

## **PUBLICATIONS**

1. **Ammous Z**, Hackett NR, Butler MW, Raman T, Dolgalev I, O'Connor TP, Harvey BG, Crystal RG. Variability in small airway epithelial gene expression among normal smokers. *Chest* 2008 Jun; 133(6):1344-53.
2. Winters KA, Jiang Z, Xu W, Li S, **Ammous Z**, Jayakar P, Wierenga KJ. Re-assigned diagnosis of D4ST1-deficient Ehlers-Danlos syndrome (adducted thumb-clubfoot syndrome) after initial diagnosis of Marden-Walker syndrome. *Am J Med Genet A*. 2012 Nov; 158A(11):2935-40.
3. Aggarwal V, Dobrolet N, Fishberger S, Zablah J, Jayakar P, **Ammous Z**. PRKAG2 mutation: An easily missed cardiac specific non-lysosomal glycolipidosis. *Ann Pediatr Cardiol*. 2015 May-Aug; 8(2):153-6.
4. Morimoto M, Maguire E, **Ammous Z**, Song X, Pehlivan D, Lau C, Karaca E, Waller-Evans H, Holst CR, Chepa-Lotrea X, Macnamara E, Tos T, Isikay S, Nehrebecky M, Gonzaga-Jauregui C, Overton JD, Brigatti KW, Klein M, Markello TC, Posey JE, Adams DR, Puffenberger EG, Strauss KA, Lloyd-Evans E, Lupski JR, Gahl WA, Malicdan MCV. Bi-allelic CCDC47 variants cause a disorder characterized by woolly hair, liver dysfunction, dysmorphic features, and global developmental delay. *Am J Hum Genet*. 2018 Nov;103(5):794–807.

## **ABSTRACTS**

**Zeinab Ammous**, Neil R. Hackett, Marcus W. Butler, Tina Raman, Igor Dolgalev, Timothy P. O'Connor, Ben-Gary Harvey, and Ronald G. Crystal. Variability in Gene Expression Levels in Small Airway Epithelium of Cigarette Smokers. In: American Thoracic Society International Conference, May 18-23, 2007, San Francisco, California, USA.

**Zineb Ammous**, Kevin A. Winters, Parul Jayakar, Klaas Wierenga. Mutations of CHST14 in a new type of Ehlers-Danlos syndrome: Case report of a 16 year old patient with an initial presumptive diagnosis of Marden-Walker syndrome. In: American College of Medical Genetics Annual Clinical Genetics Meeting, March 27-31, 2012, Charlotte, North Carolina, USA.

**Zineb Ammous**, Aparana Rajadhyaksha, Parul Jayakar. Significance of the NODAL p.H165R variant in complex cardiovascular malformations: A report of two

lethal cases with an asymptomatic parent. In: American Society of Human Genetics Annual Meeting, November 6-10, 2012, San Francisco, California, USA

Rossana Sanchez-Russo, **Zineb Ammous**, Parul Jayakar. Novel Mutation in the ADAMTSL4 gene in a child with bilateral Ectopia Lentis and Aortic Root Dilatation: expanding the phenotype?. In: American Society of Human Genetics Annual Meeting, November 6-10, 2012, San Francisco, California, USA.

**Zineb Ammous**, Aparana Rajadhyaksha, Parul Jayakar. Microduplication of 3p25.2 containing RAF1 in a newborn with cystic hygroma suggestive of Noonan syndrome. In: American College of Medical Genetics Annual Clinical Genetics Meeting, March 19-23, 2013, Phoenix, Arizona, USA.

W. Thorson, **Z. Ammous**, Aparana Rajadhyaksha, Parul Jayakar. Early detection in Multiple Acyl-Coa Dehydrogenase Deficiency (MADD): a case report. In: American College of Medical Genetics Annual Clinical Genetics Meeting, March 19-23, 2013, Phoenix, Arizona, USA.

S. Taldone, **Z. Ammous**, W. Thorson, N. Sasaki, V. John, V. Villegas, D. Hess, A. Berrocal, S. Sacharow, Y. Fan, and D. Barbouth. Severe Form of Rubinstein Taybi Syndrome Identified by Cytogenomic Microarray. In: American College of Medical Genetics Annual Clinical Genetics Meeting, March 19-23, 2013, Phoenix, Arizona, USA.

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V. Aggarwal, N. Dobrolet, P. Jayakar, J. Zablach, **Z. Ammous**, S. Fishberger. HCM and WPW Syndrome With Life Threatening Arrhythmia In An Adolescent Female: PRKAG2 Mutation. In: Journal of Atrial Fibrillation, special issue, October 2013.

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Christopher Roberson, Rebecca Evans, Olivia Wenger, Heng Wang, Kevin Strauss, **Zineb Ammous**. A Web of Rural Genetics Clinics: Advancing into the Plain. In: American College of Medical Genetics Annual Clinical Genetics Meeting, March 24-28, 2015, Salt Lake City, Utah, USA.

**Zineb Ammous**, Angela Scheid, Rebecca Evans, Jeffrey Innis. Symptomatic Generalized Epilepsy Syndrome Associated with CACNA1A E1015K Variant. In: American College of Medical Genetics Annual Clinical Genetics Meeting, March 24-28, 2015, Salt Lake City, Utah, USA.

Angela Scheid, **Zineb Ammous**, Rebecca Evans. Expanding the clinical phenotype associated with the SMAD nuclear interacting protein 1 gene mutation E366G: report of two additional Amish patients with Psychomotor Retardation, Epilepsy, and Craniofacial Dysmorphism. In: American College of Medical Genetics Annual Clinical Genetics Meeting, March 24-28, 2015, Salt Lake City, Utah, USA.

Rebecca Evans, Angela Scheid, **Zineb Ammous**. Description of an Amish Mutation for Hypomyelinating Leukodystrophy Type 2. In: American College of Medical Genetics Annual Clinical Genetics Meeting, March 24-28, 2015, Salt Lake City, Utah, USA

**Zineb Ammous**, Christopher Roberson. Propionic Acidemia Screening in the Amish and Mennonite Populations. In: APHL Newborn Screening and Genetic Testing Symposium, Feb 29 – March 3, 2016, St Louis, Missouri, USA.

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**Zineb Ammous**, Olivia Wenger, Tom Herr, Rebecca Evans, Emma Baple, Andrew Crosby, Kevin Strauss. Psychomotor retardation, epilepsy, and craniofacial dysmorphism: Expanding the clinical phenotype. What have we learned from the Old Order Amish? In: American College of Medical Genetics Annual Clinical Genetics Meeting, March 21-25, 2017, Phoenix, Arizona, USA.

Marie Morimoto, Emily Maguire, **Zineb Ammous**, Xiaofei Song, Davut Pehlivan, C. Christopher Lau, Ender Karaca, Helen Waller-Evans, Charles R. Holst, Xenia Chepa-Lotrea, Ellen Macnamara, Tulay Tos, Sedat Isikay, Michele Nehrebecky,

Claudia Gonzaga-Jauregui, John D. Overton, Karlla W. Brigatti, Matthew Klein, Thomas C. Markello, Jennifer E. Posey, David R. Adams, Erik G. Puffenberger, Kevin A. Strauss, Emyr Lloyd-Evans, James R. Lupski, William A. Gahl, May Christine V. Malicdan. Biallelic loss-of-function variants in the calcium-binding protein encoding gene *CCDC47* cause a novel disease characterized by woolly hair, liver dysfunction, pruritus, dysmorphic features, and global developmental delay. In: American Society of Human Genetics Annual Meeting, October 16-20, 2018, San Diego, California, USA.