (NATIONAL)
Date: MARCH 31, 2015

Title: **Using the *talpid*^{2} as novel model for determining the cellular and molecular etiology for Oral-facial-digital syndrome**
Author(s): Schock, Elizabeth N.; Chang, Ching-Fang; Struve, Jaime; Chang, Julie and **Brugmann, S.A.**
Conference: CILIA 2014
Location: Paris, France (INTERNATIONAL)
Date: NOV 18, 2014

Title: **Cellular, molecular and genetic characterization of the avian ciliopathic model *talpid2***
Author(s): Chang, Ching-Fang; Schock, Elizabeth N.; Delany, M.E.; **Brugmann**, **S.A.**
Conference: Society for Craniofacial Genetics & Developmental Biology
Location: Boston, MA (NATIONAL)
Date: OCT 22, 2013

Title: **Genetic and molecular characterization of the avian ciliopathic model *talpid*^{2}**
Author(s): Chang, Ching-Fang; Schock, Elizabeth N.; Delany, M.E.; **Brugmann**, **S.A.**
Conference: 72nd Annual Meeting of the Society-for-Developmental Biology
Location: Cancun, Mexico (INTERNATIONAL)
Date: JUNE 16-20, 2013

Title: **Characterization of the avian *talpid*^{2} mutant**
Author(s): **Brugmann, S.A.**; Chang, Ching-Fang; Schock, Elizabeth N.; et al.
Conference: Joint Annual Meeting of the ASPET/BPS at Experimental Biology (EB)
Location: Boston, MA (NATIONAL)
Date: APRIL 20-24, 2013

e. Books, Chapters, Reviews

1. C.F. Chang, E.N. Schock, D.A. Billmire and **S.A. Brugmann.** "Craniofacial syndromes: etiology, impact and treatment." Principles of Developmental Genetics, 2nd Edition. Ed. Sally A. Moody. Elsevier, New York. 2014. pp.654-671. ISBN:978-0-12-405945-0

2. M. Afshar, **S.A. Brugmann** and J.A. Helms. "Embryology of the craniofacial complex" Neligan Plastic Surgery 3rd Edition Vol. 2 Head & Neck. Ed. Joe Losee. Elsevier, New York. 2013. p.503.

3. **S.A. Brugmann**, R.R. Amasha, D.M. Gupta, J.A. Helms. "Craniofacial Patterning and Dysmorphologies" Craniofacial Growth and Development. Ed. Jeremy J. Mao and Hyun-duck Nah. Iowa State University Press. 2009. p.320.

4. J. A. Helms, **S.A. Brugmann**, and D.R. Cordero. "Shh and Other Genes and the Holoprosencephaly Malformation Sequence" Inborn Errors of Development Second Edition. Ed. Charles J. Epstein, Robert P. Erickson, and Anthony Wynshaw-Boris. Oxford University Press, London. 2008. p.291-300.

5. **S.A. Brugmann** and J.A. Helms. "Craniofacial formation and congenital defects" Principles of Developmental Genetics. Ed. Sally A. Moody. Elsevier, New York. 2007. p.656-679.

f. Invited talks

October, 2010: Deconstructing craniofacial development: Building a basis for understanding craniofacial disease. Invited by Mary Delany, Chair Animal Science Department, University of California, Davis.

November, 2011: Deconstructing craniofacial development: Building a basis for understanding craniofacial disease. Invited by Joshua Gross, Biology Department, University of Cincinnati.

February, 2012: Deconstructing craniofacial development: Building a basis for understanding craniofacial disease. Invited by Sally Moody, George Washington University.

February, 2013: The role of primary cilia in craniofacial development and disease. *Faculty CrossTalk, CCHMC*. Invited by Susa Wells.

February, 2014: Evolution of a new investigator's R01: from specific aim conception to funding. Invited by Susa Wells for the Linking Cancer and Development Seminar series CCHMC.

March, 2014: The cellular and molecular etiology of the craniofacial defects in the avian ciliopathic mutant, *talpid*^{2}*, Avian Model System Meeting, Cold Spring Harbor*

March, 2014: The molecular etiologies for craniofacial ciliopathies. *Gordon Research Conference for Craniofacial Morphogenesis and Tissue Engineering.* Invited by Andrea Streit, GRC organizer.

April 2014: The molecular etiologies for craniofacial ciliopathies. Invited by Trevor William, Chair in Craniofacial/Molecular Biology, University of Colorado, Denver

May 2014: The molecular etiologies for craniofacial ciliopathies. Invited by Walid Fakhouri, School of Dentistry, University of Texas, Houston

May 2014: The role of primary cilia during craniofacial development. Invited by Aaron Zorn, MDB Seminar Series, CCHMC.

August 2014: Generating human intestinal organoids with an enteric nervous system. *NCATS/DARPA Progress report meeting, Washington, DC*

October, 2014: GLI-dependent etiology of craniofacial ciliopathies. *Society for Craniofacial Genetics & Developmental Biology, La Jolla CA*

July, 2015: GLI-dependent etiology of craniofacial ciliopathies.
FASEB SRC: The Biology of Cilia and Flagella, Snowmass, CO

October, 2015: GLI-dependent etiology of craniofacial ciliopathies.
Hedgehog 2015: Puerto Varras, Chile

December, 2015: GLI-dependent etiology of craniofacial ciliopathies.
Invited by Deneen Wellik, Molecular Medicine and Genetics, University of Michigan

March, 2016: GLI-dependent etiology of craniofacial ciliopathies.
Invited by Robert Gagel, Brendan Lee, Jacqueline Hecht. Rolanette and Berdon Lawrence Bone Disease Program of Texas Bone Seminars, MD Anderson Cancer Center, Baylor College of Medicine and UT Health Science Center at Houston

September, 2016: GLI-dependent etiology of craniofacial ciliopathies.
Invited by Ophir Klein, Departments of Orofacial Sciences and Pediatrics, University of California at San Francisco

October, 2016: GLI-dependent etiology of craniofacial ciliopathies. *Midwest regional meeting of the Society for Developmental Biology, Ann Arbor, MI.* Invited by Scott Barolo.

November, 2016: GLI-dependent etiology of craniofacial ciliopathies.
Invited by Karl-Ferdinand Lechtreck, Cellular Biology, University of Georgia

April, 2017: GLI-dependent etiology of craniofacial ciliopathies.
Invited by Sally Moody, The George Washington University

June, 2017: Primary cilia and the development of the craniofacial complex. *Invited lecturer for the Cold Spring Harbor Mouse Development, Stem Cells & Cancer Course.* Invited by Tamara Caspary

November, 2017: Primary cilia and the development of the craniofacial complex. Invited by Johann Eberhart, University of Texas at Austin

December, 2017: Primary cilia and the development of the craniofacial complex. *American Society for Cell Biology*. Invited by Max Nachury, Peter Jackson and Jeremy Reiter

December, 2018: Ciliary heterogeneity and the development of the craniofacial complex. Invited by Labib Rouhana, Wright State University

April, 2019: Hand2 functions to synergistically activate Gli target genes in mandibular neural crest cells. *Molecular Signals in Craniofacial Development and Regeneration Session at AAA Experimental Biology.* Invited by Drs. Jun Wang and Ralph Marcucio

August, 2019: Etiology of Craniofacial ciliopathies: Past, present and future. *40th annual David W. Smith Workshop on Malformations and Morphogenesis.* Invited to be Keynote Speaker by Drs. Steve Braddock, John Carey and Robert Lipinski

October, 2019: Primary cilia and the development of the craniofacial complex. *26th annual Research Symposium, The School of Graduate Studies, Rutgers Biomedical and Health Sciences, Rutgers University.* Invited to be Keynote Speaker by Rutgers students.

November, 2019: Primary cilia and the development of the craniofacial complex. *Understanding Human Birth Defects in the Genomic Age.* Invited Speaker by Drs. Karen Liu, John Wallingford, Mustafa Khokha

February, 2020: Hand2 functions to synergistically activate Gli target genes in mandibular neural crest cells. *Gordon Research Conference for Craniofacial Morphogenesis and Tissue Regeneration*. Invited by Drs. Abigail Tucker and Pam Yelick

March 2020: Understanding the Etiology of craniofacial ciliopathies: what the talpid has taught us. *University of California, Davis.* Invited by Dr. Nadean Brown (Cancelled due to COVID-19)

April 2020: Understanding the Etiology of craniofacial ciliopathies: Past, Present and Future. *University of Southern California.* Invited by Dr. Amy Merrill (Cancelled due to COVID-19)

April 2020: Understanding the Etiology of craniofacial ciliopathies: Past, Present and Future. *Lawrence A. Tabak lecture in the Center of Oral biology 2020, University of Rochester School of Medicine and Dentistry*. Invited by Dr. Catherine Ovitt. (Cancelled due to COVID-19)

9. DISTRIBUTION OF EFFORT

90% Research
10% Teaching and teaching related activities

Samantha Brugmann, Ph.D.
Associate Professor Division of Developmental Biology; Department of Pediatrics
Associate Professor Division of Plastic Surgery; Department of Surgery
Cincinnati Children's Hospital Medical Center
Scientific Staff-Shriners Hospital for Children-Cincinnati