IDEFINE The Kleefstra Syndrome Foundation

LAST UPDATED AUGUST 28, 2026

A GUIDE FOR NEWLY DIAGNOSED FAMILIES

Your First Year with Kleefstra Syndrome

What to do first, what can wait, who to call, and how to find your people. Written by parents who have stood exactly where you are standing now.

FAMILIES AT THE KLEEFSTRA SYNDROME FAMILY CONFERENCE

WELCOME

You are not alone. And there is hope.

If you are reading this, your child or someone you love has recently been diagnosed with Kleefstra syndrome. You have probably heard a word you had never heard before, searched for it late at night, and found far less than you were hoping for. Take a breath. You have found the right place.

IDefine was founded in 2020 by five parents who each got this same diagnosis for their own child and decided that waiting was not an option. Everything in this guide comes from that experience: what we wish someone had handed us on day one, organized so you can pick it up whenever you need it over the next year, and put it down when you have had enough for today.

The Landstrom family, Family by Fate series

You have a community.

More than 1,100 people with Kleefstra syndrome are on the Kleefstra world map, and families talk every day in our private groups. Someone has already faced whatever you are facing this month.

There is a roadmap.

Kleefstra syndrome now has an international, evidence-based clinical guideline, a dedicated clinic at Boston Children's Hospital, and a clear set of first-year evaluations. You do not have to invent the plan.

Research is moving.

IDefine is funding gene therapy, RNA-based, and drug-repurposing programs aimed at treating Kleefstra syndrome itself. Your child's data, and your voice, are part of getting there.

Your child is the same child they were the day before the diagnosis. What has changed is that you now have a name for what you were seeing, a community that understands it, and a foundation working every day to change what it means.

Welcome to IDefine.

HOW TO USE THIS GUIDE

The guide follows the first year in order: the first 30 days, then months one through three, three through six, and six through twelve. Each stage tells you what to do now and what can wait. The back pages hold the care-team checklist, research programs, ways to get involved, and a directory of every link. Dog-ear it. Write in it. Hand it to a grandparent.

IN THIS GUIDE

What Kleefstra syndrome is, in plain language

Kleefstra syndrome (KS) is a rare genetic neurodevelopmental condition. It happens when one copy of a gene called **EHMT1** is missing or changed. EHMT1 sits at the tip of chromosome 9 (a region called 9q34.3) and acts like a dimmer switch for many other genes during brain development. With only one working copy, that switch does not work quite the way it should.

Most people with KS share a few features: **intellectual disability** (mild to severe), **low muscle tone** in infancy and childhood, **delayed speech** that is often more affected than understanding, and **distinctive facial features**. Many also experience sleep difficulties, constipation, heart differences, hearing or vision issues, seizures, or behavioral and mental-health challenges, especially in the teen and adult years. No two people with KS are alike, and the diagnosis does not predict your child's personality, humor, affection, or determination.

KS is almost always **de novo**, meaning it arose new in your child and was not inherited. Your genetics team can confirm this with parental testing and talk with you about what it means for future pregnancies and for siblings.

> Whatever you read online, remember that older articles describe the most severely affected children first. Your child's path is their own.

Kleefstra syndrome has its own code: Q87.86

Since October 1, 2024, KS has had its own ICD-10 diagnosis code in the United States, after IDefine families made the case to the CDC. It sounds like paperwork, but it is how insurers recognize the diagnosis, how researchers count how many people have KS, and how clinical trials find families. **Ask every provider to put Q87.86 in your child's chart**, and carry the wallet card from idefine.org.

Words you will hear

EHMT1
The gene involved in Kleefstra syndrome. It makes a protein (a histone methyltransferase) that helps regulate other genes.

9q34.3 deletion
A missing piece of chromosome 9 that includes EHMT1. Most people with KS have a deletion; the rest have a change within the EHMT1 gene itself.

Variant / mutation
A change in the spelling of the gene. "Pathogenic" means it is known to cause the condition; "VUS" means its significance is not yet certain. Ask your geneticist which yours is.

De novo
New in your child; not inherited from either parent.

Haploinsufficiency
One working copy of a gene is not enough. This is why KS happens and why several therapies aim to turn up the remaining copy.

Hypotonia
Low muscle tone. It affects feeding, posture, and motor milestones and is why physical therapy usually starts early.

Natural history study
Research that follows people over time to learn how a condition changes; the foundation every treatment trial is built on.

Regression
Losing skills that were previously gained. In KS it can appear in adolescence or adulthood and deserves urgent, not wait-and-see, attention.

ICD-10 code Q87.86
The diagnosis code for Kleefstra syndrome in U.S. medical records, insurance claims, and research databases. See below.

Trusted medical overviews: GeneReviews, MedlinePlus Genetics, and NORD. Links on page 11.

What to do now, and what can wait

Days 1–30
Breathe, gather, connect. Get the paperwork and the people in place.

Months 1–3
Baseline evaluations. Book the first-year checks the guideline recommends.

Months 3–6
Therapies, school, insurance. Build the everyday support system.

Months 6–12
Research, community, and a rhythm you can sustain.

Days 1–30

- [ ] **Get a copy of the genetic report.** Ask for the full lab report (not just the letter) and keep it. Note whether your child has a deletion or a variant within EHMT1, and whether it was confirmed de novo.
- [ ] **Ask for genetic counseling** if it was not already offered. A counselor will walk through the report, recurrence risk, and testing for siblings.
- [ ] **Tell us you are here.** Email **unlock@idefine.org**. A parent from IDefine will reach out to say hello and answer whatever is keeping you up at night.
- [ ] **Add your pin to the Kleefstra World Map** (kleefstraworldmap.org) and join the private caregiver group on Facebook. Read for a while; post when you are ready.
- [ ] **Sign up for Citizen Health and meet Ari. It is free for KS families.** Through IDefine's partnership, Ari gathers your child's records from every provider and portal in minutes, explains them in plain language, preps you for appointments, and drafts letters for school and insurance. It is the fastest way to get organized. (Prefer paper? A binder works too; see page 7.)
- [ ] **Give your pediatrician two things:** the clinical guideline summary (idefine.org/guideline) and the ICD-10 code, **Q87.86**. Most doctors have never seen a KS patient; the guideline tells them what to check, and the code makes sure the diagnosis is recorded properly.
- [ ] **Request an early-intervention evaluation** if your child is under three, or a school-district evaluation if older. Waitlists are long; get in line now.
- [ ] **Choose one person to update.** Let one trusted person carry the news for you this month.

Months 1–3

The 2026 International Clinical Guideline recommends these baseline evaluations at diagnosis; your pediatrician can order or refer for all of them. Full checklist on page 6.

- [ ] **Heart:** echocardiogram and ECG (cardiac MRI where available).
- [ ] **Hearing:** audiology assessment, then yearly until age six.
- [ ] **Vision:** ophthalmology referral, watching especially for strabismus and far-sightedness.
- [ ] **Speech and language:** a formal assessment now, and at least yearly until age twelve.
- [ ] **Sleep, growth, and constipation:** reviewed at this and every visit. Start a simple sleep diary.
- [ ] **Behavior and mental health:** a baseline review, repeated yearly.
- [ ] **Neurology** if there is any concern about seizures or lost skills.
- [ ] **Contact the Kleefstra Syndrome Clinic at Boston Children's Hospital** to ask about a visit. IDefine's Family Resource Packet explains how to prepare.

Building the everyday system

Months 3–6

- [ ] **Get therapies started:** physical therapy for low tone and motor skills, occupational therapy for feeding and daily skills, and speech-language therapy. Ask early about **augmentative and alternative communication (AAC)**. Signs, pictures, and devices support speech; they do not replace it.
- [ ] **Finalize the IFSP or IEP.** Bring the guideline and your one-page summary to the meeting. Other IDefine parents have been through this and will share what worked; ask in the caregiver group.
- [ ] **Look into insurance and waivers.** Many states offer Medicaid waivers for children with disabilities regardless of family income (kidswaivers.org is a good starting point). Ask about respite care while you are there.
- [ ] **Register with the research programs** that keep a permanent record of your child's journey: Citizen Health, Simons Searchlight, and RARE-X. See page 8. Each takes about an evening and matters for years.
- [ ] **Sign up for IDefine's newsletter** and follow us on social media, so research news reaches you without having to search for it.
- [ ] **Find your regional group.** Country-specific Facebook groups exist for Australia, Belgium, Brazil, France, New Zealand, and the U.K., alongside the main international group.

Months 6–12

- [ ] **Come to the conference.** IDefine's Kleefstra Syndrome Family Conference and Scientific Summit brings families, clinicians, and researchers together. Past conferences have included a newly diagnosed family orientation and programming for siblings; families facing financial hardship can email unlock@idefine.org about support.
- [ ] **Put the yearly checks on the calendar:** hearing, speech-language, behavior and mental-health review, growth, sleep, and constipation, plus any specialist follow-ups from the first round.
- [ ] **Consider joining a study.** The Natural History Study at Boston Children's Hospital and the Unravel Biosciences nasal-swab program are two ways families participate directly. Details on page 8.
- [ ] **Start your first campaign, if it feels right.** A birthday fundraiser, a walk, a bake sale, or a company match. Families fund most of IDefine's research; there is no minimum and no pressure. See page 9.
- [ ] **Mark September 17**, Kleefstra Syndrome Awareness Day, on the calendar.
- [ ] **Update your binder and medical summary.** A year in, you will be surprised how much has changed.
- [ ] **Check in on yourself and your partner.** Page 10 is for you.

> By the end of the first year, most families have a care team that knows KS, a school plan, a research profile, and a group chat full of people who get it. That is the goal. Not perfection; a system.

First-year evaluations from the clinical guideline

In January 2026 the first **International Clinical Evidence-Based Guideline for Kleefstra Syndrome** was published online in *Genetics in Medicine*, written by 27 clinical experts and 10 parent contributors across twelve care domains, with 66 recommendations. The ones that matter most in year one are below. Share this page with your pediatrician; the full guideline is at idefine.org/guideline.

DOMAIN	AT DIAGNOSIS	THEN
Genetics	Confirm the result; genetic counseling on inheritance, recurrence risk, and options for siblings.	Revisit if family planning questions arise.
Cardiology	Echocardiogram and ECG; cardiac MRI where available.	Yearly ECG from age 18; cardiology check before any medication affecting heart rhythm.
Hearing	Audiology assessment.	Yearly until age 6, then every 2 years until early adolescence. Treat ear infections and wax promptly.
Vision	Ophthalmology referral; watch for strabismus and hypermetropia.	Regular acuity checks; evaluate for cerebral visual impairment if suspected.
Speech & language	Speech-language assessment; consider AAC early.	At least yearly until age 12, tailored to hearing, vision, and cognition.
Growth	Height, weight, and head circumference.	Track at every visit; work up short stature or overweight; thyroid and glucose at puberty.
Sleep	Review sleep; start a diary.	Review at every age; use caution with melatonin and benzodiazepines (paradoxical reactions occur).
Constipation	Screen by history and exam.	Fluids, fiber, and activity first; gastroenterology if that fails.
Mental health & behavior	Baseline review.	Yearly; prompt referral when concerns arise; function-based behavioral support.
Neurology	EEG (awake and asleep) if seizures suspected; MRI if clinically indicated.	Neurologist or epilepsy specialist for confirmed seizures.
Regression	Know the signs: loss of skills, new psychosis, catatonia, severe insomnia.	Urgent referral, never wait-and-see; adaptive-behavior tracking over time.
Family support	Clear information; connection to IDefine and peer support.	Family wellbeing asked about at every visit; adult-transition planning by mid-adolescence.

The Kleefstra Syndrome Clinic

One of only a few clinics in the world built entirely around Kleefstra syndrome, at Boston Children's Hospital and supported by IDefine. Specialists see your child in one coordinated visit and send a plan home to your local team.

KleefstraSyndromeClinic@childrens.harvard.edu
617-355-3645 · idefine.org/family-resource-packet

If your local doctor has never seen KS

That is normal. Bring this page and say: "This is a rare condition with a published care guideline. Can we go through the baseline evaluations together?" IDefine can help connect your clinician with KS-experienced colleagues: unlock@idefine.org.

The practical scaffolding

Your records, in one place: Citizen Health and Ari

IDefine partners with Citizen Health so KS families can stop chasing paper, **at no cost to you**. Sign up (citizen.health/ai-advocate/kleefstra-syndrome), connect your patient portals, and **Ari**, Citizen Health's AI advocate, pulls your child's history together and keeps it current. Then it works for you: plain-language summaries, a briefing before each appointment, drafts of school and insurance letters, help with appeals, and answers to "what did the neurologist say in March?" Make sure it holds:

- The full genetic lab report and any letters from genetics.
- A **one-page medical summary**: diagnosis, key findings, medications and doses, allergies, specialists and their phone numbers, and the two or three things a new provider must know first.
- The guideline summary (page 6 works) and the ICD-10 code, Q87.86.
- Evaluation reports, therapy plans, and the IFSP or IEP.
- Insurance and waiver paperwork, and a running list of questions for the next visit.

With your permission, Citizen Health also shares de-identified data with KS researchers, so getting organized counts as research participation too. Prefer paper? A three-ring binder with the same contents, brought to every visit, still works.

Early intervention, IFSP, and IEP

In the U.S., children under three qualify for state early-intervention services through an **Individualized Family Service Plan (IFSP)**; from three on, the school district provides services through an **Individualized Education Program (IEP)**. Request an evaluation in writing at any time; no referral needed. Bring the guideline and ask specifically about speech-language, physical, and occupational therapy, and AAC.

Therapies that usually start early

Physical	Low muscle tone, sitting, crawling, walking, balance. Often the first therapy a family starts.
Occupational	Feeding, fine motor skills, sensory needs, dressing and daily routines.
Speech-language	Understanding and expression, oral-motor skills, and AAC (signs, pictures, devices).
Feeding	If sucking, chewing, or swallowing are hard, or growth is a concern.
Behavioral	Personalized, function-based support when behavior is getting in the way of learning or family life.

Insurance, waivers, and money

- **Medicaid waivers.** Many states cover children with significant disabilities regardless of parental income. Waitlists can be years long; apply early. Start at kidswaivers.org.
- **Respite care.** Often included in waiver programs. Use it.
- **ABLE accounts and special-needs trusts.** Ways to save for your child without affecting benefits. Worth a conversation with a special-needs planner in year one or two.
- **Appeal denials.** Therapy and equipment denials are routinely overturned on appeal with a letter of medical necessity. Ask the caregiver group for sample letters.

> The IDefine caregiver groups are the best IEP and insurance resource we know of. Ask; someone has the template.

RESEARCH YOU CAN JOIN

Be counted. It is the most powerful first step.

Treatments for rare conditions get built on data: how many people have it, how it changes over time, what a good outcome looks like. Every family that registers makes Kleefstra syndrome more visible to researchers, regulators, and companies deciding where to invest.

Citizen Health & Ari

IDefine's records partner, free for KS families. Ari gathers and organizes your child's medical records and, with your permission, shares de-identified data with KS researchers. Getting organized and being counted in one step.

citizen.health/ai-advocate/kleefstra-syndrome

Simons Searchlight

A long-running international registry for genetic neurodevelopmental conditions, with an EHMT1 community. Surveys, an optional blood sample, and regular newsletters on findings.

simonssearchlight.org · search EHMT1

RARE-X

A family-owned data platform where you complete structured surveys about your child's health and development that researchers can access.

rare-x.org/kleefstra

What IDefine is funding right now

IDefine believes intellectual disability disorders are treatable. These are the active programs and how families take part. Updates at idefine.org/research.

PROGRAM	WHAT IT IS	HOW FAMILIES PARTICIPATE
Natural History Study Boston Children's Hospital	A three-year longitudinal study gathering standardized data on how KS presents and changes over time, building the biomarkers and outcome measures any future trial will need.	Enroll through the Kleefstra Syndrome Clinic. Visits and assessments over time.
Gene therapy UT Southwestern	A two-year program led by Steven Gray, PhD, evaluating the feasibility and safety of EHMT1 gene replacement using next-generation delivery designed to reach the central nervous system.	Follow updates; registry data supports trial readiness.
RNA upregulation University of Chicago	Work in the Bryan Dickinson lab to develop targeted activators that "turn up" the remaining working copy of EHMT1.	Follow updates via the CSO Corner and newsletter.
"mRNA boosters" Johns Hopkins	A multi-foundation collaboration led by Jeff Coller, PhD, testing an RNA platform that boosts gene expression in haploinsufficiency disorders, using patient-derived cell models.	Follow updates.
Drug repurposing Unravel Biosciences	An AI-driven collaboration that builds "Living Molecular Twins" from patient RNA to screen tens of thousands of existing medicines for KS.	Families anywhere can contribute a non-invasive nasal-swab RNA sample.
Neural organoids UC San Diego	Dr. Angels Almenar-Queralt's lab grows brain-like cell models from IDefine-developed stem-cell lines to understand how KS affects the developing brain and to screen treatments.	Follow updates; IDefine's cell lines are shared with researchers.

> Our Chief Scientific Officer writes a plain-language "CSO Corner" about every project. It is the best way to understand where the science stands without a biology degree.

Ways to connect, volunteer, and fuel the research

IDefine is run by families. Every program on the previous page exists because a parent made a phone call, hosted an event, or gave what they could. When you are ready (and only then), here is how to join in.

Connect

- **Email unlock@idefine.org** to be paired with a parent who will talk with you one-on-one.
- **Private caregiver groups on Facebook** for day-to-day questions, IEP advice, and milestones, plus regional groups by country and an "All Grown Up" group for families of teens and adults.
- **The Kleefstra World Map** shows you families near you. Many local meetups start with a map search.
- **The annual Family Conference and Scientific Summit,** where families, clinicians, and researchers spend a weekend together.
- **Newsletter and social media** (Facebook, Instagram, LinkedIn, YouTube) for research news, event dates, and family stories.

Volunteer

IDefine has no large staff; it has parents with skills. Whatever you are good at, from writing and social media to fundraising, events, data, or translation, there is a place for it. Represent IDefine at a conference, help run an event, or share your family's story. Write to **unlock@idefine.org** with what you are good at.

THE MCDONALD FAMILY

Fundraise

Family-led fundraising is the engine behind IDefine's research. You do not need experience; you need a story, and you already have one.

- **Start a campaign** for a birthday, a milestone, or a diagnosis anniversary at idefine.org/fundraise-for-research. IDefine provides the page, the materials, and the support.
- **Host or join an event:** golf outings, walks and runs, community reunions, or your own idea; there is a custom fundraiser request form on the site.
- **Ask your employer about matching gifts.** Many companies double donations; IDefine's corporate matching page walks you through it.
- **Give directly,** including stock, crypto, or donor-advised funds, at idefine.org.
- **Wear it.** "I Love Someone Rare" apparel from the IDefine store starts conversations and raises funds.

Advocate

September 17 is Kleefstra Syndrome Awareness Day. Share your child's story when you are ready, tell your pediatrician's office about the guideline, or ask your state for a proclamation. Awareness leads to earlier diagnoses, and earlier diagnoses lead to better care.

This is a marathon. Pace yourself.

Rare-disease parents describe the first year as grief and joy at the same time, often in the same hour. Both are allowed. A few things other IDefine parents want you to know:

- **You do not have to do everything in this guide this month.** The roadmap exists so you can put things down and pick them up later.
- **Your relationship matters.** Partners often process a diagnosis differently and at different speeds. Say that out loud to each other early.
- **Siblings are watching.** Simple, honest language works. The caregiver groups have good resources for brothers and sisters.
- **Ask for help with specifics.** "Can you take Tuesday pickup?" is easier for friends to say yes to than "let me know if you need anything."
- **Talk to someone.** A counselor who works with medical families, a faith leader, or a parent who is a year ahead of you. Your own doctor is a fine place to start.
- **Celebrate small.** First sign, first bite, first night of sleep. Post it in the group. We will celebrate with you.

> The guideline itself says clinicians should ask about the wellbeing of the whole family at every visit. If they do not, tell them how you are doing anyway.

Questions for our care team

Our team

Pediatrician	
Geneticist	
Cardiology	
Audiology	
Ophthalmology	
Speech-language	
PT / OT	
Neurology	
Service coordinator	
IDefine parent contact	
ICD-10 code	**Q87.86** Kleefstra syndrome

RESOURCE DIRECTORY

Every link in this guide, in one place

IDEFINE

Home
idefine.org

Talk to a parent, questions, volunteering
unlock@idefine.org

Newly diagnosed page
idefine.org/newly-diagnosed

Clinical guideline & PDF
idefine.org/guideline

Clinic prep: Family Resource Packet
idefine.org/family-resource-packet

ICD-10 wallet cards (Q87.86)
idefine.org/wp-content/uploads/2024/09/Kleefstra_Syndrome-_ICD-10_code_IDcards-1.pdf

Research programs
idefine.org/research

Fundraise for research
idefine.org/fundraise-for-research

Corporate matching
idefine.org/corporate-matching-program

Stock, crypto, DAF gifts
idefine.org/ways-to-give-crypto-stock-daf

News & events, conference
idefine.org/news-events

Awareness Day (September 17)
idefine.org/awareness-day

Newsletter sign-up
idefine.org/newsletter

Store
bonfire.com/store/idefine

Social
facebook.com/IDefine.org · instagram.com/idefineorg · linkedin.com/company/idefine501c3 · YouTube: IDefine

COMMUNITY

Kleefstra World Map
kleefstraworldmap.org

Private caregiver group (Facebook)
facebook.com/groups/Kleefstra.Syndrome

All groups, including regional and All Grown Up
idefine.org/caregiver-community

CLINICAL CARE

Kleefstra Syndrome Clinic, Boston Children's Hospital
childrenshospital.org/services/kleefstra-syndrome-clinic
KleefstraSyndromeClinic@childrens.harvard.edu · 617-355-3645

Guideline citation
Bouman A, et al. International clinical evidence-based guideline for Kleefstra syndrome. *Genetics in Medicine*. 2026. doi:10.1016/j.gim.2026.102070

RESEARCH REGISTRIES

Citizen Health & Ari (free records + research)
citizen.health/ai-advocate/kleefstra-syndrome

Simons Searchlight (EHMT1)
simonssearchlight.org/research/what-we-study/ehmt1

RARE-X
rare-x.org/kleefstra

MEDICAL OVERVIEWS

GeneReviews
ncbi.nlm.nih.gov/books/NBK47079

MedlinePlus Genetics
medlineplus.gov/genetics/condition/kleefstra-syndrome

NORD
rarediseases.org/rare-diseases/kleefstra-syndrome

PRACTICAL SUPPORTS

Medicaid waivers by state
kidswaivers.org

Global Genes resource hub
resource-hub.globalgenes.org

NORD patient & caregiver center
rarediseases.org/for-patients-and-families

This guide is for information and community support. It is not medical advice. Always work with your child's care team. Content reflects IDefine programs and the 2026 clinical guideline as of the date on the cover; the online version at idefine.org/newly-diagnosed is kept current.

IDEFINE
The Kleefstra Syndrome Foundation

Together, we are turning barriers into bridges.

IDefine is a 501(c)(3) nonprofit founded by parents in 2020, dedicated to advancing research and finding a cure for Kleefstra syndrome, and finding it quickly.

REACH US

idefine.org
unlock@idefine.org

THIS EDITION

Last updated August 28, 2026
Current version: idefine.org/newly-diagnosed