FAMILIES / CARE TEAMS

Prepare for an ASH1L-related appointment

A spacious, print-ready workbook for defining the current question, organizing relevant evidence, and leaving with a shared plan.

USE THIS WHEN

Before a first, new-specialty, or complex visit.

TIME NEEDED

20 to 30 minutes

TAKE OR SHARE

Bring only the pages relevant to this visit.

Bring the smallest useful packet.

The goal is not to prove that every concern is caused by ASH1L. The goal is to define the person, the current question, the reliable baseline, and what information would change care.

01 / START HERE

The one-page visit brief

Complete this page first. It should let a clinician understand the person and the current question before opening the larger record.

REASON FOR THIS VISIT

What question needs an answer today?

RELIABLE BASELINE

What can the person usually access, and with what supports?

WHAT CHANGED - AND WHEN

Separate longstanding features from a new concern.

COMMUNICATION, SENSORY, MOVEMENT, AND VISIT SUPPORTS

TOP THREE QUESTIONS

Write them in priority order.

02 / PACKET, BASELINE, AND ACCESS

Prepare the packet and describe the person

Bring original reports connected to today's question. Reliable baseline is the reference point for deciding whether something is longstanding, variable, or new.

- [ ] Complete signed genetic laboratory report
- [ ] Current medication, supplement, allergy, and reaction list
- [ ] Age-ordered chronology on page 4
- [ ] Original reports relevant to today's question
- [ ] Existing emergency plan, if one applies

PREFERRED NAME, COMMUNICATION, AND PAIN SIGNALS

How does the person best understand, answer, and show discomfort?

RELIABLE FUNCTION

Communication, movement, attention, participation, eating, sleep, bowel pattern, and daily living.

WHAT MAKES ACCESS EASIER - AND EARLY CHANGE SIGNALS

People, pacing, sensory supports, positioning, preparation, recovery time, and observable early changes.

SAFETY THRESHOLDS AND ACCESS NEEDS FOR THE VISIT

Use the person's existing plan and clinician guidance.

03 / TIMELINE

Age-ordered chronology

Use an exact date only when it matters. Otherwise, an age band or month/year is enough. Record baseline or event, context, objective source, and recovery.

1 **AGE / DATE**

BASELINE OR EVENT

2 **AGE / DATE**

BASELINE OR EVENT

3 **AGE / DATE**

BASELINE OR EVENT

4 **AGE / DATE**

BASELINE OR EVENT

04 / DURING THE VISIT

Connect each test to a decision

A test is useful when everyone understands the question, the acquisition conditions, and what result would change the plan.

QUESTION OR PROPOSED TEST 1

What will it answer? What must be captured? What result changes next steps? Who reviews it?

QUESTION OR PROPOSED TEST 2

What will it answer? What must be captured? What result changes next steps? Who reviews it?

QUESTION OR PROPOSED TEST 3

What will it answer? What must be captured? What result changes next steps? Who reviews it?

QUESTION OR PROPOSED TEST 4

What will it answer? What must be captured? What result changes next steps? Who reviews it?

Interpret a normal result narrowly.

Timing, acquisition quality, sleep or event capture, sensitivity, and the relevant clinical window determine what a negative result can reasonably exclude.

05 / CLOSE AND FOLLOW UP

Leave with a shared plan

Before the visit ends, assign an owner and date to the next step. Unowned follow-up is not a plan.

LEADING EXPLANATIONS

Keep competing explanations visible.

NEXT TEST, OBSERVATION, OR REFERRAL

What exact question will it answer?

WHAT CAN BE DONE NOW

Supports or care that do not require the evaluation to be complete.

URGENT CHANGE THAT SHOULD TRIGGER EARLIER REVIEW

Use the existing emergency plan and clinician guidance.

RESULTS OWNER AND FOLLOW-UP DATE

Who communicates what, to whom, and by when?

Keep ASH1L visible without diagnostic overshadowing.

The molecular diagnosis can guide attention, but a new symptom, loss of function, pain signal, spell, sleep change, medication reaction, infection, or mental-health change still deserves its own differential.