



Compiled by @b12unme

Inborn errors of B12 metabolism

2003: Transcobalamin and methionine synthase reductase mutated polymorphisms aggravate the risk of neural tube defects in humans.

<http://www.ncbi.nlm.nih.gov/m/pubmed/12812837/>

2005: Transcobalamin 776C3G polymorphism negatively affects vitamin B-12 metabolism¹⁻³

<http://ajcn.nutrition.org/content/81/6/1436.full.pdf>

2008: Common variants of FUT2 are associated with plasma vitamin B12 levels

<http://www.ncbi.nlm.nih.gov/pmc/articles/PMC2673801/pdf/nihms86874.pdf>

2009: Genome-wide significant predictors of metabolites in the one-carbon metabolism pathway. (TCN1)

<http://www.ncbi.nlm.nih.gov/pubmed/19744961?dopt=Abstract>

2010: Genetic disorders of vitamin B12 metabolism: eight complementation groups – eight genes

<http://www.ncbi.nlm.nih.gov/pmc/articles/PMC2995210/pdf/S1462399410001651a.pdf>

2012: Structural features of recombinant MMADHC isoforms and their interactions with MMACHC, proteins of mammalian vitamin B12 metabolism.

<http://www.ncbi.nlm.nih.gov/m/pubmed/22832074/>

2012: Mutations in ABCD4 cause a new inborn error of vitamin B12 metabolism.

<http://www.ncbi.nlm.nih.gov/m/pubmed/22922874/?i=4&from=b12%20AND%20abcd4>

2013: Molecular and cellular effects of vitamin B12 in brain, myocardium and liver through its role as co-factor of methionine synthase.

<http://www.ncbi.nlm.nih.gov/m/pubmed/23415654/?i=2&from=mtr%20and%20b12>

2013: Lessons in biology from patients with inborn errors of vitamin B12 metabolism.

<http://www.ncbi.nlm.nih.gov/pubmed/23402785>

2013: Vitamin B12 absorption: mammalian physiology and acquired and inherited disorders.

<http://www.ncbi.nlm.nih.gov/pubmed/23178706>

2013: Proteomics of vitamin B12 processing.

<http://www.ncbi.nlm.nih.gov/pubmed/23241609>

2013: Cobalt: its role in health and disease.

<http://www.ncbi.nlm.nih.gov/pubmed/24470095>

2013: Disorders of Intracellular Cobalamin Metabolism

<http://www.ncbi.nlm.nih.gov/books/NBK1328/>

2013: Genetic architecture of vitamin B12 and folate levels uncovered applying deeply sequenced large datasets.

<http://www.ncbi.nlm.nih.gov/m/pubmed/23754956/>

2013: An X-linked cobalamin disorder caused by mutations in transcriptional coregulator HCFC1.

<http://www.ncbi.nlm.nih.gov/m/pubmed/24011988/>

2014: Methyl nutrients, DNA methylation, and cardiovascular disease

<http://onlinelibrary.wiley.com/store/10.1002/mnfr.201200636/asset/mnfr1988.pdf?v=1&t=htj3r634&s=8acdb92f372d4284e52e3dda5280a14a39201e99>

2014: Maternal micronutrient imbalance alters gene expression of BDNF, NGF, TrkB and CREB in the offspring brain at an adult age.

<http://www.ncbi.nlm.nih.gov/m/pubmed/24462543/>

2014: A novel mutation of the transcobalamin II gene in an infant presenting with hemophagocytic lymphohistiocytosis.

<http://www.ncbi.nlm.nih.gov/m/pubmed/24563082/>