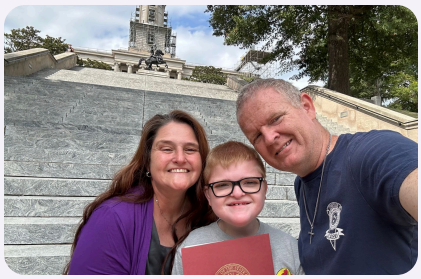
IDEFINE The Kleefstra Syndrome Foundation

NEWLY DIAGNOSED · UPDATED AUG 28, 2026

Welcome to your family by fate.



If your child or someone you love has just been diagnosed with Kleefstra syndrome, please know this: **you are not alone**. You have found a community that understands how overwhelming this moment feels, a foundation started by parents who have been exactly here, and a clear set of next steps. Take a deep breath. We will walk this together.

What it is

Kleefstra syndrome is a rare genetic condition caused by a missing or changed copy of the *EHMT1* gene. It commonly involves intellectual disability, low muscle tone, delayed speech, and distinctive features. It ranges from mild to severe, and every child is their own person.

There is a roadmap

A 2026 international clinical guideline spells out exactly what to check and when, a dedicated Kleefstra clinic at Boston Children's Hospital sees families from everywhere, and KS has its own diagnosis code, **Q87.86**. You do not have to invent the plan.

There is hope

IDefine funds gene therapy, RNA, and drug-repurposing research aimed at treating Kleefstra syndrome itself. More than 1,100 people with KS are on the Kleefstra world map. Research moves because families like yours join in.

FOR YOUR CHILD'S CARE

First steps this month

1. **Get the full genetic report and ask for genetic counseling.** Note deletion vs. variant, and whether it was de novo.
2. **Give your pediatrician the clinical guideline and the ICD-10 code Q87.86.** Most doctors have never seen KS; the guideline tells them what to check and the code gets the diagnosis on the record.
   idefine.org/guideline
3. **Book the baseline evaluations:** heart (echo + ECG), hearing, vision, speech-language, plus sleep, growth, and behavior reviews. Neurology if seizures or lost skills are a concern.
4. **Request early intervention (under 3) or a school evaluation (3+).** Waitlists are long. Get in line now, in writing.
5. **Sign up for Citizen Health and meet Ari. It's free.** Through IDefine's partnership, Ari gathers your child's records from every provider in minutes, explains them, and preps you for appointments. Then ask about the Kleefstra Syndrome Clinic.
   citizen.health/ai-advocate/kleefstra-syndrome · idefine.org/family-resource-packet

FOR YOU, WITH IDEFINE

First steps into the community

1. **Tell us you're here.** A parent will reach out one-on-one.
   unlock@idefine.org
2. **Add your pin to the Kleefstra World Map** and join the private caregiver group on Facebook. Read for a while; post when you're ready.
   kleefstraworldmap.org · facebook.com/groups/Kleefstra.Syndrome
3. **Be counted.** Say yes to research sharing in Citizen Health, and register with Simons Searchlight and RARE-X. Your child's data is the foundation every future treatment is built on.
   idefine.org/research
4. **Stay in the loop.** Sign up for the newsletter and follow IDefine on Facebook, Instagram, LinkedIn, and YouTube. Plan to join us at the annual Family Conference.
5. **When you're ready: volunteer or fundraise.** A birthday campaign, a company match, a few hours of your skills. Families fund the research.
   idefine.org/fundraise-for-research · unlock@idefine.org

Keep going: Your First Year with Kleefstra Syndrome

Our companion guide walks month by month through the first 6–12 months: the evaluation checklist, therapies and IEPs, insurance and waivers, research, ways to get involved, and a page just for you. Download it at **idefine.org/newly-diagnosed**.

Talk to a parent unlock@idefine.org
Kleefstra Clinic 617-355-3645
Everything else idefine.org
ICD-10 code Q87.86 · **Awareness Day** Sept 17