

## Publication List - Agilent SureSelect Platform

### Mendelian Diseases

- Rafiq MA, Kuss AW, Puettmann L, Noor A, Ramiah A, Ali G, Hu H, Kerio NA, Xiang Y, Garshasbi M, Khan MA, Ishak GE, Weksberg R, Ullmann R, Tzschach A, Kahrizi K, Mahmood K, Naeem F, Ayub M, Moremen KW, Vincent JB, Ropers HH, Ansar M, Najmabadi H. **Mutations in the Alpha 1,2-Mannosidase Gene, MAN1B1, Cause Autosomal-Recessive Intellectual Disability. Am J Hum Genet. 2011 Jul 15;89(1):176-82.**
- Sergouniotis PI, Davidson AE, Mackay DS, Li Z, Yang X, Plagnol V, Moore AT, Webster AR. **Recessive Mutations in KCNJ13, Encoding an Inwardly Rectifying Potassium Channel Subunit, Cause Leber Congenital Amaurosis. Am J Hum Genet. 2011 Jul 15;89(1):183-90.**
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- Rope AF, Wang K, Evjenth R, Xing J, Johnston JJ, Swensen JJ, Johnson WE, Moore B, Huff CD, Bird LM, Carey JC, Opitz JM, Stevens CA, Jiang T, Schank C, Fain HD, Robison R, Dalley B, Chin S, South ST, Pysher TJ, Jorde LB, Hakonarson H, Lillehaug JR, Biesecker LG, Yandell M, Arnesen T, Lyon GJ. **Using VAAST to identify an X-Linked Disorder Resulting in Lethality in Male Infants Due to N-Terminal Acetyltransferase Deficiency. Am J Hum Genet. 2011 Jun 22.**

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## Complex Diseases

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