Resolving catatonia with vitamin infusions? Late-onset cobalamin C deficiency diagnosed through medical evaluation for progressive catatonia

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Background

Cobalamin C (CblC) deficiency is an inborn error of metabolism leading to impaired ability to convert dietary vitamin B12 into its active forms, which manifests as neurological, psychiatric, and renal pathology.[1]

Case

Early 20s male with several months of progressive catatonia

- No premorbid psychiatric history
- PMH: chronic kidney disease, gout
- Initial BFCRS 12
  - Immobility, mutism, staring, rigidity, withdrawal, fever
- Brisk reflexes, bilateral positive Hoffman's sign, ankle clonus, rash from mites
- Leukopenia (low of 1.1 10^3/μL)
- MRI/MRA/MRV Brain – interval mild parenchymal loss (over 1 month)
- EEG - mild encephalopathy
- Broad CSF studies - largely unremarkable
  - Acetylcholine receptor antibody mildly elevated (0.22 nmol/L)

Empiric high dose IV steroids and IVIG for initially presumed autoimmune encephalitis → no improvement.

Modest improvement (BFCRS 12 → 4) with:

- Lorazepam (up to 16 mg/day IV)
- Memantine (20 mg/day) transitioned to amantadine (400 mg/day)
- 2 sessions of ECT – complicated by post-ECT seizure

ECT and amantadine were discontinued in the setting of new onset generalized tonic-clonic seizures, which were stabilized on levetiracetam and lacosamide

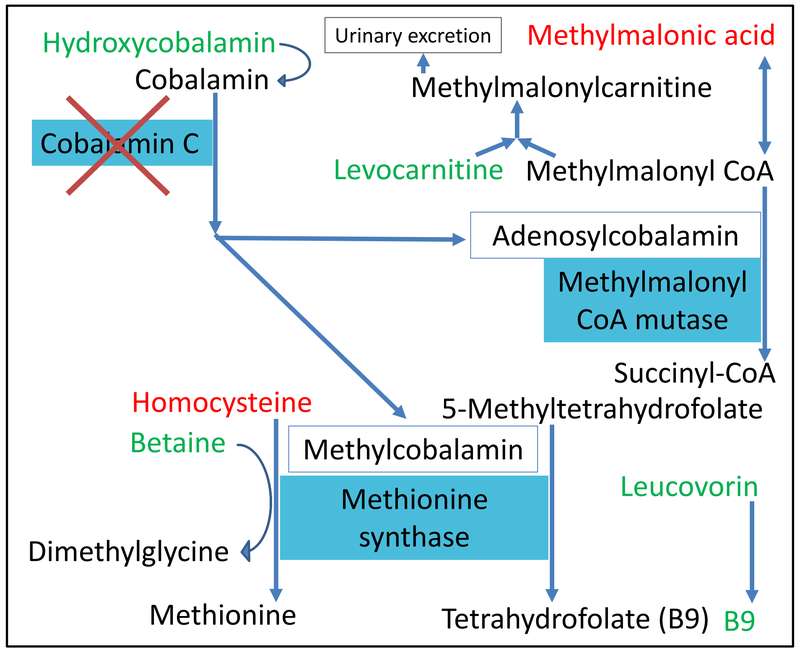

Figure 1: Key toxic metabolites and supplementary treatments for cobalamin C deficiency

Urine amino acid analysis - highly elevated homocysteine
Subsequent serum testing:

- **Homocysteine** (>50.0 umol/L; normal range 0-15)
- **Methylmalonic** acid (452,000 nmol/L; normal range 87-318)

Genetics consultation clarified cobalamin C deficiency diagnosis

Eventual full resolution of catatonia and other neuropsychiatric symptoms (cognitive baseline) with vitamin infusions (see Fig 1):

- **Hydroxocobalamin** (injectable B12; increases cobalamin)
- **Leucovorin** (injectable B9; bypasses folate synthesis)
- **Betaine** (decreases homocysteine)
- **Levocarnitine** (decreases methylmalonic acid)

Discussion

High index of clinical suspicion prompted multisystem testing for medical etiology of catatonia, revealing a metabolic etiology that was treatable with vitamin infusions. Late-onset adult CblC deficiency is rarer than early onset presentations, but cases can feature neurological, behavioral, renal, hematological, and dermatologic findings.[2] Case reports describe cognitive decline, psychosis, and abnormal neurological exams,[3,4] but this is the first case of CblC deficiency demonstrating catatonia.

Conclusions

Catatonia is a rare presentation of inborn errors of metabolism, and amino acid analysis can aid in screening during evaluation for medical etiology of catatonia. In such cases vitamin repletion may lead to resolution of catatonia.

References

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