

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; 133:1344–1353 (First published online on March 13, 2008).
2. Winters KA1, 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. doi: 10.1002/ajmg.a.35613. Epub 2012 Sep 14.
3. Aggarwal V, Dobrolet N, Fishberger S, Zablah J, Jayakar P, **Ammous Z**. PRKAG2 mutation: an easily missed cardiac specific non-lysosomal glycogenosis. *Ann Pediatr Cardiol*. 2015 May-Aug;8(2): 153-6. doi: 10.4103/0974-2069.154149.

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

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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. Barbooth. 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.

Zineb Ammous, Aparana Rajadhyaksha, Parul Jayakar, Erick Hernandez, Olaf Bodamer. Severe chronic pancreatitis in propionic acidemia and treatment with liver transplantation. In: 12th International Congress of Inborn Errors of Metabolism, September 3-6, 2013 in Barcelona, Spain.

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.

Angela Scheid, **Zineb Ammous**. New CNS anomalies found in a patient with 16p11.2 microdeletion. In: American College of Medical Genetics Annual Clinical Genetics Meeting, March 25-29 2014, Nashville, Tennessee, USA.

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.

Zineb Ammous, Julia Szekely, Heng Wang, Baozhong Xin. Primary microcephalies (MCPH) and Seckel syndrome (SCKS) spectrum disorders: expanding the phenotype with a novel CENPJ mutation in the Old Order Amish. In: American College of Medical Genetics Annual Clinical Genetics Meeting, March 7-12, 2016, Tampa, Florida, USA

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