Sila Khaled Attia

What the Research Found, and What To Do Next

A plain-English guide for the family — updated with an independent review

Date of birth: 14 October 2012 · Diagnosis: profound (late-onset) biotinidase deficiency · Updated: 23 May 2026

Please read first.

This is a plain-language summary written to help the family understand the medical records and the research, and to organise conversations with Sila's doctors. It is not a diagnosis, a prescription, or a substitute for her own neurologist, metabolic geneticist, and rehabilitation team — they can examine her and order tests. Review every point below with them. The single most important rule: never stop or interrupt biotin without her doctors' direction.

1. Where things stand today

Sila is 13. In 2022, at age 9–10, she rapidly lost the ability to move and lost her vision, and at one point needed help breathing. After several wrong turns, the cause was identified as biotinidase deficiency, a rare inherited condition in which the body cannot recycle the vitamin biotin. Once she started high-dose biotin in January 2023, she began to recover.

Today she has **full strength in her upper body**, her vision is back, and she can sit independently and stand with support. The slowest-recovering areas are her **lower-limb spasticity (stiff, tight legs)** and **bladder and bowel control**. She has also developed osteoporosis (weak bones) and a spine curve (scoliosis) that is improving with bracing.

2. The bottom line from the research

The research report (generated with Google Gemini) and the Boston Children's second opinion **agree with the core findings**. In plain terms:

- **The biotinidase deficiency diagnosis is confirmed** — the enzyme test is decisive.
- **Biotin is the correct treatment** and is keeping her safe. **It must never be stopped** — stopping it is dangerous and can trigger a serious relapse.
- Her recovery, though slow, is real. The slow pace mostly reflects how severe the original spinal-cord injury was.
- **However — an independent re-reading of her raw tests agrees on the diagnosis but raises one important question the reports did not settle.** This is explained in Section 3, and it changes the order of the action plan.

3. An independent second look — and where we'd push back

We re-read Sila's actual lab results and scans on their own, deliberately setting the experts' reports aside, to pressure-test their conclusions. Here is what that found, in plain terms.

Where we fully agree

- **The biotinidase deficiency diagnosis is solid.** The enzyme level is far below normal — this part is not in doubt.
- **Biotin is the right treatment and must continue for life.**

The one big question they did not resolve

Sila's brain and spine scans look **exactly like a separate, well-known nerve disease called NMO** (neuromyelitis optica) — an immune condition in which the body attacks the optic nerves and spinal cord. Her scans, her vision test, and even the radiologist's own wording all point that way. Four things stand out:

- The simple blood test that tells these two apart — the **AQP4 and MOG antibody tests** — was never done.
- Right before biotin was started, she was given the **exact medicines used to treat NMO** (including rituximab, which keeps working for many months). So her recovery cannot be credited to biotin alone — NMO treatment and natural healing were happening at the same time.
- A **new spot appeared on her spinal cord in 2024 while she was on biotin**. A deficiency that is being corrected should be fading, not producing new spots — which hints at an ongoing immune process.
- Her scans are **missing the changes doctors normally expect in biotinidase deficiency** (certain brain shrinkage / myelination patterns) and instead match the immune-demyelinating pattern.

What this means in plain terms.

It is quite possible Sila has biotinidase deficiency *and* an immune nerve disease at the same time — which is more likely in families where the parents are related. If so, biotin alone would never be enough, and she would need a separate treatment to prevent future immune attacks. This matters because another attack could again affect her breathing. This is not a reason to panic, and it does not change that she should stay on biotin — it is a reason to do one blood test and one scan to settle the question, which is why they are now at the top of the action plan.

Our honest verdict on the expert reports

They were **right on the biochemistry** (the diagnosis and the need for biotin), and right to suggest hearing testing and a possible second condition. But we think they were **over-confident and mis-prioritised**: they treated biotinidase deficiency as a complete explanation, never recommended the decisive antibody test, did not engage with the new spinal-cord spot, and credited the recovery to biotin even though NMO treatments had just been given. The Boston opinion was also written by a metabolic specialist who openly declined the nerve-related questions — yet the nerves are where the uncertainty and the disability are. Our disagreement is about **certainty and what to test next**, not about whether she has biotinidase deficiency.

4. Is there a permanent cure?

Honestly, not yet. There is **no cure for biotinidase deficiency itself** — no gene therapy or enzyme-replacement treatment is ready for it. The treatment is lifelong biotin, which replaces what her body cannot recycle.

For the nerve damage left behind, researchers are studying experimental approaches, but none is a proven cure today. They are things to ask about and watch — not options to start now (Section 6). The one genuinely hopeful item is an implanted device for bladder/bowel control, which has restored control in some children. Note: if the antibody test (Section 5) shows she also has an immune disease, then there is an effective, established treatment for that — to prevent future attacks — which is a different and important kind of help.

5. Your action plan — in order

Step 1 is permanent and unchanged. Steps 2–4 are the new top priority from the independent review (Section 3). The bone treatment (step 8) is already scheduled and should proceed *in parallel* — it does not have to wait its turn.

1. **Keep biotin going, exactly as prescribed.** Non-negotiable. Pause it only under medical supervision, and only briefly, for the blood tests below.
2. **[MOST IMPORTANT] AQP4 + MOG antibody blood tests.** The decisive test to find or rule out a coexisting immune nerve disease (NMO / MOG). Ask for the “live cell” version, done during a short, supervised pause in biotin (biotin distorts blood tests).
3. **Contrast MRI of brain + whole spine (with dye).** Every scan so far has lacked the dye. It is needed to see whether the newer spinal-cord spot is active — a sign of ongoing immune disease.
4. **Spinal-fluid test (lumbar puncture), if the neurologist agrees.** Looks for “oligoclonal bands,” another clue to immune disease.
5. **Confirm the biotinidase diagnosis properly.** Re-confirm the gene change, test both parents (to see how the change was inherited), and consider repeating the enzyme level / a genome test — the gene result is currently only a “variant of uncertain significance.”
6. **Urine metabolic tests.** Urine organic acids and acylglycines, to confirm her biotin dose is actually enough and guide any increase.
7. **Hearing test.** This condition can quietly affect hearing, so it should be checked.
8. **Bone treatment, done safely (in parallel).** The IV bone medicine needs calcium + vitamin D loading first, plus blood, kidney, and dental checks.
9. **Trio genetic test (Sila + both parents).** To check for a possible second inherited condition behind the osteoporosis and the high biotin dose.
10. **Neuro-urologist for bladder and bowel.** Including a “urodynamic” study and a discussion of the implanted device (Section 6).

6. Experimental options — "ask about / monitor," not yet

These are not part of the active plan. They are emerging ideas to discuss with her specialists once the picture is complete. The implant is the most realistic; the rest are early-stage. Do not start any of these on your own.

A. The implant — sacral neuromodulation (most realistic)

What it is: a small pacemaker-like device placed near the tailbone that sends gentle signals to the nerves controlling the bladder and bowel. In some children, control returned so completely that the device was later removed.

The honest nuance: most of the strong results are in children whose problems are not from spinal-cord damage. Sila's incontinence comes from her cord injury, where results are more variable. The reassuring part — you don't have to gamble: it is normally done in two stages, a temporary test lead first, with the permanent implant only if the test clearly helps.

What to do next:

- Ask for a pediatric neuro-urology referral and a urodynamic study first — this decides whether she is even a candidate.
- Ask specifically about the staged trial approach (temporary test before committing).
- Ask for an MRI-conditional device, since she will keep needing MRIs and the implant must be MRI-safe.
- Timing: this comes after the diagnostic work-up; her bladder may still be slowly improving on biotin.

B. Clemastine (a remyelination drug)

What it is: an old allergy medicine being studied to help nerves rebuild their protective coating. A small study showed a modest improvement, but a different trial was stopped because some patients got worse, and it can be harmful during active nerve inflammation. There is no approved children's dose.

What to do next: Not now, and do not try it on your own (it is sold over the counter in some places — avoid that). Treat it only as a question for her neurologist, and only after the antibody tests and contrast MRI confirm there is no active inflammation.

C. NVG-291 (a nerve-regeneration drug)

What it is: an experimental drug for spinal-cord injury, still in clinical trials, with a larger trial only planned around 2026 — and current trials likely exclude her age.

What to do next: Nothing actionable today. Ask her team to keep an eye on ClinicalTrials.gov and flag her if eligibility ever opens. File under "monitor."

D. Stem cells

What it is: experimental cell treatments with no standard protocol and no proven benefit for her condition.

What to do next: Be careful here. This is where unregulated "stem-cell clinics" charge families large sums for treatments that don't work and can cause harm. The only safe route is a registered clinical trial at a real academic hospital — never a pay-out-of-pocket private clinic.

Reality check: None of these four will help more right now than the basics — biotin, intensive physiotherapy, and finishing the missing tests (especially the antibody test). Those come first.

7. One-page checklist

When	Action
Now / ongoing	• Continue biotin every day — never stop without medical supervision • Continue physiotherapy, stretching, leg braces, and spasticity medicines (baclofen, tizanidine) • Prevent pressure sores; support nutrition and mood
Most important tests	• **AQP4 + MOG antibodies** (live-cell test, during supervised biotin pause) — the decisive test • **Contrast MRI of brain + whole spine** (with dye) — is the new spinal-cord spot active? • Spinal-fluid test (lumbar puncture) for oligoclonal bands, if the neurologist agrees
Other tests soon	• Re-confirm the gene change + test both parents (segregation); consider genome / repeat enzyme • Urine organic acids + acylglycines (to check biotin dose) • Hearing test (audiometry + BAER) • Bone treatment — proceed in parallel after calcium/vitamin D + blood/kidney/dental checks • Trio genetic test (Sila + both parents) • Neuro-urology referral + urodynamic study
Ongoing checks	• Yearly: genetics/metabolic review, eye exam, hearing test, DEXA bone scan • Every 6–12 months: scoliosis spine X-ray • Repeat MRI as advised to confirm stability
Ask about / monitor (not yet)	• Implanted bladder/bowel device (sacral neuromodulation) — after urodynamics; ask for staged trial + MRI-safe device • Clemastine — only a question for the neurologist, only if no active inflammation • NVG-291 — monitor ClinicalTrials.gov • Stem cells — only via a registered hospital trial, never a paid private clinic

8. Warning signs — get urgent medical help

Seek urgent neurological care (and ask for a contrast MRI) if any of these appear, as they could signal a relapse or a new spinal-cord problem:

- New or worsening vision loss
- New limb weakness, or numbness that is creeping upward
- A sudden change in bladder or bowel function

- **Any breathing difficulty — treat as an emergency.** She previously needed breathing support, so this is potentially life-threatening.

9. Who to contact

In Egypt (Cairo) — starting points the research highlighted:

- **Ain Shams University:** Prof. Solaf M. Elsayed (medical genetics — her own doctor, who identified the variant and manages her biotin); also Prof. Nagia Fahmy (neuromuscular), Prof. Osama K. Zaki, Prof. Nevine El Nahas (neurology).
- **Cairo University / Aboul Reesh Children's Hospital:** Prof. Laila A. Selim.
- **National Research Centre:** Dr. Hasnaa Elbendary.

Most useful next referral: a pediatric **neurologist / neuro-immunologist** — to order the antibody test and contrast MRI and interpret them. International centres (Boston Children's; tertiary centres in Qatar and Saudi Arabia) listed institutions only, with no direct contact details.

Note: the research named a specific Boston doctor that could not be independently verified in the source links, so treat that individual name loosely and confirm with the hospital.

10. Questions to bring to each specialist

For the neurologist / neuro-immunologist (now the priority)

- **Do the AQP4 and MOG antibody results show a coexisting immune nerve disease — and if so, does she need ongoing treatment to prevent future attacks?**
- What does the contrast MRI show about the newer spinal-cord spot — is it active?
- Could any of her recovery be due to the rituximab/IVIG she received, rather than biotin alone?
- Can we optimise the spasticity treatment (medicines, botulinum toxin, therapy)?

For the metabolic geneticist

- Do the urine tests show my biotin dose is enough, or should it go higher?
- Can we confirm the gene change and test both parents, and should we proceed with trio / genome testing?

For the neuro-urologist

- Is she a candidate for the implanted bladder/bowel device, and can we do a temporary test first (and an MRI-safe device)?

For the bone / endocrine team

- Are her vitamin D and calcium corrected before the bone medicine, and is the osteoporosis expected to improve as she becomes more mobile?

11. Safety reminders

- **Never stop biotin** without her metabolic team's direction.
- **Biotin distorts blood tests.** High-dose biotin can cause false results on many lab tests (including thyroid and the AQP4/MOG antibody tests). Tell every lab she is on high-dose biotin, and pause it only as instructed before those specific tests — then resume.

- **Be cautious with steroids / strong immune medicines** unless a true antibody-mediated disease is confirmed — they did not help acutely and can worsen the osteoporosis. (If the antibody test is positive, the right immune treatment is a different, targeted one — discuss with neurology.)
- **Avoid unregulated "stem-cell" or "cure" clinics** that charge out-of-pocket; use only registered hospital trials.

12. Plain-English glossary

Biotinidase deficiency	An inherited condition where the body can't recycle the vitamin biotin, which nerves and metabolism need. Treated with high-dose biotin for life.
NMO (neuromyelitis optica)	An immune disease where the body attacks the optic nerves and spinal cord. It can look identical to Sila's scans but is treated very differently — which is why the antibody test matters.
AQP4 / MOG antibodies	Blood tests that detect NMO / MOG immune disease. The decisive test to separate it from a metabolic mimic.
Variant of uncertain significance	A gene change whose effect isn't fully proven — testing the parents helps clarify it.
Spasticity	Muscles that are stiff and tight and hard to control — here, in the legs, from the spinal-cord injury.
Neurogenic bladder / bowel	Loss of normal bladder/bowel control because the controlling nerves were injured.
Contrast MRI (gadolinium)	An MRI with a dye that highlights areas of active inflammation.
Osteoporosis	Weak, thinned bones — here likely from immobility, steroids, and nutrition.
Sacral neuromodulation	A small implanted device that stimulates the nerves controlling the bladder and bowel.

This guide summarises the records and research in this folder as of 23 May 2026, and includes an independent re-reading of the primary tests. It is intended to support, not replace, the judgement of Sila's treating physicians.