



Maple Syrup Urine Disease: Pathophysiology Management, and Outcomes in the Modern Era

Kevin A. Strauss, MD

Medical Director, Clinic for Special Children

Wednesday, July 15, 2020

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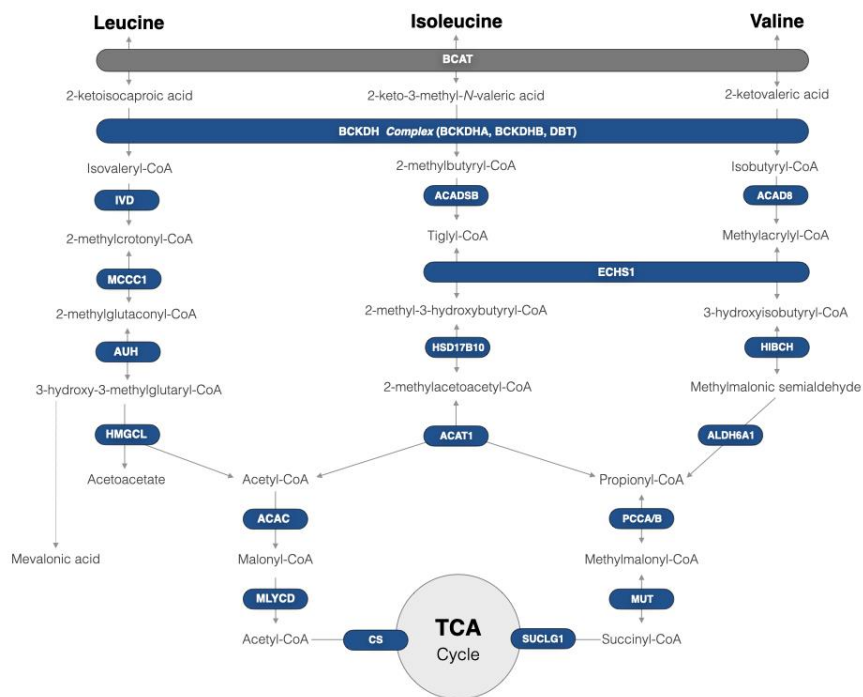


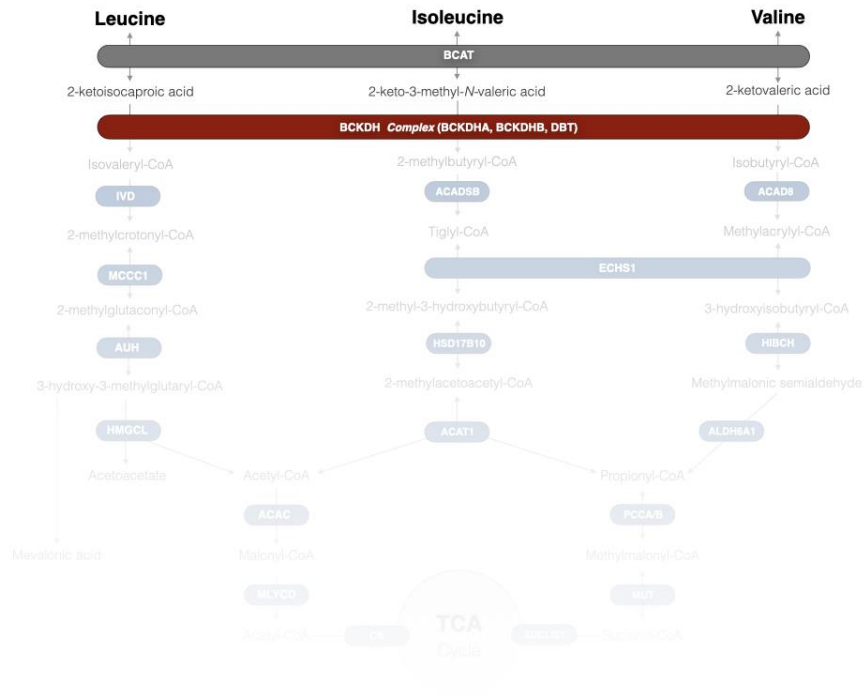
Learning Objectives



1. Understand the pathophysiology of MSUD and the theory behind management with medical food.
2. Identify the components of management that are the primary contributors to good metabolic control in MSUD.
3. Apply learnings to develop nutrition interventions in one's own clinical setting and practice to contribute to positive outcomes for MSUD patients.

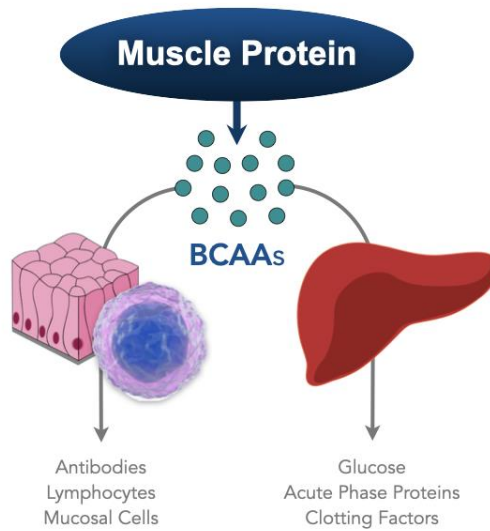
pathophysiology



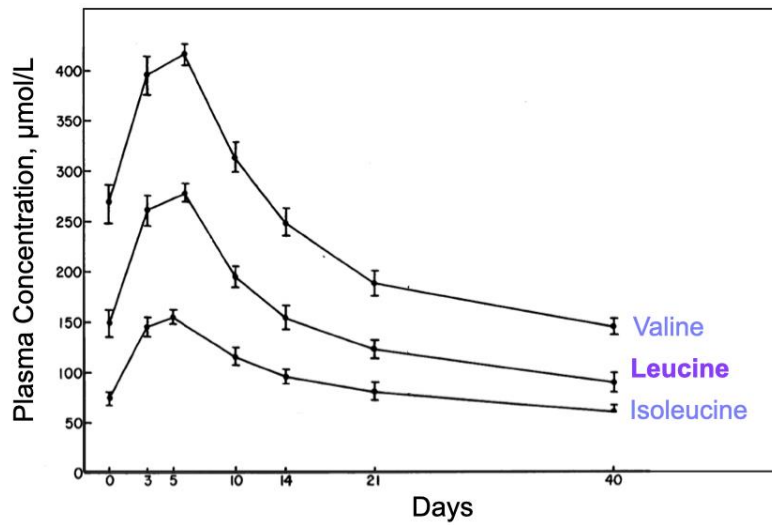


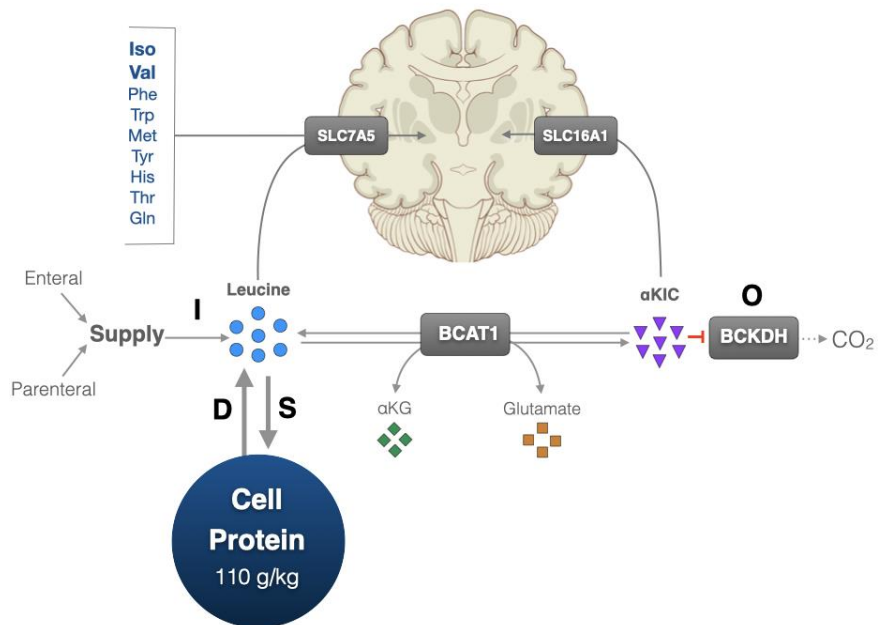
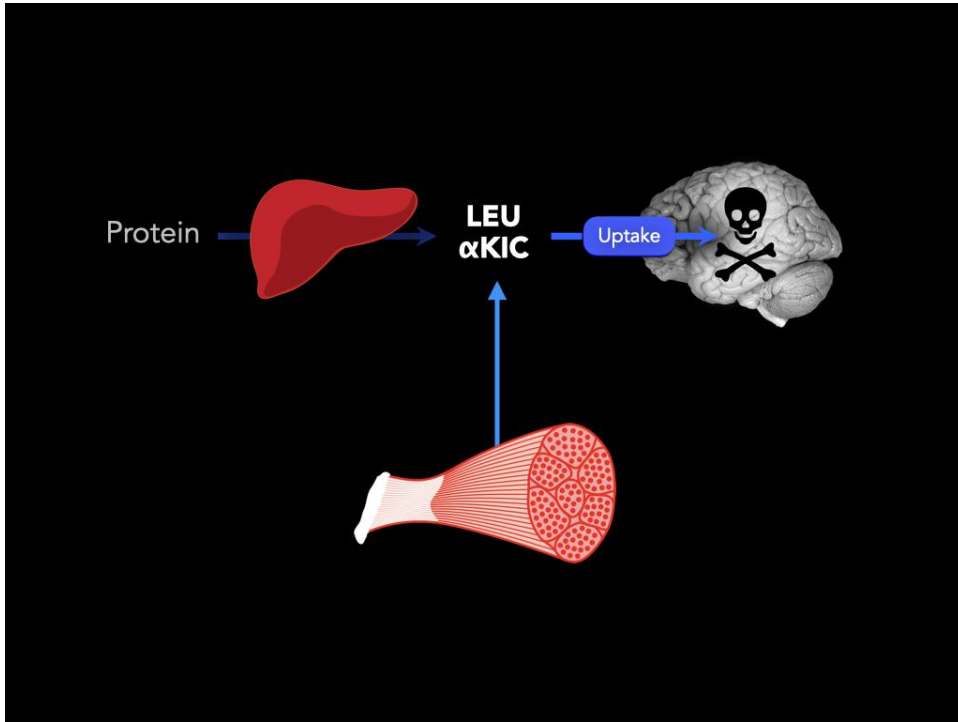
a c u t e
neurotoxicity

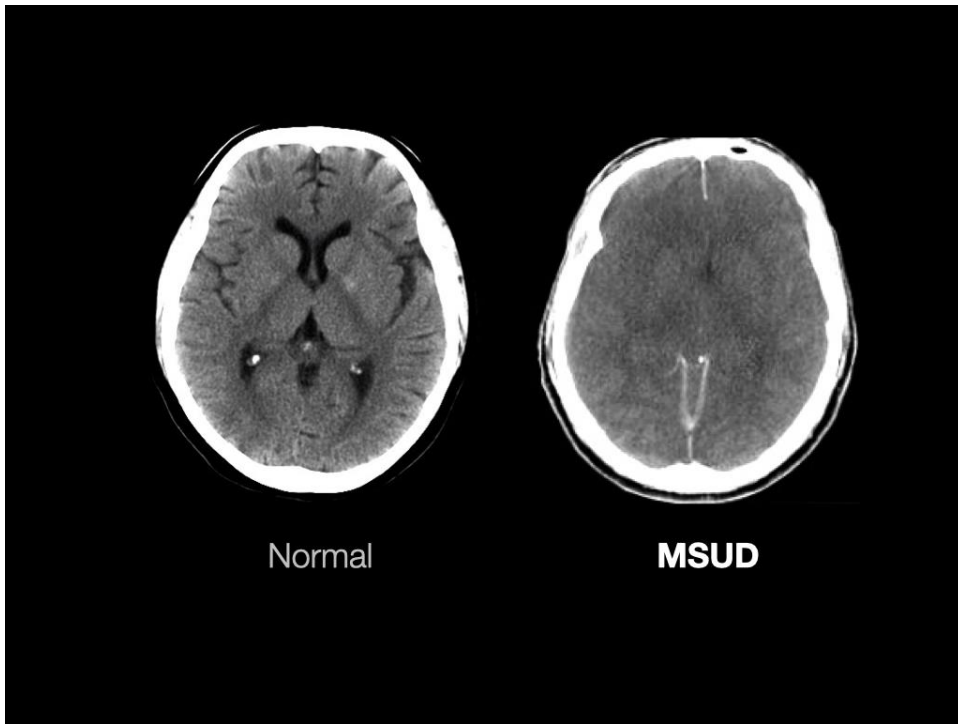
Catabolic 'Stress' Response



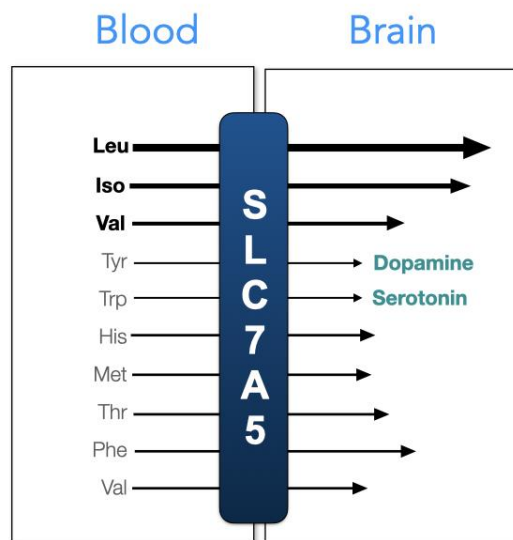
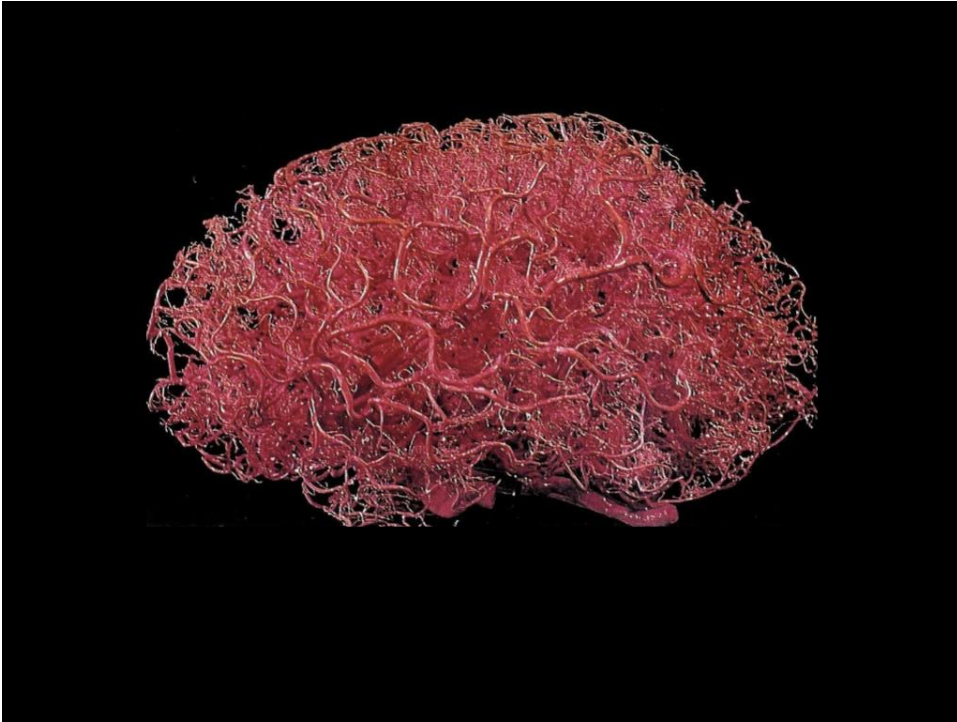
BCAA Release During Catabolism

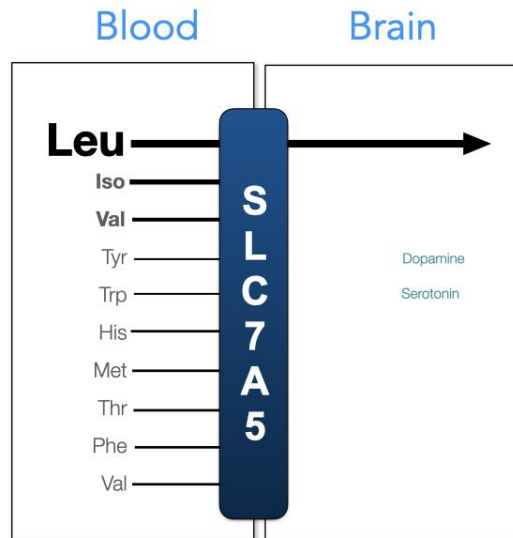


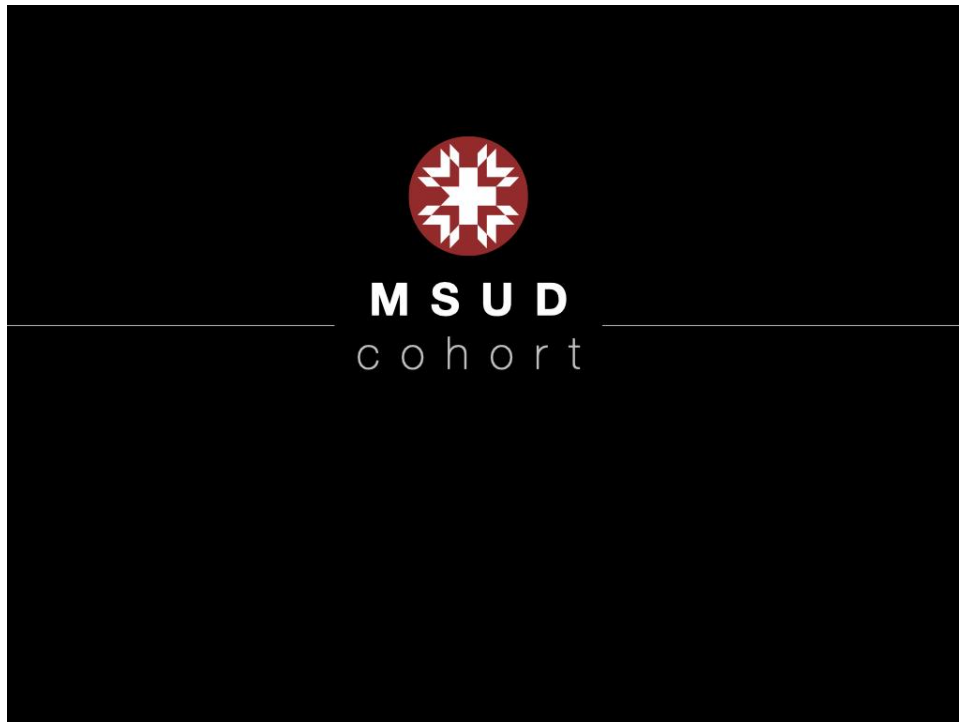




c h r o n i c
— n e u r o t o x i c i t y —







2020: 50-year Natural History Study

► *Mol Genet Metab*. 2020 Mar;129(3):193–206. doi: 10.1016/j.jymgme.2020.01.006. Epub 2020 Jan 16.

Branched-chain α -ketoacid dehydrogenase deficiency (maple syrup urine disease): Treatment, biomarkers, and outcomes

Kevin A Strauss¹, Vincent J Carson², Kyle Soltys³, Millie E Young⁴, Lauren E Bowser⁴, Erik G Puffenberger⁴, Karla W Brigatti⁴, Katie B Williams⁴, Donna L Robinson⁴, Christine Hendrickson⁴, Keturah Beller⁴, Cora M Taylor⁵, Barbara Haas-Givler⁵, Stephanie Chopko⁶, Jennifer Hailey⁷, Emilie R Muelly⁸, Diana A Shellmer³, Zachary Radcliff⁹, Ashlin Rodrigues⁴, Kalynn Loeven⁴, Adam D Heaps⁴, George V Mazariegos³, D Holmes Morton¹⁰

Affiliations

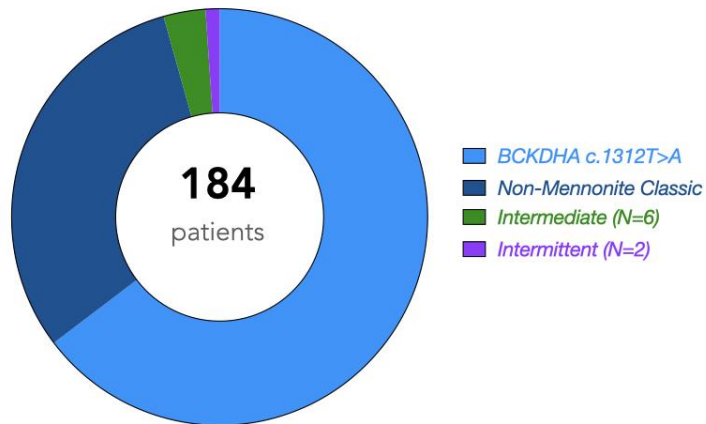
Affiliations

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- 6 Department of Pediatrics, Nemours Alfred I. duPont Hospital for Children, Wilmington, DE, USA; Sidney Kimmel Medical College, Thomas Jefferson University, Philadelphia, PA, USA.
- 7 Wellspan Philhaven, Mount Gretna, PA, USA.
- 8 Department of Internal Medicine, The Permanente Medical Group, Santa Clara, CA, USA.
- 9 Department of Pediatrics, Nemours Alfred I. duPont Hospital for Children, Wilmington, DE, USA.
- 10 Clinic for Special Children, Strasburg, PA, USA; Department of Pediatrics, Penn Medicine-Lancaster General Hospital, Lancaster, PA, USA; Central Pennsylvania Clinic, Belleville, PA, USA.

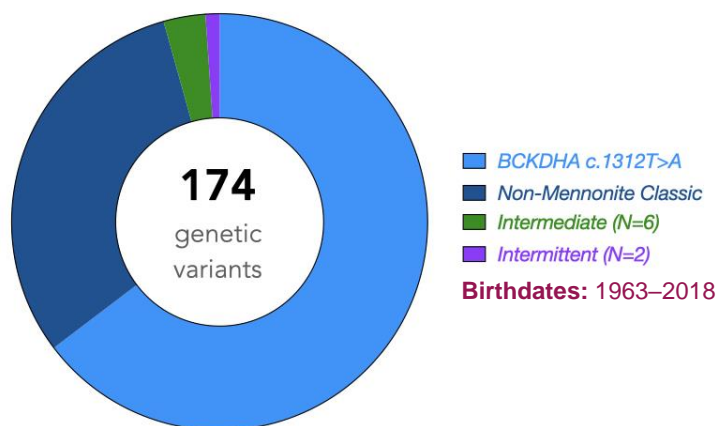
PMID: 31980395 DOI: 10.1016/j.jymgme.2020.01.006

KA Strauss et al., *Mol Genet Metab* 2020

Clinic for Special Children Cohort (1989–2020)



Clinic for Special Children Cohort (1989–2020)



d i e t a r y m a n a g e m e n t

2005: Novel Medical Formula

Clinical Trial > [Mol Genet Metab.](#) 2010 Apr;99(4):333-45. doi: 10.1016/j.ymgme.2009.12.007.

Epub 2010 Jan 12.

Classical maple syrup urine disease and brain development: principles of management and formula design

Kevin A Strauss¹, Bridget Wardley, Donna Robinson, Christine Hendrickson, Nicholas L Rider, Erik G Puffenberger, Diana Shellmer, Ann B Moser, D Holmes Morton

Affiliations — collapse

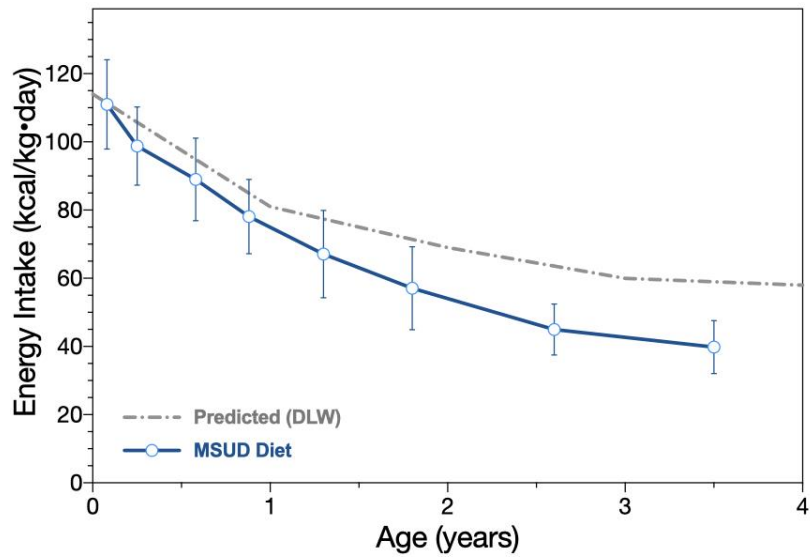
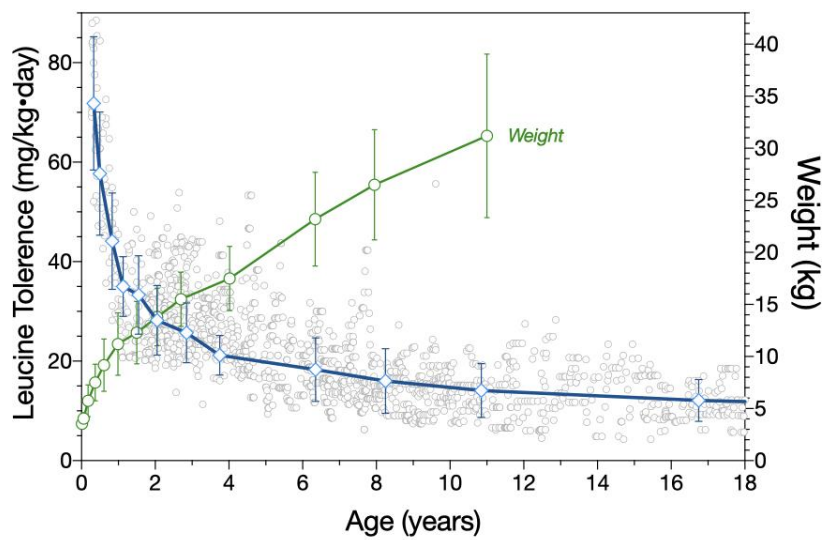
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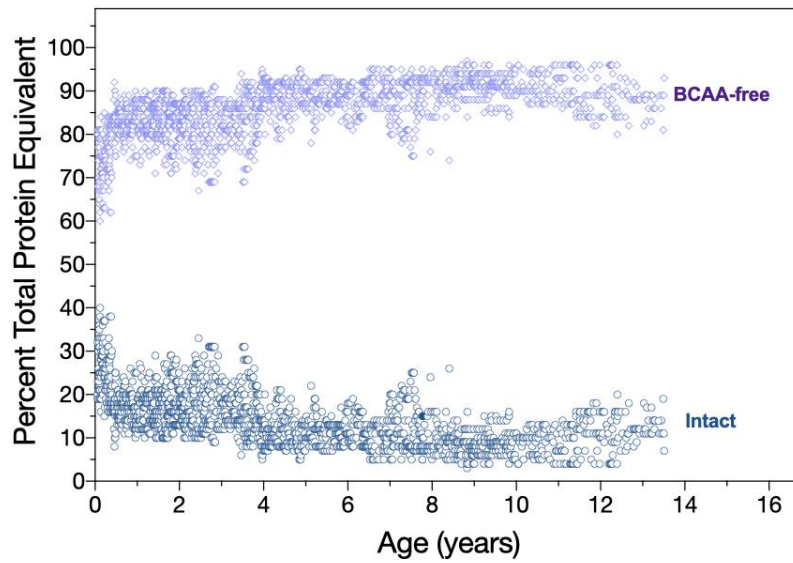
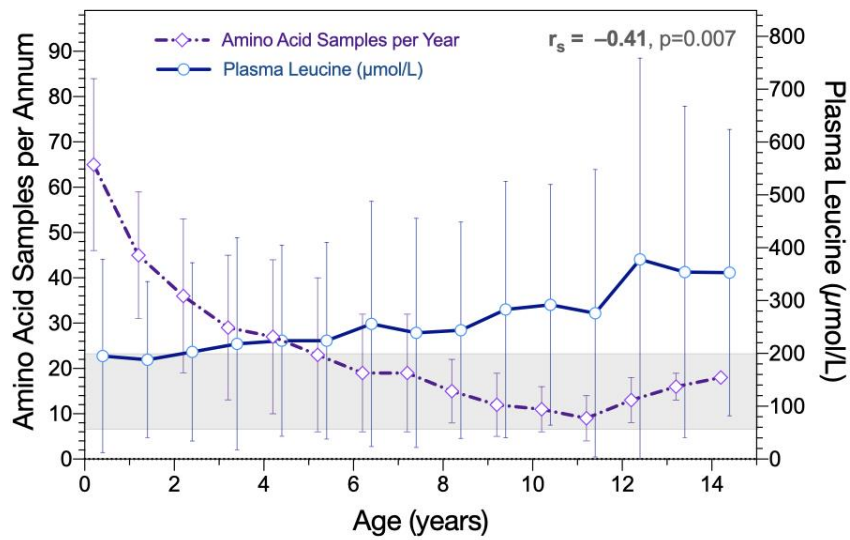
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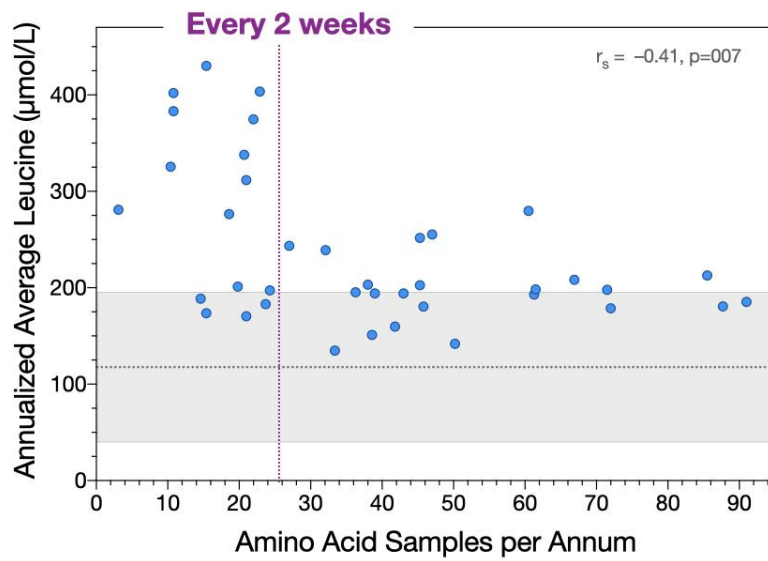
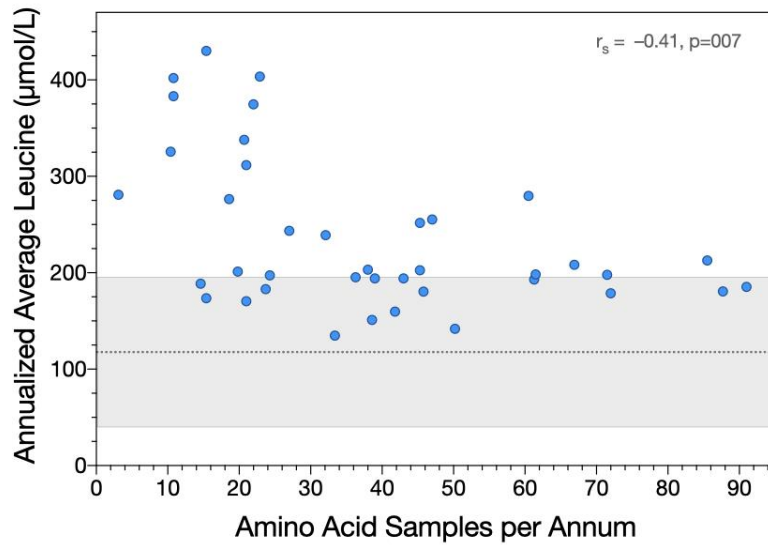
PMID: 20061171 PMCID: PMC3671925 DOI: 10.1016/j.ymgme.2009.12.007

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Formula Cohort: N=41, DOB 2005–2018

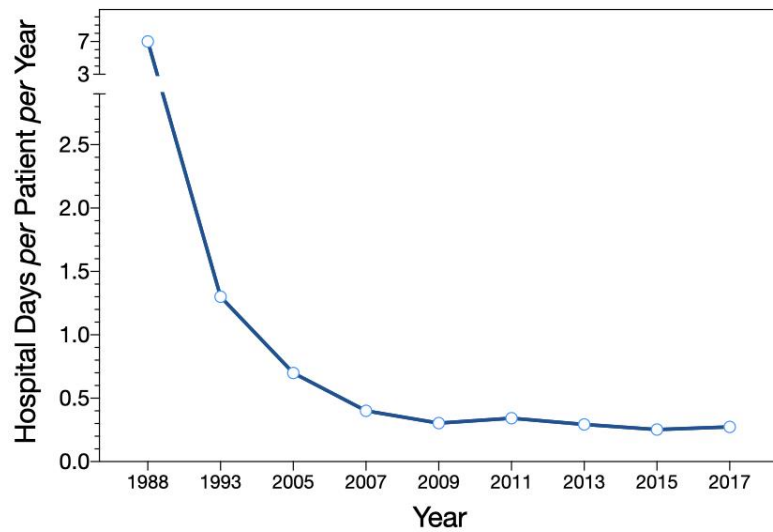
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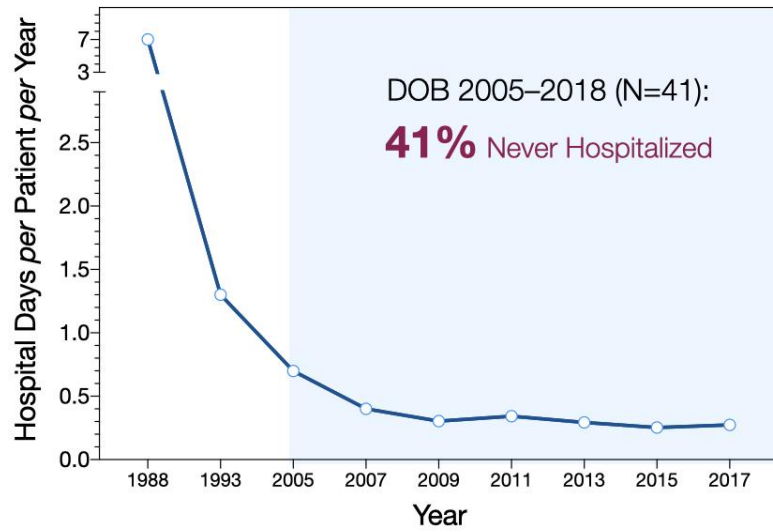
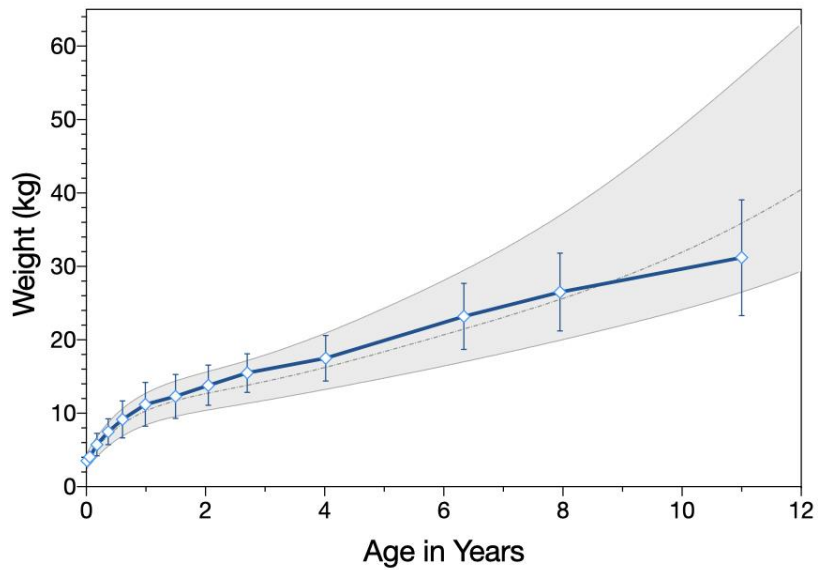


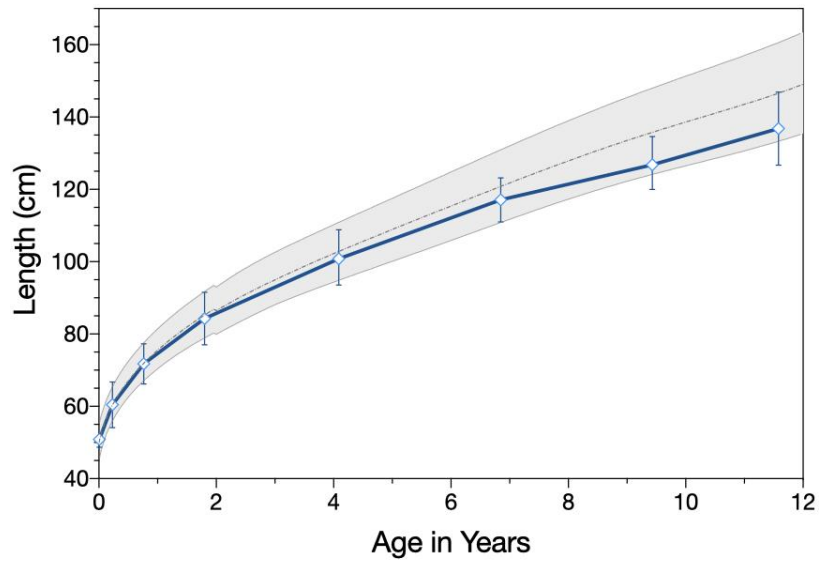
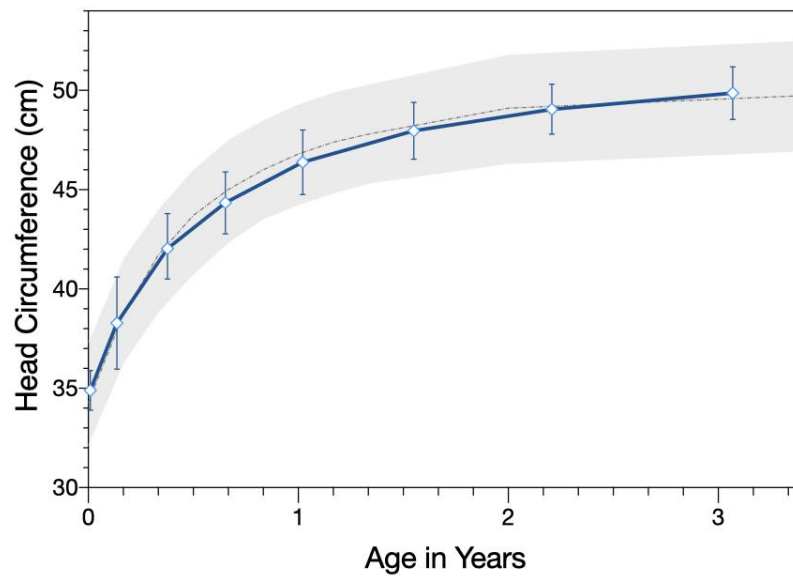
outcomes

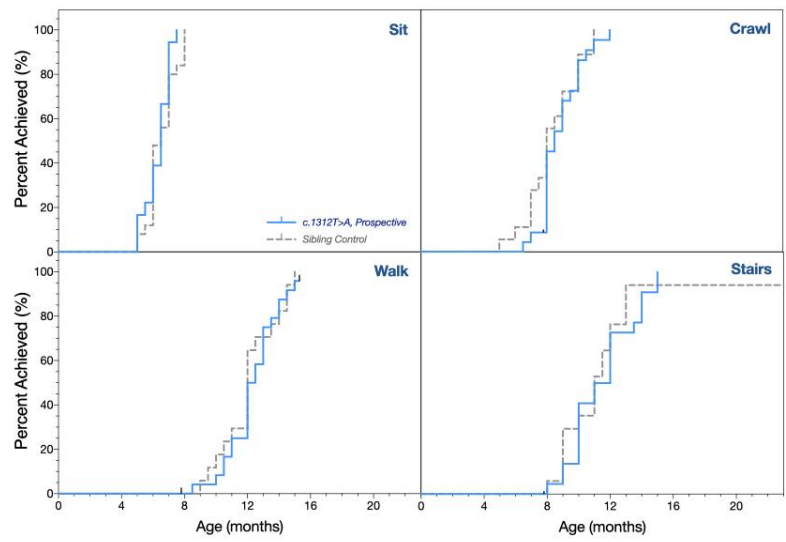
dietary cohort



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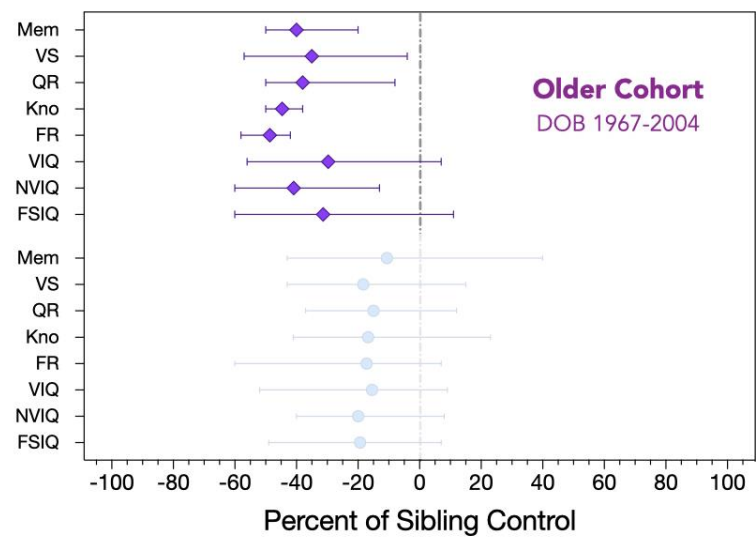
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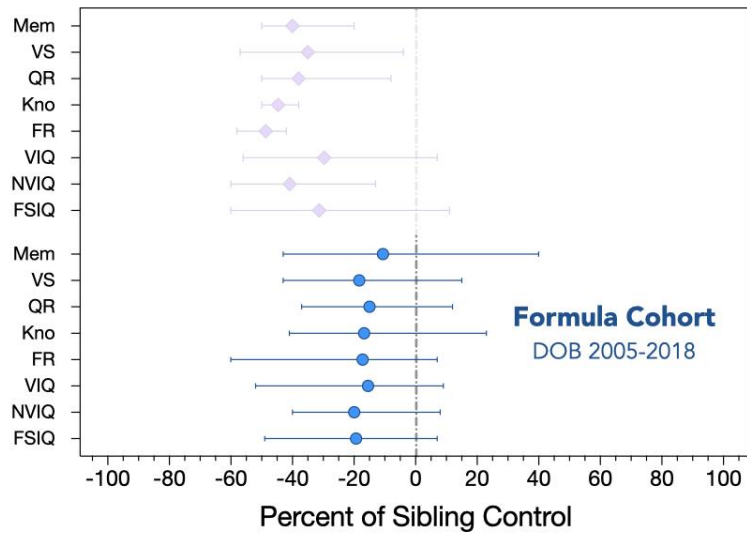
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Stanford-Binet Intelligence Scales



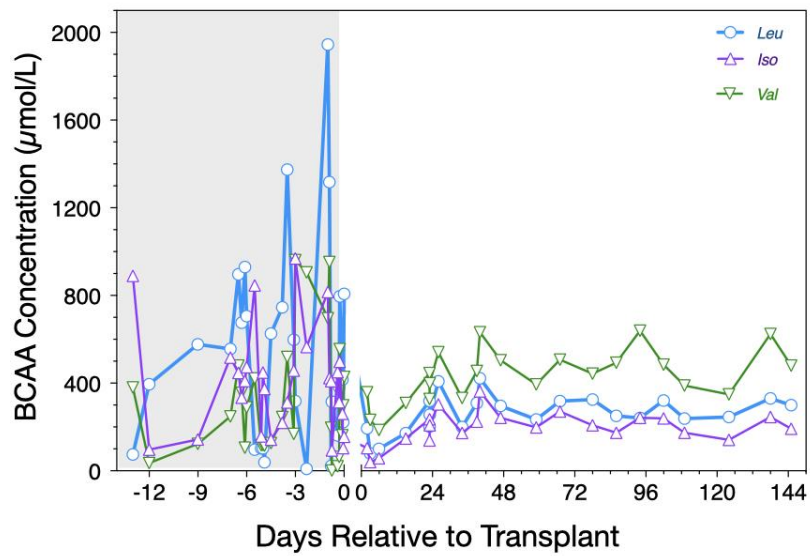
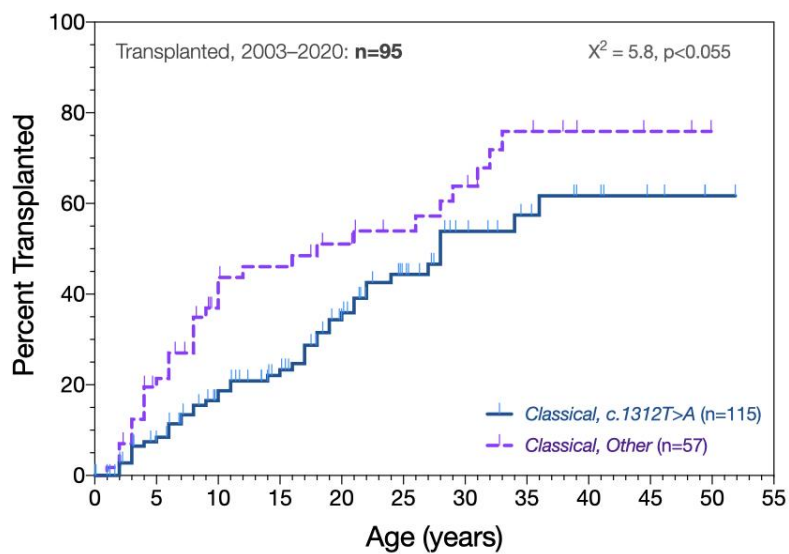
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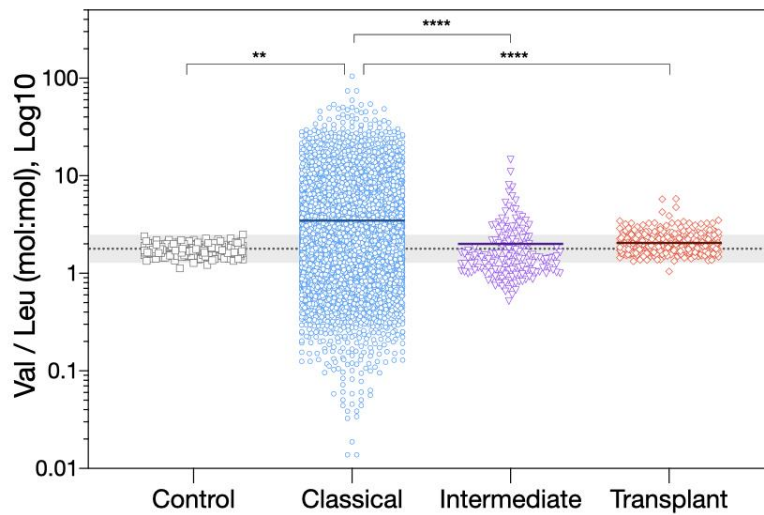
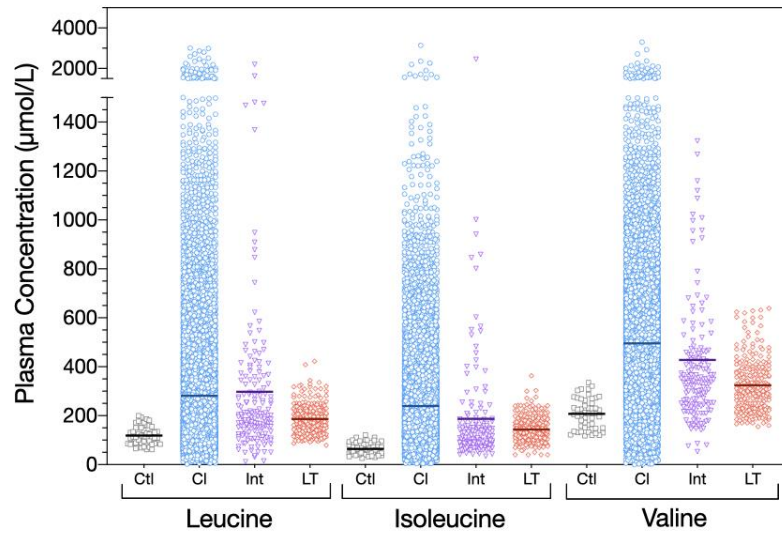
Stanford-Binet Intelligence Scales

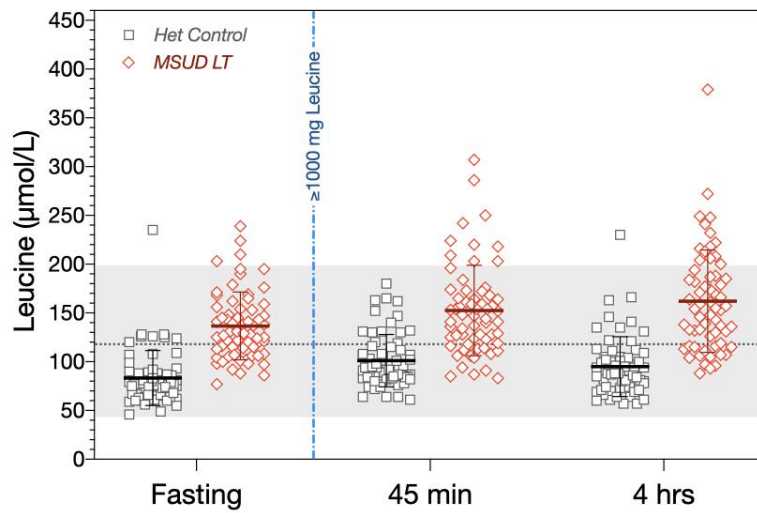
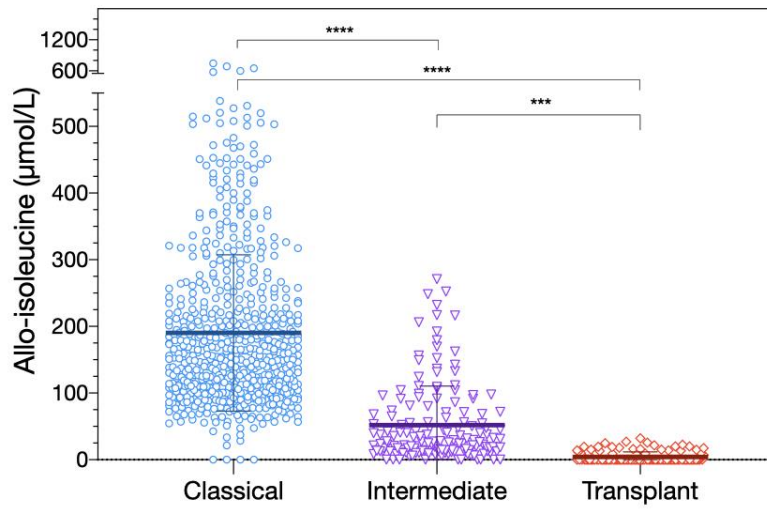


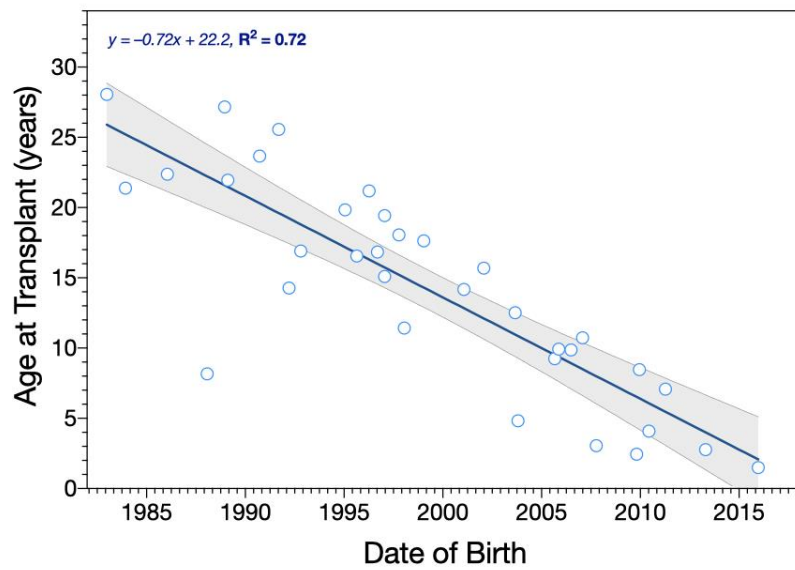
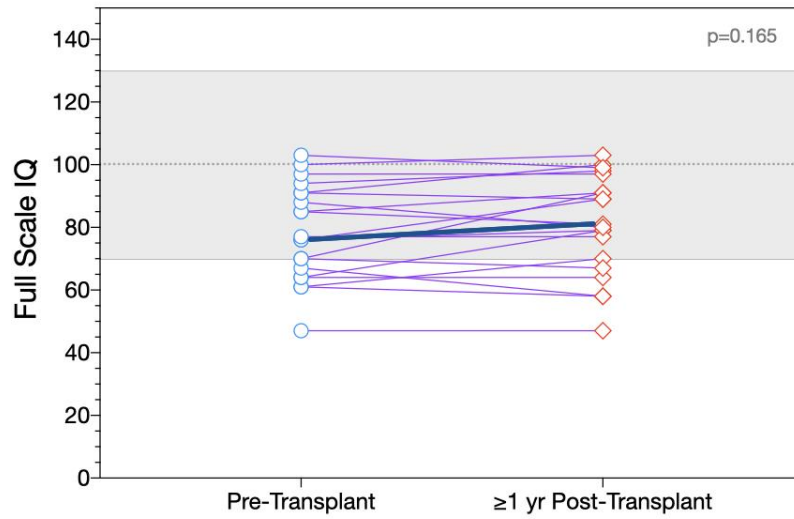
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liver
transplant

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Thank you! Questions?

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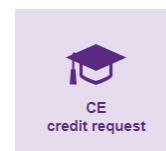
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