

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.
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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.
5. Bowser LE, Young M, Wenger OK, **Ammous Z**, Brigatti KA, Carson VJ, Moser T, Deline J, Aoki K, Morlet T, Scott EM, Puffenberger EG, Robinson DL, Hendrickson C, Salvin J, Gottlieb S, Heaps AD, Tiemeyer M, Strauss KA. Recessive GM3 synthase deficiency: Natural history, biochemistry, and therapeutic frontier. *Molec Genet Metab*. 2019 Jan; epub

ABSTRACTS

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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.

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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.

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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.