Example only. A fictional child, not a real person or record.

Sample Case File for Sam

Case File summary · version 1 · 23 Nov 2026

Born 14 Mar 2022 · 4 years 8 months

R = medical record · P = parent · T = care team

OUR QUESTION

"Sam used about 20 words at age 2 and then stopped using most of them. We want to understand why." P

SNAPSHOT

Doctors have said: Speech delay, low muscle tone as a baby P
Medicines: Melatonin P
Allergies: Amoxicillin R

MAIN CONCERNS

In our words	Clinical term	Started	Now	Source
stopped using words	Developmental regression	2 years 1 month	present	P
not putting words together	Delayed speech and language development	2 years	present	P
staring spells		3 years	present	P

3 more in Section B of the full Case File

GROWTH

Weight 16.2 kg (Oct 2026) [P] · Height 89.5 cm (Apr 2024) [R] · Head size 47.8 cm (Apr 2024) [R]

FAMILY

Yes; Who: Mother's brother; What: Seizures as a young child; Parents related by blood: No P

TESTING ON FILE

Chromosomal microarray · May 2024 (2 years 2 months) · "No clinically significant copy number changes detected." R
Fragile X (FMR1) · May 2024 (2 years 2 months) · "Normal. 29 CGG repeats." R
2 more in Section I of the full Case File

OUR TOP QUESTIONS

1. Is a broader test, such as exome or genome sequencing, worth doing now?
2. Sam lost words at 2 years 1 month. Does that change how soon we should be seen or what should be checked?
3. Are you satisfied with the EEG and MRI we have, or should either be repeated?

WHAT WE ARE ASKING FOR

A plan for which tests make sense next, and what we can start at home now P

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A. Timeline

When	What	Source
July 2022 (4 months)	Diagnosis recorded: Hypotonia	R
February 2023 (10 months)	Started Ferrous sulfate drops	R
August 2023 (1 year 4 months)	Stopped Ferrous sulfate drops	R
April 2024 (2 years)	Diagnosis recorded: Speech delay	R
April 2024 (2 years)	Clinic visit at Example Children's Hospital (speech)	R
May 2024 (2 years 2 months)	Test: Chromosomal microarray	R
May 2024 (2 years 2 months)	Test: Fragile X (FMR1)	R
January 2025 (2 years 9 months)	Clinic visit at Example Children's Hospital (staring spells)	R
January 2025 (2 years 9 months)	Test: EEG	R
March 2025 (2 years 11 months)	Started Melatonin	P

How old was Sam when you first noticed it?
Age: 2 years [P]

B. Concerns in clinical terms

In our words	Clinical term	Started	Status	Source
trouble hearing			checked and absent	P
floppy as a baby	Generalized hypotonia (HP:0001290)	4 months	resolved	R
stopped using words	Developmental regression (HP:0002376)	2 years 1 month	present	P

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In our words	Clinical term	Started	Status	Source
not putting words together	Delayed speech and language development (HP:0000750)	2 years	present	P
staring spells		3 years	present	P
wakes many times a night		2 years 6 months	present	P

In your own words, what worries you most about Sam right now?
Sam used about 20 words at age 2 and then stopped using most of them. We want to understand why. [P]

Which areas concern you?
Talking and understanding, Moving and muscle tone, Sleep [P]

C. Pregnancy, birth and newborn

Was Sam born early?
No [P]

D. Development

Has Sam lost any skill they used to have, like words or walking?
Yes; Which skill: words; When: 2 years 1 month [P]

E. Growth

Measure	Value	When	Source
Head size	47.8 cm	April 2024 (2 years)	R
Height	89.5 cm	April 2024 (2 years)	R
Weight	12.4 kg	April 2024 (2 years)	R
Weight	16.2 kg	October 2026 (4 years 6 months)	P

Has a doctor ever said Sam's head is small or large for their age?
No [P]

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F. Medical history

Diagnosis	Status	Since	Source
Hypotonia (P94.2)	active	July 2022 (4 months)	R
Speech delay (F80.9)	active	April 2024 (2 years)	R

Allergies
Amoxicillin (hives, mild) [R]

Has Sam ever had a seizure or a staring spell?
Yes [P]

What have doctors called it so far?
Speech delay, low muscle tone as a baby [P]

G. Medicines and therapies tried

Medicine	Dose	Dates	For, what happened	Source
Melatonin	1 mg at bedtime	From March 2025 (2 years 11 months)	For night waking	P
Ferrous sulfate drops	15 mg once a day	From February 2023 (10 months) To August 2023 (1 year 4 months)	For iron level	R

H. Family history

Relative	Living	Age	Conditions	Started at	Source
maternal uncle	living	34	seizures as a young child	3	P
sister	living	7			P

Does anyone else in the family have similar concerns?
Yes; Who: Mother's brother; What: Seizures as a young child [P]

Are Sam's parents related by blood, for example cousins?
No [P]

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I. Testing done

Test	When	Ordered by, lab	Result as written	Source
Chromosomal microarray	May 2024 (2 years 2 months)	Developmental pediatrics, Example Genetics Lab Parents tested: no	“No clinically significant copy number changes detected.”	R
Fragile X (FMR1)	May 2024 (2 years 2 months)	Developmental pediatrics, Example Genetics Lab Parents tested: no	“Normal. 29 CGG repeats.”	R
EEG	January 2025 (2 years 9 months)	Neurology	“Normal awake and sleep EEG.”	R
Hearing test	at about 2 and a half		No report on file	P

What tests has Sam had that you remember?
Genetic test, EEG, Hearing test [P]

K. Questions for this visit

1. Is a broader test, such as exome or genome sequencing, worth doing now?
Answer:

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2. Sam lost words at 2 years 1 month. Does that change how soon we should be seen or what should be checked?
Answer:

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3. Are you satisfied with the EEG and MRI we have, or should either be repeated?
Answer:

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4. Should Sam see an eye doctor?
Answer:

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5. Is there a research program for undiagnosed children we should apply to? Would you write the recommendation letter?
Answer:

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6. Could the staring spells be related to how often Sam wakes at night?
Answer:

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7. What should make us call you or go to the emergency room before the next visit?
Answer:

What do you most want from the next appointment?
A plan for which tests make sense next, and what we can start at home now [P]

What would an answer change for your family?
School support, Planning for another child [P]

L. Care team and records

Records connected
1 hospitals, 6 documents, March 2022 to October 2026

Reports still to request
Hearing test

SAMPLE