



01 / START HERE

ASH1L Core Family Profile

A concise, person-first snapshot for a new clinician, appointment, school meeting, therapy handoff, or family planning. Complete only what is useful today.

COMPLETE ONLY WHAT HELPS

No field is required. Leave anything blank. A blank means unanswered—not absent. Save your copy and choose which pages are useful for each conversation.

Use this short profile first.

The separate 80-page Comprehensive Parent & Caregiver Questionnaire contains optional deep-dive modules. It is designed to be saved and completed over time—not in one sitting.

FAMILY-CHOSEN NAME OR CASE LABEL

Optional

CURRENT AGE OR AGE RANGE

Optional

DATE UPDATED

COMPLETED BY / RELATIONSHIP

BEFORE YOU SEND A COPY

Remove identifiers the recipient does not need. Exact genetic information can identify a person, especially in a rare-disease community.



02 / STRENGTHS, COMMUNICATION & ACCESS

Start with the person

Describe the person before the diagnosis. Use the person's and family's own words wherever possible.

STRENGTHS AND INTERESTS

PERSON-DEFINED GOALS

PREFERRED LANGUAGES

COMMUNICATION MODES

HOW YES, NO, STOP, PAIN, HELP AND CHOICE ARE EXPRESSED

HOW TO INCLUDE THE PERSON IN DECISIONS

PROCESSING, SENSORY AND ACCESS SUPPORTS

CONDITIONS FOR COMFORT AND SUCCESSFUL PARTICIPATION



03 / CURRENT SNAPSHOT

Priorities, reliable baseline & safety

A reliable baseline helps a new team recognize meaningful change. Describe usual function and the supports already known to help.

PRIORITY 1

PRIORITY 2

PRIORITY 3

USUAL COMMUNICATION BASELINE

USUAL MOVEMENT BASELINE

USUAL SLEEP AND ENERGY BASELINE

USUAL FEEDING, GI AND COMFORT BASELINE

USUAL PARTICIPATION AND REGULATION BASELINE

HOW PAIN OR DISTRESS MAY APPEAR

FIRST-LINE SAFETY AND SUPPORT INFORMATION



04 / COPY, DO NOT REINTERPRET

Genetic & health essentials

Preserve the laboratory wording and source. Exact genetic information is optional and may be identifying.

LABORATORY / REPORT DATE / CLINICIAN SOURCE

GENE, TRANSCRIPT AND VARIANT - EXACTLY AS REPORTED

LABORATORY CLASSIFICATION

☐ Pathogenic ☐ Likely pathogenic ☐ VUS ☐ Other ☐ Unknown

INHERITANCE AS REPORTED

☐ De novo ☐ Maternal ☐ Paternal ☐ Inherited, source unclear ☐ Not tested ☐ Unknown / other

CURRENT DIAGNOSES AND UNRESOLVED CONCERNS

MEDICATIONS AND SUPPLEMENTS

ALLERGIES, INTOLERANCES AND ADVERSE REACTIONS

IMPORTANT PROCEDURES OR HOSPITALIZATIONS

KEY CLINICIANS, SERVICES AND SAFETY INFORMATION



05 / SUPPORT IN CONTEXT

Function & participation

Choose the description that fits most often, then note the support, setting, or state that changes access. “Varies” is a valid answer.

COMMUNICATION AND CHOICE

☐ Independent ☐ Some support ☐ Full assistance ☐ Varies by state ☐ Not assessed / prefer not

SUPPORT / CONTEXT NOTE

MOBILITY AND TRANSFERS

☐ Independent ☐ Some support ☐ Full assistance ☐ Varies by state ☐ Not assessed / prefer not

SUPPORT / CONTEXT NOTE

EATING, DRINKING AND PERSONAL CARE

☐ Independent ☐ Some support ☐ Full assistance ☐ Varies by state ☐ Not assessed / prefer not

SUPPORT / CONTEXT NOTE

LEARNING, WORK AND COMMUNITY PARTICIPATION

☐ Independent ☐ Some support ☐ Full assistance ☐ Varies by state ☐ Not assessed / prefer not

SUPPORT / CONTEXT NOTE

SLEEP AND REGULATION

☐ Independent ☐ Some support ☐ Full assistance ☐ Varies by state ☐ Not assessed / prefer not

SUPPORT / CONTEXT NOTE

HEALTH SELF-MANAGEMENT

☐ Independent ☐ Some support ☐ Full assistance ☐ Varies by state ☐ Not assessed / prefer not

SUPPORT / CONTEXT NOTE



06 / BASELINE → CONTEXT → CHANGE → MEASURE → RECOVERY

One important change over time

Chronology is useful evidence. Timing alone does not establish causation, and a difficult window may have more than one explanation.

EVENT OR CHANGE

AGE OR DATE

BASELINE BEFORE THE WINDOW

CONTEXT / ACTIVE FACTORS

WHAT CHANGED

WHAT WAS OBJECTIVELY MEASURED

ACTION OR SUPPORT TRIED

RECOVERY

MOST USEFUL NEXT QUESTION

CURRENT STATUS

☐ Returned to baseline ☐ Partial recovery ☐ New baseline ☐ Ongoing ☐ Uncertain

SOURCE

☐ Person/family ☐ Original record ☐ Clinician ☐ School/therapy ☐ Multiple ☐ Unclear



07 / PREPARE ONE USEFUL HANDOFF

Records, questions & next steps

This is a records inventory—not a request to upload or email records. The family controls access and can redact or withhold any item.

KEY RECORDS AVAILABLE

WHERE RECORDS ARE HELD

SHARING OR REDACTION LIMITS

CURRENT CARE-TEAM QUESTIONS

NEXT STEP 1

OWNER AND TARGET DATE 1

NEXT STEP 2

OWNER AND TARGET DATE 2

MATCH THE COPY TO THE READER

A new clinician, teacher, therapist, or researcher may need different pages. Choose the pages that answer the question in front of them.



08 / FAMILY CONTROL

Review this copy before it leaves your hands

Check names, dates, private details, and the intended reader before you send selected pages.

INFORMATION THAT MAY BE UNCERTAIN OR OUTDATED

INTENDED RECIPIENT

PAGES SELECTED FOR SHARING

OPTIONAL CONTACT METHOD

INFORMATION THAT SHOULD REMAIN PRIVATE

INTENDED PURPOSE

CONTACT PREFERENCE

REVIEWED BY / ROLE / DATE

STATUS OF THIS COPY

☐ Draft ☐ Current ☐ Recipient-specific ☐ Archive ☐ Uncertain

- ☐ I saved a private copy before sharing.
- ☐ I removed identifiers the recipient does not need.
- ☐ Blank fields are understood as unanswered—not negative.
- ☐ Your observations and original records remain clearly separated.
- ☐ I chose the reader, purpose, and pages for this copy.

YOU CONTROL THIS COPY

Share it only when it is useful, with the people you choose, for the purpose you choose. Keep your own copy so you can update it later.