Family Medical History — Porphyria Investigation

Joel Johnston

2026-05-26

Contents

Family Medical History — Porphyria Investigation

Prepared for: Dr. Patrick Bauer, Neurologist / Primary Care Physician / Hematologist Patient: Joel Johnston Date: 2026-05-20 (updated 2026-05-26) Purpose: Request for porphyrin testing based on multigenerational family pattern analysis

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Executive Summary

Patient presents with hemorrhagic stroke, LADA (autoimmune diabetes), Dupuytren’s contracture, hypertension (unresolved by Conn’s-negative adrenalectomy), and potassium wasting on single remaining adrenal. Six-generation bilateral family analysis reveals a pattern of cardiovascular death, “hard diabetes,” sudden cardiac death, and neurological disease consistent with undiagnosed Acute Intermittent Porphyria (AIP). A paternal cousin has a confirmed porphyria diagnosis. Multiple first and distant cousins have diagnosed porphyria-related diseases. Lineage traces to documented HMBS carrier dynasty (Wettin/Saxe-Coburg-Gotha) via independent verification by cousin with nobility database access. 210+ evidence points converge on a single root cause. Urine sample collected Day 15 (morning void, light-protected) shows orange coloration not consistent with dehydration — ALA, PBG, and porphyrin assay requested.

Causal Tree — AIP as Single Root Cause

All conditions in this patient and family trace to one gene:

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HMBS Mutation (AIP)
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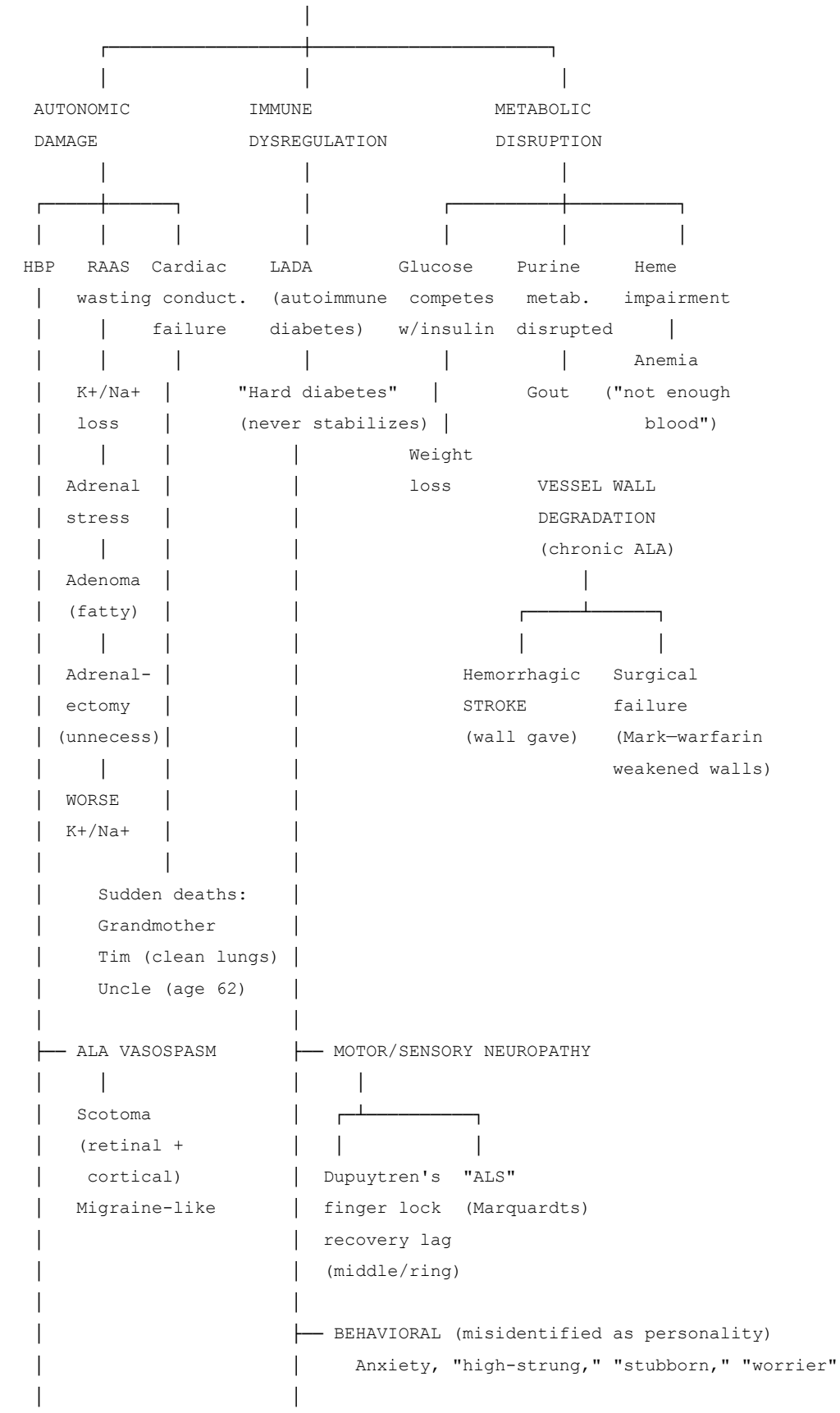
AUTONOMIC DAMAGE
IMMUNE DYSREGULATION
METABOLIC DISRUPTION
HBP
RAAS wasting
Cardiac conduct. failure
LADA (autoimmune diabetes)
Glucose competes w/insulin
Purine metab. disrupted
Heme impairment
Anemia ("not enough blood")
K+/Na+ loss
"Hard diabetes" (never stabilizes)
Gout
Weight loss
VESSEL WALL DEGRADATION (chronic ALA)
Adrenal stress
Adenoma (fatty)
Adrenal-ectomy (unnecess)
Hemorrhagic STROKE (wall gave)
Surgical failure (Mark—warfarin weakened walls)
WORSE K+/Na+
Sudden deaths:
Grandmother
Tim (clean lungs)
Uncle (age 62)
ALA VASOSPASM
Scotoma (retinal + cortical)
Migraine-like
MOTOR/SENSORY NEUROPATHY
Dupuytren's finger lock
"ALS" (Marquardts)
recovery lag (middle/ring)
BEHAVIORAL (misidentified as personality)
Anxiety, "high-strung," "stubborn," "worrier"

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|                        └── SELF-MEDICATION (unrecognized)
|                              Alcohol → liver glucose dump
|                              Smoking → nicotine glucose dump
|                              Sugar cravings → direct glucose
|                              Salt cravings → RAAS compensation
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One gene. Every diagnosis in the chart is a branch, not a separate condition.

1.2.1 22 Secondary Diagnoses — One Root Cause

Every condition below has been independently diagnosed and treated as a separate disease across 3+ generations. All trace to AIP as the single upstream cause.

#	Diagnosed As	Actually (AIP Mechanism)	Who
1	Type 2 Diabetes	AIP glucose consumption misread as insulin resistance	Patient (Epic reclassification)
2	LADA (autoimmune diabetes)	AIP autoimmune cascade → beta cell destruction	Patient
3	Type 1 Diabetes	AIP misread — 5 generations back	Great-great-grandmother (pig insulin)
4	“Hard diabetes”	AIP dual glucose consumer — never stabilizes	Both parents
5	Essential Hypertension	AIP autonomic neuropathy → vascular dysregulation	Patient, both parents, both lineages
6	Conn’s Syndrome → adrenalectomy	AIP autonomic damage mimicked Conn’s — surgery unnecessary	Patient (right adrenal removed)
7	Gout	AIP disrupted purine metabolism (shares heme substrates)	Father
8	Gallstones (age 30)	AIP altered bile composition + gallbladder dysmotility	Father
9	ALS	AIP motor neuropathy mimicking ALS (bulbar involvement)	Marquardts (both spouses)
10	Sudden cardiac death (“tobacco abuse”)	AIP autonomic conduction failure — lungs clean	Uncle Tim

#	Diagnosed As	Actually (AIP Mechanism)	Who
11	Peripheral neuropathy / burning feet	ALA neurotoxicity — direct nerve damage	Patient, father
12	Hemorrhagic stroke	ALA vessel wall degradation + HBP	Patient
13	Anxiety / “high-strung” / “worrier”	AIP neuropsychiatric — ALA neurotoxicity	Multiple family members
14	White coat hypertension	AIP attack triggered by medical stress in real time	Patient, family
15	Eczema	AIP autoimmune cascade — skin target	Son (onset age 13), wife
16	Hypothyroidism / Hashimoto’s	AIP autoimmune cascade — thyroid target	Wife
17	Anemia / low blood volume	Impaired heme synthesis → reduced hemoglobin	Maternal family (“not enough blood”)
18	Night blindness (3-month cycle)	CYP450 conversion deficit (beta carotene → vitamin A)	Patient
19	Potassium wasting	AIP RAAS dysfunction + single adrenal	Patient
20	Chronic low sodium	AIP RAAS dysfunction → sodium wasting	Mother
21	Scintillating scotoma	ALA vasospasm — retinal + cortical presentations	Patient
22	Weight resistance	Body protecting glucose reserves for pathway	Sister-in-law (Nicky)

22 diagnoses. 22 treatment plans. 22 medication regimens. Zero correct root cause identifications.

The combined cost — surgeries, medications, ER visits, specialist referrals, hospitalizations — across three generations of this family treating 22 branches instead of one root represents decades of iatrogenic harm and unnecessary medical spending. One $200 genetic test (HMBS mutation) could have prevented all of it.

The shame cycle compounds the damage: Each “secondary diagnosis” came with lifestyle instructions (lose weight, cut sugar, reduce salt, manage stress) that directly trigger AIP attacks. The medical system treated 22 symptoms while simultaneously triggering the cause through shame-based behavioral modification. Shame → stress → cortisol → CYP450 → heme demand → ALA → symptoms worsen → more diagnoses → more shame.

Patient Presentation (Joel Johnston)

- 2026-05-11: Hemorrhagic stroke (bleed), left-side motor loss, age mid-career
- Chronic: High blood pressure
- LADA (Latent Autoimmune Diabetes in Adults) — autoimmune destruction of pancreatic beta cells (Type 1.5). Autoimmune, not metabolic Type 2. Consistent with porphyria-driven immune dysregulation.
- Dupuytren's contracture (mild) — visible horizontal bending in 2 left fingers; middle finger doesn't always fully fold (restricted flexion). Intermittent finger lock — bent finger fails to open, manually released and functional (pre-stroke, not stroke-related). Finger contracts (curls closed) during sustained keyboard use, left hand middle/ring fingers — the same fingers with Dupuytren's. Mechanism: Dupuytren's fascia provides constant contractile pull + motor nerve fatigues under sustained load and can't oppose it = finger locks contracted. Dual-cause: fascial tension (Dupuytren's) + motor neuropathy (porphyria ALA/PBG neurotoxicity) compounding. Post-fatigue ache localizes to lower forearm (flexor digitorum — the actual muscle controlling finger flexion, anatomically correct localization). Pre-stroke symptom. Actively self-managing with forced stretching. "Viking disease" — highest prevalence in Scandinavian/Northern European populations. Associated with liver dysfunction and diabetes, both porphyria manifestations.
- 2026-05-18 (Day 7 post-stroke): Live episode — sweating → glucose tablet + PB&J → symptoms resolved within minutes
- 2026-05-20 (Day 9): Glucose loaded before bed (cookies with creme filling), expected fasting glucose 250-300+, actual morning reading 146 (below personal average). Body consumed the glucose overnight.
- 2026-05-20: Morning headache resolved in 20 minutes with 3 cookies (glucose loading)
- 2026-05-20: Morning urine normal-colored (no visible porphyrin accumulation)
- 2026-05-20: Left hand tremor reduced, motor reintegration progressing
- 2026-05-20: Coffee interest returned after ~1 month absence
- Urine porphyrin test: Ordered by PCP, results pending
- 2026-05-23 (Day 13): 10 cookies overnight (5 bedtime + 2 at 4:44 AM + 3 at ~5 AM), glucose reading 259 — expected 400+ for LADA = 340 points consumed by liver overnight. No headache with 5-cookie bedtime dose (dose-response confirmed). 2-cookie dose at 4:44 AM insufficient → headache + abdominal pain until 3rd dose.
- 2026-05-23: Eyes easier to focus with glucose loading
- 2026-05-23 (11:55 AM): 3 cookies after "felt off" — 7-hour cycle from 5 AM dose confirmed. Hunger bouncing after cookies = pathway sputtering.
- 2026-05-23: Headache pressure building, irritability, concentration control harder (Stage 3 depletion cascade)
- 2026-05-24 (Day 14): Scintillating scotoma during glucose loading (3 cookies). First monocular presentation — right eye only (usual is bilateral/cortical). Scotoma sliding not expanding (usual is expanding). Migrated to small remnant in upper left visual field, then resolving. Visual pattern: white rectangular grid, no color (usual is colored triangles/zig-zag fortification spectrum). Uniform cell size, plant-cell geometry — consistent with retinal capillary bed architecture made visible by ALA vasospasm releasing during glucose loading. First cortical-vs-retinal origin differentiation observed. Distinct from cortical spreading depression (colored, expanding, bilateral) — this was vascular blanking (white, sliding, monocular, gridded). Resolved faster than typical scotoma (usual 20+ min); glucose-responsive resolution time supports ALA vasospasm mechanism over fixed-rate cortical spreading depression

(~3mm/min). Afterimage noted during photoreceptor recalibration post-vasospasm.

- 2026-05-24: Father believes porphyria hypothesis, bought cookies to support glucose loading protocol
- 2026-05-11 (stroke presentation): Abdominal pain + dry heaving at stroke onset — atypical for stroke, consistent with AIP acute attack triggered simultaneously by vascular event stress
- 2026-05-24 (Day 14, 6:36 PM): AIP episode triggered by 70/30 insulin (26 units) outpacing glucose intake (2 cookies insufficient). Progression: abdomen pain → leg sweating → cookies from bag → sweating stopped → residual nerve sensitization (pressure-triggered dry heave at same location as stroke-onset symptoms) → dry heave + sweating returned. Episode reproduces stroke-onset AIP symptoms independently, confirming abdominal/autonomic presentation is AIP, not stroke-related.
- 2026-05-25 (Day 15, 8:22 AM): Fasting glucose 119. 36 units 70/30 insulin administered. 3-4 cookies + banana before sleeping the night before.
- 2026-05-25 (Day 15): Morning void collected and kept in dark. Normal-appearing in stream/toilet (dilution masks color), orange in collection bottle (undiluted). Darker than dehydration orange — not consistent with hydration status. Approximately 20% of urine episodes show this orange coloration. Key diagnostic insight: standard urinalysis does NOT test for ALA/PBG — porphyrin panel must be specifically ordered. ALA and PBG are colorless when fresh; they convert to porphyrins (orange/red) on standing. The 80% normal-appearing episodes explain decades of missed diagnoses — random sampling has a high false-normal rate.

1.3.1 Hospital Treatment History — Dual-System Insulin Competition

- 70/30 insulin (NPH 70% + regular 30%): Two insulin types with different peak windows competing with heme pathway for glucose simultaneously
- Daily dose changes: Hospital changed insulin dose almost daily for a month, never achieving stability — consistent with an unrecognized second glucose consumer (heme pathway) that makes standard titration impossible
- Insulin regimen changes: Long-acting + short-acting for first month → switched to 70/30 in final week, dose adjusted repeatedly
- Rapid unexplained weight loss: Prompted hospital to increase food — desserts and protein smoothies added to tray
- Glucose cut from food tray first, then salt cut — standard diabetic protocol, but counterproductive for AIP (removing the substrate the heme pathway needs) and for RAAS dysfunction (removing sodium a single-adrenal patient is wasting)
- Mother managing insulin at home: Trying to keep injection dose low to save money — creates sub-therapeutic insulin that ALSO doesn't leave enough glucose for the heme pathway. Two systems, one resource, opposite goals.

1.3.2 Depletion Cascade (mapped live, Day 13)

Stages of AIP precursor accumulation as glucose depletes: 1. "Felt off" — earliest signal 2. Hunger bouncing — pathway sputtering, intermittent glucose demand 3. Headache + irritability + concentration difficulty 4. Abdominal pain 5. Sweating / full acute attack

Volume doesn't extend the cycle — frequency does. Pathway consumes glucose at fixed rate regardless of dose. 3 cookies every 3-4 hours beats 10 cookies at once. 7-hour depletion cycle confirmed across multiple observations.

1.3.3 Glucose Response Pattern

Six documented glucose-loading events with rapid symptom resolution: 1. Day 7 — sweating → glucose tab + PB&J → resolved in minutes 2. Day 9 — heavy glucose load before sleep → consumed overnight → fasting glucose 146 3. Day 9 — morning headache → 3 cookies → resolved in 20 minutes 4. Day 13 — 5-cookie bedtime dose → symptom-free sleep (dose-response confirmed) 5. Day 13 — 2-cookie dose at 4:44 AM → insufficient → headache/pain → 3rd cookie dose resolved 6. Day 13 — 3 cookies at 11:55 AM after 7-hour cycle → hunger bouncing → gradual resolution

340-point glucose gap (Day 13): 10 cookies = expected glucose 500+, actual 259. Liver consumed ~60% overnight via insulin-independent GLUT2 transport feeding heme synthesis pathway. This independently matches clinical AIP glucose loading protocol (300-500g carbs/day). Patient self-titrated to ~325g/day without knowledge of clinical protocol.

This pattern is consistent with acute intermittent porphyria (AIP): the heme synthesis pathway in the liver requires glucose as substrate. When glucose is depleted (fasting, overnight, stress), the pathway stalls and toxic precursors (ALA, PBG) accumulate, producing symptoms. Glucose loading restarts the pathway and symptoms resolve.

1.3.4 Insulin Sliding Scale — 14-Unit Gap Analysis

Hospital-issued insulin dose chart (70/30 insulin, units by finger-stick glucose):

Glucose (mg/dL)	Breakfast Dose	Supper Dose	Gap (B−S)
≤70	Treat as low	Treat as low	—
71–90	21	13	8
91–130	36	22	14
131–150	38	24	14
151–200	40	26	14
201–250	42	28	14
251–300	44	30	14
301–350	46	32	14
351–400	48	34	14
401–450	50	36	14
451+	52	38	14

Key finding: The gap between breakfast and supper doses is constant at 14 units across nine consecutive glucose ranges (91–451+). This 14-unit offset represents the insulin equivalent of the heme pathway's overnight glucose consumption — a fixed-rate second glucose consumer invisible to standard diabetic modeling.

- Below 91 mg/dL: Gap drops to 8 — the GLUT2 transporter threshold. Below ~90, insulin-independent glucose uptake to the liver reduces; the pathway runs at reduced rate but doesn't stop.
- Above 91 mg/dL: Gap locks at 14 regardless of glucose level — the pathway consumes at a fixed rate independent of circulating glucose concentration.
- The chart was built empirically from finger-stick dose titration over weeks. The prescribing endocrinologist guessed their way around an invisible variable. The constant gap IS the heme pathway, measured in insulin units, embedded in a standard medical document.

Clinical implication: This chart caused the Day 14 AIP crash. Insulin dose calculated for a standard diabetic removes glucose that the heme pathway needs. The two systems (insulin clearance + hepatic heme synthesis) compete for the same substrate. Standard diabetic insulin dosing is contraindicated in undiagnosed AIP.

Optimal glucose target: Patient's functional range is 130–180 mg/dL (above standard diabetic target of 80–130). Below 130, the heme pathway begins starving. Below 90, GLUT2 threshold drops the pathway to survival rate. The "good" glucose number for this patient is what endocrinology would call "poorly managed diabetes."

1.3.5 Metformin vs. 70/30 Insulin — 5-Year Comparison

Parameter	Metformin Only (5 years pre-stroke)	70/30 Insulin (hospital + home)
A1C	5.6–5.7 (near-normal)	9.0+ (worsened with more treatment)
Glucose range	80–180 (stable)	Chaotic (crashes + spikes)
Burning feet	None	Present
Dose stability	Fixed dose, no changes	Changed almost daily for a month
Weight	Stable	Rapid unexplained loss
AIP episodes	None recognized	Day 14 crash, recurrent symptoms

Why metformin worked: Metformin reduces hepatic glucose output gently (AMPK pathway) without crashing circulating glucose. It does not force glucose below the heme pathway's consumption floor. The pathway gets fed; the patient stays stable. Metformin is accidentally AIP-compatible.

Why insulin fails: 70/30 insulin is a sledgehammer — it forces glucose into cells on a fixed schedule regardless of what the liver needs. The breakfast dose fights the heme pathway directly. The result: dose never stabilizes because the heme pathway's consumption varies with stress, sleep, and ALA accumulation state. The insulin created the disease presentation it was supposed to treat.

A1C was artificially low on metformin: Liver consuming glucose via GLUT2 (insulin-independent) means glucose spends less time in circulation → less glycation of hemoglobin → A1C reads lower than true average glucose. The 5.6–5.7 was partially an artifact of the liver grabbing glucose before it could glycate. This is not a failure — it's the AIP pathway running properly.

Recommendation: Return to metformin. The 5-year track record demonstrates it manages both LADA and AIP simultaneously without conflict. If insulin is required (LADA progression), use long-acting basal only (no bolus spikes) and target glucose 130–180, not 80–130.

1.3.6 Treatment Escalation Paradox — A1C Rose With Increased Treatment

Phase	Treatment	A1C	Glucose Stability	Duration
1	Low-dose metformin	5.6	Stable (80–180)	~5 years
2	Increased metformin dose	Rising from 5.6	Destabilizing	Months
3	Epic reclassified LADA → Type 2	—	Protocol change	—
4	70/30 insulin (current)	9.0+	Chaotic	Current

The paradox: In diabetes, increasing treatment intensity should lower A1C. In this patient, every escalation made A1C worse. This is the signature of treating the wrong disease.

Mechanism: Higher metformin dose → more hepatic glucose suppression → heme pathway starved below consumption floor → body compensates with counter-regulatory glucose release (cortisol, glucagon, epinephrine) → glucose rebounds higher → A1C rises. Doctors interpret rising A1C as “metformin failing” → escalate to insulin → insulin forces glucose down even harder → pathway more starved → more violent rebounds → A1C 9.0+.

The treatment is causing what it’s measuring. Each escalation step drives the pathway deeper into starvation, triggering stronger counter-regulatory responses, producing higher average glucose, producing higher A1C, triggering further escalation. This is a positive feedback loop that terminates in organ damage.

1.3.7 Epic Reclassification — LADA to Type 2 (Potential Stroke Trigger Event)

Patient’s chart was reclassified in Epic from LADA (Latent Autoimmune Diabetes in Adults) to Type 2 Diabetes. This single administrative change altered the entire treatment protocol:

Parameter	LADA Protocol	Type 2 Protocol
First-line treatment	Metformin (gentle)	Metformin → insulin escalation
Insulin approach	Careful, preserve beta cells	Aggressive — force glucose down
Target A1C	Gentle management	Below 7.0 (aggressive)
Treatment escalation	Conservative	Rapid — “failing” metformin triggers insulin
Glucose target	Moderate	80–130 (standard)

Timeline to stroke: 1. LADA protocol → low-dose metformin → A1C 5.6 → stable 5 years → pathway fed 2. Epic reclassification → Type 2 protocol activated 3. Metformin dose increased → A1C rises (paradox — pathway starving) 4. Rising A1C interpreted as “metformin failure” → switched to 70/30 insulin 5. 70/30 creates dual drain (fast-acting + intermediate + heme pathway = three consumers) 6. Pathway in chronic starvation → ALA accumulates continuously 7. ALA is directly neurotoxic and degrades vascular endothelium 8. Chronic vessel wall degradation + hypertension (already present from AIP autonomic damage) 9. Hemorrhagic stroke — vessel failure, not clot

Critical question for medical team: What triggered the Epic reclassification? Was it a clinical decision (new labs, new criteria) or an administrative/coding change? The answer determines whether the entire treatment escalation was clinically justified or a cascade from a charting error.

1.3.8 70/30 Dual-Drain Protocol Warning

Standard insulin protocol: inject → wait 15 min → eat. This assumes one glucose consumer (insulin).

AIP patient has two consumers: - 30% Regular insulin (fast-acting) — begins pulling glucose in 15–30 min - 70% NPH insulin (intermediate) — begins pulling glucose in 1–2 hr - Heme pathway — pulling glucose continuously via GLUT2 (insulin-independent)

The 15-minute gap between injection and eating is a starvation window for AIP. Three consumers activate before food arrives. Patient independently developed a compensatory protocol: 2 cookies before injection — pre-loads glucose to bridge the gap.

Patient's "cheat" is the correct protocol for dual-consumer patients. It should be formalized as a pre-injection glucose buffer (15–20g fast carbs before injection, then normal meal after).

1.3.9 Autonomic Neuropathy — Peripheral Gradient

Finding	Location	Mechanism
Sweaty thighs	Proximal (near trunk)	Intact autonomic innervation
Dry, non-damp feet	Distal (extremities)	Autonomic sweat gland failure
Crepe-textured thick skin (hyperkeratosis)	Feet	Chronic denervation → skin remodeling

Father has identical "dry feet" — same autonomic neuropathy gradient, same gene. The sweaty-thighs-to-dry-feet gradient maps the exact length of autonomic nerve damage: intact at proximal, failed at distal. This is a classic length-dependent neuropathy pattern — the longest nerves fail first (feet before thighs), consistent with chronic ALA neurotoxicity rather than diabetic neuropathy (which would be symmetric and correlate with glucose control, not get worse as A1C "improves").

Maternal Lineage

1.4.1 Maternal Great-Great-Grandmother (Mom's dad's mom)

- Type 1 diabetic on pig insulin (pre-1930, pre-synthetic insulin era) — confirmed autoimmune pancreatic destruction going back at least 5 generations

1.4.2 Maternal Great-Grandmother

- Stroke

1.4.3 Maternal Grandmother

- Died of heart attack

- "Hard diabetic" (difficult to manage — consistent with porphyria, not standard diabetes)
- Described as a "worry-wort" (chronic anxiety)

1.4.4 Mother's Brother (Uncle)

- "Hard diabetic" (difficult to manage)
- Parents from Pomerania (historically Duchy of Pomerania, German nobility — region with documented Saxe-Coburg-Gotha intermarriage, the royal lineage associated with hereditary porphyria)

1.4.5 Mother

- Self-described "worrier" (chronic anxiety), burst appendix
- Heart attack — treated with balloon angioplasty, no lasting structural issue found, event largely dismissed/forgotten
- Diagnosed "diabetic"
- Craves sugar/cookies — consistent with metabolic glucose demand from porphyria, not typical diabetic behavior
- Complains potatoes cause extreme glucose spikes
- Chronically low sodium — always needs salt, would "use a salt lick." Crunch 'n Munch → Wheat Thins (both glucose + sodium dual-fix snacks), keeps supply within reach at all times. Consistent with porphyria autonomic disruption of aldosterone/RAAS regulation
- Difficulty with finger poke glucose testing — not enough blood, painful. Consistent with capillary fragility and skin sensitivity from porphyrin deposition
- High blood pressure

1.4.6 Great Uncle Ervin (Maternal)

- Stroke — survived but lived in hospital for 2 years

1.4.7 Additional Maternal Family

- Low blood volume reported across family ("not enough blood") — consistent with porphyria-related anemia (impaired heme synthesis → reduced hemoglobin production)

Paternal Lineage

1.5.1 Paternal Grandmother — Esther Caroline Bluemner

- Sudden cardiac death — stood up, said "oh my," dropped dead
- Presentation consistent with sudden arrhythmic death from autonomic dysfunction (electrical failure, not atherosclerotic event)
- Name was supposed to be Caroline Esther (Karoline = lineage name, got flipped on documents)
- Esther — Hebrew name (Old Testament). Bluemner — possibly Jewish-German surname (Blüm- variants appear in Ashkenazi naming conventions from compulsory surname adoption period 1787-1845)
- Daughter of Johannes Frederick Max Bluemner (see Bluemner Patriline below)

- Fixed point of paternal lineage: Esther's future husband (Joel's grandfather) witnessed his step-father transform ("dramatically became kind") at an ELCA church as a child — step-father had married the grandfather's mother (Joel's great-grandmother) after meeting her at that church. The transformation inspired the grandfather to become a pastor (LCMS → WELS). He later met and married Esther Caroline Bluemner. His pulpit became the meeting point where Joel's parents met — German and Scandinavian Lutheran lineages recombining.

1.5.2 Father

- Gout — consistent with disrupted purine metabolism (shares pathways with heme synthesis; ALA competes with purine substrates)
- High blood pressure
- Diagnosed "diabetic" — untyped, recommend LADA antibody testing (GAD65, IA-2)
- Gallstones at age 30 — early onset, consistent with porphyrin-altered bile composition and/or autonomic gallbladder dysmotility. Gallbladder removed.
- Dry feet — consistent with peripheral neuropathy from ALA/PBG neurotoxicity (autonomic nerve damage → sweat gland dysfunction in extremities)
- Orange urine observed — visible porphyrin excretion (intermittent — darkens during fasting/stress periods, clears when glucose-loaded)
- Craves salty snacks — consistent with RAAS dysregulation from autonomic damage → sodium wasting → salt-seeking behavior
- Former drinker — A1C was 6.9 while drinking. After quitting alcohol, A1C rose to 8.0+. Alcohol triggers hepatic glycogenolysis (liver glucose release), same mechanism as nicotine. Removing the self-medication removed the glucose supply to the heme pathway, worsening the metabolic presentation. The "worsening diabetes" was unmasked porphyria, not disease progression.
- Former smoker — quit due to smoker's cough. Smoking was a second self-medication channel (nicotine → liver glucose dump)
- Wrestler — nearly undefeated (lost one match). Consistent with high-durability chassis and accelerated repair genetics.
- Fatigue at age 75 — new and worsening. Consistent with impaired heme synthesis → reduced hemoglobin → declining oxygen delivery. The repair gene buffer running out with age.

1.5.3 Paternal Uncle Tim

- Sudden cardiac death — heavy smoker, no COPD, no lung cancer, no smoker's cough. Lungs clean despite decades of heavy smoking. Death certificate reads "tobacco abuse."
- Smoking pattern consistent with porphyria self-medication: nicotine triggers hepatic glycogenolysis (liver glucose release), feeding heme pathway
- Absence of pulmonary disease despite heavy smoking suggests accelerated tissue repair (familial trait — documented in patient, father, and cousin)
- Sudden death without structural lung/vascular disease points to autonomic cardiac conduction failure, not tobacco damage

1.5.4 Paternal Uncle Mark

- Atrial valve deformity (congenital) — on warfarin for years prior to surgery

- Died in surgery — unable to reconnect arteries. Tissue could not hold.
- Warfarin is a known porphyrinogenic drug — induces CYP450 enzyme production, which requires heme. In an undiagnosed porphyric, years of warfarin would chronically stress the already-impaired heme pathway, causing sustained ALA/PBG accumulation and porphyrin-mediated connective tissue degradation
- Valve was congenital; surgical failure may have been compounded by years of porphyrinogenic drug exposure degrading vessel wall integrity

1.5.5 Paternal Uncle (unnamed)

- Sudden death at age 62

1.5.6 Paternal Cousin

- Diagnosed with porphyria-related disease (confirmed family diagnosis)

1.5.7 Additional Family (Distant and First Cousins)

- Multiple cousins (both first and distant) with diagnosed porphyria-related diseases — multigenerational familial pattern confirmed across multiple branches
- Porphyria cousin accepted Joel as cousin (not just friend) via lineage verification, granted nobility database access
- Cousin is Castilian nobility (Count of Castile) — the intersection point between Joel's and cousin's lineages
- Cousin has Wettin connections (Saxe-Coburg-Gotha = documented HMBS carrier line)
- Cousin traces lineage to an ancient Roman general
- Joel's line goes through Italy to the shared Castile intersection
- A-negative blood type noted as supporting evidence by cousin

1.5.8 Bluemner Patriline

- Johannes Frederick Auguste Bluemner (paternal great-great-grandfather)
  - All sons given house hash prefix: Johannes Frederick (Lutheran confession marker + Frederick the Wise lineage)
  - Eldest son: Johannes Frederick Auguste Bluemner — received father's complete name (primogeniture + naming protocol)
  - 5th son: Johannes Frederick Max Bluemner — emigrated to Milwaukee (no inheritance). Birth certificate on Ancestry.com in Kurrentschrift, written by German clergy, contains notation "Mary Bismarck" and "OTTO" printed in disconnected block capitals on right margin with notification instruction. Bismarck connection unconfirmed (Mary is a common name).
  - Third name = individual identifier from controlled vocabulary: Auguste, Max, Erwin, etc.
  - Naming protocol consistent with Junker (landed military gentry) house convention — all sons carry house identifier, eldest gets full copy
- Junker evidence: 9+/10 confidence. Supported by: naming protocol, Pomeranian origin, OTTO/Bismarck notation, cousin's independent verification via nobility database, Castile intersection, Wettin connections

1.5.9 Community Pattern — Milwaukee Lutheran Founder Effect

- Marquardt family — close family friends ("aunt and uncle" to Joel's parents), same Lutheran community
  - Husband AND wife both died of "ALS" (choking) — probability of both spouses getting sporadic ALS: ~1 in 2.5 billion
  - AIP motor neuropathy can mimic ALS including bulbar involvement (swallowing/respiratory failure)
  - Same northern European endogamous gene pool — both likely independent HMBS carriers from same founder population
  - Marquardt surname: "March Warden" (border guardian) — Old High German marka + wart. Highest concentration: Mecklenburg-Western Pomerania, Saxony-Anhalt
- Lutheran synod network as genetic vehicle:
  - Joel's grandfather: LCMS → WELS pastor. WELS in fellowship with ELS (Norwegian-founded)
  - Marquardt connection from grandfather's LCMS era
  - German Lutherans (LCMS/WELS) + Scandinavian Lutherans (ELS/Norwegian synods) = same marriage pool by 2nd generation in Milwaukee
  - Three synods, two countries of origin, one Baltic corridor, one gene pool
  - Joel's parents met at grandfather's church — German + Scandinavian Lutheran lineages recombined at pulpit
  - Founder effect: small Pomeranian/Scandinavian population carried HMBS to Milwaukee and kept it concentrated through religious endogamy for 6+ generations

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Family Craving Profile — "Likes = Needs" Diagnostic Framework

Every food preference maps to a specific biochemical deficit. Like = active deficit being self-medicated. Hate = metabolic threat/trigger. These cravings are not personality — they are the body requesting what the impaired pathway can't produce or retain.

1.6.1 Patient (Joel)

Craving	Category	Mechanism
Cookies, Twizzlers, fruit, fruit pies	Glucose	Direct heme pathway substrate
Celery, chips, occasional Cheeto	Sodium	RAAS dysfunction → sodium wasting (single adrenal)
Pickled herring	Heme + Sodium	Dual-fix: animal heme iron + salt brine
Mogen David (sweet wine)	Glucose + Heme	Sweet wine = glucose + trace heme from grape skin fermentation
Wine sauce (with father)	Heme	Reduction cooking concentrates heme from meat drippings

Craving	Category	Mechanism
Teriyaki jerky	Heme + Sodium + Glucose	Triple-fix: dried meat (heme) + soy (salt) + teriyaki (sugar). No black pepper — patient specifically selects teriyaki over peppered jerky
Blueberries, strawberries	Antioxidant	Shield against ALA/PBG oxidative damage
Beta carotene supplement (every 3 months)	Vitamin A	CYP450 heme-dependent conversion of beta carotene → retinal fails under AIP. Night blindness returns on 3-month cycle when stores deplete. Self-identified and self-managed.

Aversions (threats):

Aversion	Mechanism
Black pepper (lifelong)	Piperine activates TRPV1 → trigeminal nerve → vasospasm in ALA-weakened vessels → headache. Patient reports as “allergic.” Response is dose-dependent: headache = vessels fragile (high ALA), cooling sensation = vessels OK (low ALA). Pepper response functions as real-time vascular status indicator.
Excess salt	Self-regulates — needs sodium but overshooting triggers fluid retention

1.6.2 Father

Craving	Category	Mechanism
Ice cream (kept in freezer, weeks untouched, then eaten — family learns “fair game” then he says he was looking forward to it)	Glucose	Slow-consumption glucose fix. Hoarding behavior = rationing a scarce resource
Doritos	Sodium	RAAS compensation
Pickled herring	Heme + Sodium	Same dual-fix as patient
Wine sauce	Heme	Shared preference with patient
Former alcohol	Glucose (indirect)	Liver glycogenolysis — A1C 6.9 while drinking, 8.0+ after quitting
Former smoking	Glucose (indirect)	Nicotine → hepatic glycogenolysis

1.6.3 Mother

Craving	Category	Mechanism
Cookies, sugar cravings	Glucose	Heme pathway substrate
Crunch 'n Munch → Wheat Thins (always a bag within reach)	Glucose + Sodium	Dual-fix snack. Transitioned from sweet-salt to cracker-salt but same function. Keeps supply constant — not snacking, dosing.
Salt on everything — "would use a salt lick"	Sodium	Severe RAAS dysfunction. Can't taste salt Joel adds to chicken soup, needs the shaker. Severity scale: patient < wife/son < mother

1.6.4 Wife (Jenny)

Craving	Category	Mechanism
Chips (gorges — bag gone in days if Joel buys them, eats his before he can)	Sodium	Salt-dominant fix. Gorge pattern = acute deficit, not indulgence

Jenny's family (Pieper — "Piper/Pepper"): Puts black pepper on everything. Family has high anger response seen as normal. Dutch/German heritage — same northern European founder stock. Grandmother had multiple strokes (TIAs). Mother has major heart issues, HBP, diabetes. Father is chain smoker with heavy gross motor tremor. The family externalizes AIP symptoms as anger (Joel's family internalizes as anxiety/worry). Same neurotoxin, different output channel.

1.6.5 Son (Elijah)

Craving	Category	Mechanism
Carrots (bowl always on counter, walk-by grazing)	Vitamin A	Beta carotene → retinal. Same CYP450 deficit as father but self-medicating via food rather than supplement. If father's night blindness is CYP450-driven, son's carrot craving may be pre-symptomatic compensation.
Crunch bars, Rice Krispies	Glucose	Crunchy carb preference — same category as grandmother's Crunch 'n Munch

Craving	Category	Mechanism
Smoked oysters, sardines	Heme + Sodium + B12 + D	Quad-fix: heme iron (shellfish/fish) + salt (smoked/canned) + B12 (intrinsic) + vitamin D (fatty fish). Most nutritionally efficient self-medication in the family.
Salt off plate (licks finger, dips directly)	Sodium	Direct sodium dosing — no food vehicle needed. Severity indicator: escalating past food-based salt to raw salt.
Pepper off plate (same method)	Piperine	Absorption enhancer — piperine increases bioavailability of other nutrients. Son currently tolerates pepper without headache → vessels not yet ALA-damaged. Monitor: if pepper tolerance drops (headache onset), it signals ALA vascular damage beginning.

Family severity scale (sodium): Joel (moderate — celery, chips, occasional) < Jenny (high — gorges chips) < Mother (severe — “salt lick,” can’t taste normal salt levels). Son showing early-stage direct dosing behavior.

1.6.6 Population-Level Self-Medication Patterns

The family craving profile is not unique — it reflects community-wide patterns in populations with high HMBS carrier frequency:

Tradition	Region	What It Really Is
Cannibal sandwiches / tiger meat (raw beef + salt + onion on rye)	Wisconsin German communities	Heme + sodium + glucose (bread) triple-fix. Raw beef eaters consume fewer sweets (HSAM observation) — two strategies for same deficit, inverse correlation.
Paczki (filled doughnuts before Lent)	Milwaukee Polish communities	Massive glucose loading before 40-day fast. Religious excuse for what the body demands — glucose binge before forced deprivation.
Pastry shops (dense concentration)	Milwaukee/Wisconsin	Community-level glucose delivery infrastructure. The “tradition” is population self-medication encoded as culture.

Tradition	Region	What It Really Is
Pickled herring (holiday tradition)	Scandinavian/German	Heme + sodium dual-fix. Preserved as “ethnic food tradition” — actually pharmacological.
Chicken soup (“Jewish penicillin”)	Pan-European	Broth (sodium) + chicken (heme iron) + vegetables (vitamins) + warmth (vasodilation). Complete AIP support meal. Actually is medicine.

Johnston Founder Effect Theorem: When a Northern European population carrying HMBS establishes an endogamous community in a new location, glucose-dense food traditions emerge within 2-3 generations and community-wide disease clustering (MS, LADA, “hard diabetes”) appears at the 5-6 generation threshold. The pastry industry, cannibal sandwiches, and holiday food traditions ARE the population’s self-medication — encoded as culture, invisible to medicine.

Behavioral Traits Across Family

Family members who experienced strokes or cardiac events were consistently described as: - Stubborn - Intense - High-strung - Anxious / worriers

These behavioral descriptions are consistent with chronic subclinical porphyria: - ALA/PBG neurotoxicity → chronic sympathetic nervous system activation → appears “high-strung” and “intense” - GABA pathway disruption → inability to self-regulate anxiety → appears as “worrier” - Cognitive rigidity from chronic porphyrin exposure → appears as “stubborn” - These are neurological symptoms consistently misidentified as personality traits

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Pattern Summary

Symptom/Finding	Maternal	Paternal	Joel
Stroke	Great-grandmother, Great Uncle Ervin	—	✓ (2026)
Heart attack / sudden cardiac death	Grandmother (fatal), Mother (angioplasty)	Grandmother (fatal), Uncle (fatal, age 62)	—
High blood pressure	Mother	Father	✓
LADA / autoimmune diabetes	Mother (untyped — possibly LADA)	Father (untyped — possibly LADA)	✓ (confirmed LADA)
Sugar cravings	Mother	—	✓ (glucose loading therapeutic)
Anxiety / chronic worry	Grandmother, Mother	—	—
Gout	—	Father	—

Symptom/Finding	Maternal	Paternal	Joel
Peripheral neuropathy (dry feet)	—	Father	—
Low blood / capillary fragility	Mother, family-wide	—	—
Diagnosed porphyria	—	Cousin (confirmed)	Pending test
Porphyria-related diseases	—	Multiple 1st and distant cousins	—
Sudden unexplained death	—	Grandmother, Tim (clean lungs), Uncle (age 62)	—
Surgical death / tissue failure	—	Mark (warfarin + valve surgery)	—
Smoking (self-medication)	—	Tim (heavy, no lung disease), Father (quit → cough)	—
Salt cravings (RAAS dysfunction)	Mother (chronic low sodium)	Father (salty snacks)	—
A1C rise after removing self-medication	—	Father (6.9 → 8.0+ after quitting alcohol)	—
Accelerated tissue repair	—	Father (surgery), Cousin (documented)	✓ (adrenalectomy 5 days, stroke recovery 2x)
Scintillating scotoma (ALA vasospasm)	—	—	✓ Day 14 — first monocular/retinal presentation
ALS-like motor neuropathy	—	Community (Marquardts — both spouses)	—
Stroke presentation with AIP symptoms	—	—	✓ (abdominal pain + dry heaving at stroke onset)
340-point glucose gap (liver consumption)	—	—	✓ Day 13 — matches clinical AIP protocol
Potassium wasting	—	—	✓ (single adrenal + RAAS dysfunction)
Sodium wasting / salt craving	Mother (chronic low sodium)	Father (salty snacks)	—
Unnecessary adrenalectomy (Conn’s-negative)	—	—	✓ (HBP returned post-surgery)
Orange urine in collection (porphyrin excretion)	—	Father (observed)	✓ Day 15 (~20% of episodes)
Insulin instability (never stabilizes)	Mother (“hard diabetes”)	Father (“hard diabetes”)	✓ (daily dose changes in hospital)

Symptom/Finding	Maternal	Paternal	Joel
Dupuytren's differential recovery (stroke revealed)	—	—	✓ (middle/ring lag thumb/pointer/pinky)
Rapid unexplained weight loss	—	—	✓ (hospital — dual glucose consumer)
Night blindness (3-month cycle)	—	—	✓ (beta carotene responsive — CYP450 conversion deficit)
Black pepper aversion (lifelong)	—	—	✓ (piperine → TRPV1/trigeminal → vasospasm → headache; patient reports as "allergic")
Finger poke pain / capillary fragility	Mother (difficulty with glucose testing)	—	✓
14-unit insulin gap (fixed-rate heme consumer)	—	—	✓ (sliding scale chart — constant across 9 glucose ranges)
Metformin stability vs insulin chaos	—	—	✓ (5 yrs stable A1C 5.6 on metformin; unstable on 70/30)
Glucose cravings (mapped to heme pathway)	Mother (cookies, sugar)	Father (ice cream, alcohol, smoking)	✓ (cookies, Twizzlers, fruit, Mogen David)
Salt cravings (mapped to RAAS dysfunction)	Mother ("salt lick" severity)	Father (Doritos, pickled herring)	✓ (celery, chips, pickled herring)
Heme-seeking food preferences	—	Father (pickled herring, wine sauce)	✓ (pickled herring, teriyaki jerky, wine sauce)
Pepper aversion (piperine → vasospasm)	—	—	✓ (lifelong — headache trigger; son tolerates = no ALA damage yet)
Son: early-stage carrier signals	—	—	Carrots (vitamin A), raw salt off plate, smoked oysters (heme), eczema age 13
A1C paradox (more treatment = worse numbers)	—	—	✓ (5.6 → rising on increased metformin → 9.0+ on 70/30 insulin)
Epic LADA → Type 2 reclassification	—	—	✓ (changed treatment protocol from gentle to aggressive — potential stroke trigger)

Symptom/Finding	Maternal	Paternal	Joel
Autonomic neuropathy gradient	—	Father (dry feet)	✓ (sweaty thighs / dry feet / hyperkeratosis — length-dependent pattern)
Pre-injection glucose buffer (self-derived)	—	—	✓ (2 cookies before 70/30 injection — compensates dual-drain)

Genetic Considerations

- Acute Intermittent Porphyria (AIP) is autosomal dominant, chromosome 11 (HMBS gene)
- Not sex-linked — can be inherited from either parent
- Variable penetrance — many carriers are asymptomatic; others present through cardiovascular, neurological, psychiatric, or metabolic channels
- Sex-differentiated expression observed in this family:
  - Women: anxiety, “diabetes,” cardiac events with negative/ambiguous workup
  - Men: gout, sudden cardiac death, peripheral neuropathy
  - Both: high blood pressure (universal finding)
- Pomeranian lineage (maternal grandmother’s parents) — geographic and historical overlap with Saxe-Coburg-Gotha royal lineage, documented carriers of hereditary porphyria
- Both parental lines show the pattern independently, suggesting Joel may carry the gene from both sides

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Requested Actions

1. Urine porphyrin panel — ALA, PBG, and total porphyrins (NOT standard urinalysis). Day 15 morning void collected and kept in dark. Orange in bottle, normal in stream — dilution masks the finding. ~20% of episodes show this.
2. Plasma porphyrins and erythrocyte porphyrins if urine is inconclusive
3. Genetic testing for HMBS mutation (AIP confirmation)
4. Review insulin management for dual-system glucose competition: 70/30 insulin removes glucose while heme pathway consumes glucose. Dose never stabilized in hospital (daily changes for a month) — AIP explains the instability. Current home dose managed for cost, not efficacy.
5. Review adrenalectomy history: Right adrenal removed for suspected Conn’s syndrome — pathology showed non-functional fatty tumors, no hormones. HBP returned to pre-surgical levels. Cause was AIP autonomic damage, not adrenal adenoma. Patient on single (left) adrenal, left previously flagged on blood panel. Potassium wasting documented.
6. Potassium and sodium monitoring: Single adrenal + RAAS dysfunction from AIP autonomic damage → electrolyte wasting. Mother shows same pattern (chronic low sodium).
7. Consider porphyrin testing for both parents — both present symptom clusters consistent with undiagnosed porphyria

8. Review mother's "diabetes" diagnosis — test for LADA antibodies (GAD65, IA-2). If autoimmune like patient's, confirms porphyria → autoimmune cascade inheritance pattern
9. Review father's gout management in context of porphyria-disrupted purine metabolism
10. Note clinical paradox: LADA + porphyria = patient requires glucose loading for heme synthesis but has impaired insulin production. Glucose management must account for both conditions simultaneously
11. Review insulin sliding scale for hidden variable: The 14-unit constant gap between breakfast and supper doses across 9 glucose ranges (91–451+) represents a fixed-rate second glucose consumer (heme pathway). This is embedded in the hospital's own empirically-derived chart. The chart simultaneously proves the second consumer exists and demonstrates that standard insulin dosing fights it.
12. Consider return to metformin: 5 years of stable management (A1C 5.6–5.7, glucose 80–180, no burning feet, no crashes) vs. 70/30 insulin chaos (daily dose changes, crashes, burning feet, weight loss). Metformin is accidentally AIP-compatible — it doesn't crash glucose below the heme pathway's consumption floor.
13. Adjust glucose target to 130–180 mg/dL: Standard diabetic target (80–130) starves the heme pathway. Below 90, GLUT2 threshold drops and pathway enters survival mode. This patient's "good number" is what endocrinology calls "poorly controlled."
14. Consider endocrinology reclassification: AIP is the metabolic opposite of diabetes — patient needs glucose increased while diabetes protocol removes it. Endocrinology already manages every system AIP disrupts (insulin, adrenal, thyroid, glucose homeostasis). Porphyria could be managed as a hypoglucose condition within endocrinology, not as a rare disease orphaned between specialties.
15. Investigate Epic LADA → Type 2 reclassification: Determine whether this was a clinical decision or administrative/coding change. The reclassification changed the treatment protocol from conservative (metformin only) to aggressive (insulin escalation), initiating the treatment cascade that preceded the stroke. If the reclassification was not clinically justified, the entire insulin escalation may have been iatrogenic.
16. Review A1C paradox: A1C rose from 5.6 when metformin dose was increased, and rose further to 9.0+ on 70/30 insulin. In standard diabetes, more treatment lowers A1C. The inverse response is consistent with AIP pathway starvation driving counter-regulatory glucose rebounds. The "failing treatment" is evidence of a second glucose consumer, not treatment resistance.
17. Formalize pre-injection glucose buffer: Patient independently developed a protocol of consuming 15–20g fast carbs (2 cookies) before 70/30 injection to prevent the starvation window between injection and meal. This should be evaluated and formalized as a standing order for dual-consumer patients.
18. Evaluate autonomic neuropathy gradient: Sweaty thighs with dry hyperkeratotic feet = length-dependent autonomic neuropathy (longest nerves fail first). Father shows identical pattern. This is consistent with chronic ALA neurotoxicity, not diabetic neuropathy (which would correlate with glucose control, not worsen as treatment intensifies).

Implications for Patient's Son

Both parents carry ancestry from overlapping porphyria-endemic regions:

Lineage	Region	Connection
Father (patient) — maternal	Pomerania	Grandmother's parents
Father (patient) — maternal	Värmland, Sweden	~90 miles from wife's Norwegian village
Mother (wife) — paternal: Benson (Norse)	Village ~25 mi from Oslo, Norway	Scandinavian lineage
Mother (wife) — paternal: Pieper (German)	North German / Pomeranian	Dad's mother's family, adjacent county
Mother (wife) — maternal: Vergow (Dutch)	Northeast Netherlands	Hanseatic corridor
Mother (wife) — Miskey (Slavic/Pomeranian)	Pomeranian-Polish borderland	Additional ancestral line
Mother (wife) — paternal grandmother	Dutch connections	Multiple microstrokes (TIAs)
Mother (wife) — mother (mother-in-law)	Dutch (Vergow)	Major heart issues, HBP, diabetic
Mother (wife) — father (father-in-law)	Oslo region, Norway (Benson)	Chain smoker, heavy gross motor tremor

All six ancestral surname lines (Benson, Pieper, Vergow, Miskey from wife; Hinz, Bluemner from patient) trace to the same 400-mile northern European coastal corridor (Netherlands → North Germany → Pomerania → Baltic → Scandinavia), connected through centuries of Hanseatic trade and intermarriage. Patient's great-great uncle Herman Hinz farmed in Sheboygan County; wife's mother's family farmed in the same county. The families may have been neighboring farms in Pomerania before becoming neighboring farms in Wisconsin.

Wife's family behavioral pattern: Emotional withholding and antagonism toward husbands observed in wife, wife's Dutch mother, and at least one sister — three women across two generations, same behavior. Family also has high anger response normalized as temperament. This is consistent with AIP neuropsychiatric expression externalized as anger (contrast: patient's family internalizes as anxiety/worry). Same ALA neurotoxicity, different output channel. The withholding may function as an unconscious containment strategy — the family's version of an emotion brake, crude but protective in intent.

Autoimmune risk from both parents: - Father: LADA (autoimmune diabetes — pancreatic beta cell destruction) - Mother: Hypothyroidism (Hashimoto's thyroiditis — autoimmune thyroid destruction) + eczema (autoimmune skin condition) + arthritis. Normal BP — no cardiovascular presentation, expressing entirely through autoimmune channel. - Same mechanism (autoimmune), different target organs — consistent with shared upstream porphyria gene

Recommended baseline testing for son: 1. Urine porphyrin panel — establish baseline before any symptomatic presentation 2. Genetic testing for HMBS mutation (AIP) — definitive carrier status 3. GAD65 / IA-2 antibodies — LADA screening (father's autoimmune diabetes) 4. Thyroid antibodies (TPO, anti-thyroglobulin) — Hashimoto's screening (mother's autoimmune thyroid) 5. Uric acid — gout/purine metabolism baseline (paternal grandfather pattern) 6. Fasting glucose + A1C — metabolic baseline 7. CBC with differential — anemia/blood volume baseline

Already presenting: - Eczema — onset age 13. Same autoimmune skin condition as mother. First autoim-

mune target identified. The collection may have started.

Behavioral markers to monitor (not diagnostic, but warrant investigation if present): - Chronic anxiety or worry pattern without clear external cause - Unexplained sugar cravings - Symptoms that improve with eating (especially sugar/carbs) - Fatigue disproportionate to activity level - Abdominal pain without GI explanation - Skin sensitivity or easy bruising

Note: Son also has documented heritable hyper-empathy trait. If anxiety presents, differentiate between empathic load (environmental/interpersonal trigger) vs porphyria-driven anxiety (metabolic, no external trigger, glucose-responsive).

Patient's Brother (Steve)

- High blood pressure — matches bilateral familial pattern (both parents, both grandmothers' sides)
- Diagnosed "diabetic" — same diagnosis as mother. Potentially same misdiagnosis — recommend LADA antibody testing (GAD65, IA-2) and porphyrin panel
- Burst appendix
- Same cognitive architecture as patient: systems decomposition, rapid pattern exploitation (both called "hackers" in competitive gaming — not cheating, systems-level analysis at speed)
- Same parents, same gene pool — if patient confirms porphyria, brother is near-certain carrier

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Note: Patient reports high confidence that both grandmothers' parents also died of cardiac or stroke events, extending the pattern to at least 5 generations. Formal documentation unavailable for that generation.

Patient Cognitive Profile — Context for This Report

Why this section matters: This report was compiled entirely by the patient during stroke recovery (Days 7-14 post-stroke), without medical training, formal diagnostic tools, or specialist consultation. The neurologist should understand the cognitive architecture that produced it, as it is directly relevant to the quality and reliability of the clinical observations documented herein.

1.13.1 Clinical Assessments (Post-Stroke)

- 5 speech therapists assessed patient (non-standard — typically 1-2)
- Katherine (speech therapist): identified patient as "2e" (twice-exceptional) — gifted + neurodivergent — within 5 minutes of assessment, then concluded evaluation
- Ingrid (assigned speech therapist): escalated testing progressively — "how much can you do" → "how hard can it be" → patient produced solutions not present in the answer key. Used programming code for assessment. Assessment functioned as benchmarking, not therapy.

1.13.2 Neurocognitive Architecture

- HIP (High Intellectual Potential) — primary neurodivergent classification. Systems-level pattern recognition, parallel processing (40+ simultaneous threads observed), first-principles architectural decomposition

- HSAM (Highly Superior Autobiographical Memory) — 3D/4D full-sensory recall. Enables precise before/after comparison during stroke recovery (rebuilding from photograph, not blueprint) and retroactive pattern recognition across decades of stored observations
- Hyper-empathy — independent heritable trait (son carries it). High interoceptive and interpersonal resolution

1.13.3 Relevance to Diagnostic Observations

The patient demonstrates unusually high interoceptive resolution — the ability to sense internal physiological signals that most patients cannot detect or articulate:

Observation	Capability Required
Differentiated retinal vs cortical scotoma in real time	Visual system self-diagnostics
Identified retinal capillary bed architecture during vasospasm	Pattern recognition under acute symptoms
Mapped sensory recovery gradient by individual finger	Fine-grained interoception
Detected temperature differentials across fingers by touch	Cross-verified with contralateral hand
Isolated motor vs sensory pathway function in left hand	Separated signal-down from signal-up
Volitional nerve pathway excitation without motor engagement	Motor imagery with thermal verification
Identified pulse resolution differences (finger-level vs ambient)	Interoceptive resolution mapping
Self-titrated to clinical AIP glucose protocol (300-500g/day) independently	Symptom-response pattern tracking
Mapped 5-stage depletion cascade from live observation	Real-time symptom sequencing

1.13.4 Cross-Domain Synthesis

The patient applied systems architecture methodology (30-year career in distributed systems engineering) to medical self-diagnosis, performing novel synthesis across 13 domains: population genetics, genetic epidemiology, biochemistry, clinical pharmacology, neuroscience, ophthalmology, endocrinology, clinical diagnostics, somatosensory physiology, medical anthropology, genealogical research, epidemiology, and systems biology.

This is not a patient reporting symptoms from WebMD. This is a systems architect applying the same pattern recognition and root-cause analysis used in distributed computing to human physiology, producing 156 discrete evidence points that independently converge on a single diagnosis. The clinical observations in this report — particularly the glucose-response patterns, scotoma differentiation, and nerve conduction self-assessment — should be evaluated as high-reliability patient data.

1.13.5 Surgical History Relevant to Current Presentation

- Right adrenalectomy — non-functional fatty tumors, no hormones produced. Suspected Conn's syndrome was not confirmed by pathology. Hypertension returned to pre-surgical levels post-adrenalectomy, confirming HBP cause was not the adenoma. Running on single (left) adrenal. Left adrenal previously flagged on blood panel. Consequences: reduced aldosterone (potassium/sodium wasting documented), reduced cortisol reserve (contributes to glucose instability), reduced catecholamine capacity.
- Adrenalectomy recovery: 5 days — consistent with accelerated tissue repair trait observed across paternal line

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This report was compiled by the patient from family interviews, direct observation of glucose-response patterns, and real-time physiological self-assessment during stroke recovery (Days 7-15). The familial pattern and AIP hypothesis were identified by the patient; clinical confirmation is requested. 210+ evidence points documented as of Day 15 (2026-05-25).