

The Angelman Syndrome Online Registry

In this study we want to collect clinical and genetic findings of Angelman Syndrome and collect data of affected individuals in a local patient registry at the Institute of Human Genetics at the University of Leipzig. You have the possibility to view the detailed study information at any time. After registration you can start entering data and interrupt this at any time and continue at a later time by means of a login and ID. For various questions you have the possibility to upload documents (e.g. molecular genetic findings, electroencephalography, magnetic resonance imaging, doctor's letters, etc.), which may be viewed and pseudonymized by a member of the study staff and transferred to the study database. The evaluation and storage of the data is pseudonymized. The aim of the study is to establish an Angelman Syndrome patient registry, a better understanding of genotype-phenotype correlations and in the future an improvement in the medical treatment of affected patients. This registry study was approved by the Ethics Committee of the University of Leipzig under the reference number 465/19-ek. You are welcome to fill out the questionnaire in great detail and comprehensively, but it is not a problem if you cannot or only want to answer some of the questions very briefly. If you have any uncertainties, questions or suggestions, please contact the Institute of Human Genetics at the University of Leipzig (angelman@medizin.uni-leipzig.de).

Patient Information

Last name of patient: _____

First name of patient: _____

Registration of contact person

Last name of contact person: _____

First name of contact person: _____

E-Mail of contact person: _____

Legal disclosures

ID of contact person: _____

(The contact ID was either sent to you in a previous confirmation email, or (for new contacts) is entered from researcher of this study).

A medical validation has been carried out? Yes

(filled from researcher)

Consent form

The voluntarily informed consent is the legal framework for processing the data according to the General Data Protection Regulation (GDPR) and the Declaration of Helsinki (Declaration of the World Medical Association on Ethical Principles for Medical Research on Humans).

Below you will find the patient's consent to the use of patient data for participation in the project Angelman Registry at the Institute of Human Genetics of the Leipzig University Hospital.

Project manager:

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I have the right not to agree to this declaration of consent - but since this project depends on the collection and processing of personal and medical data, non-signature would preclude participation in the project and the resulting further studies. Please read through the information for volunteers and contact us if any questions arise.

We are sufficiently informed about the purpose and procedure of the study. The data is entered by me/us on behalf of and in agreement with the legal guardians. We read the study information (link) and are aware that participation in the study is voluntary and free of charge for us, that we do not receive any remuneration, bonus or other share in financial benefits and profits that may be obtained on the basis of the research with our data. We are aware that this consent can be retract at any time without giving reasons and without any disadvantages for us.

Ja

We agree that the clinical and genetic data of our child or guardian may be used in pseudonymized form in the study "Genotype-Phenotype Correlation in Angelman Syndrome" and entered into the database and published if necessary.

Ja

In case of future questions or new findings that could be relevant to us, we agree to be re-contacted.

Ja

Nein

The e-mail address entered on the previous page can be used for this purpose.

Ja

Nein

if not:

alternative e-mail address: _____

Place, date and

Signature of the contact person: _____

General Information

Unless otherwise stated, please tick the most appropriate answer.

Questionnaire was answered by:

- parents (part) / relative / legal representative
- responsible doctor
- other

The questionnaire was answered by (other):

Sex of the patient:

- female
- male
- unknown

Patient's nationality:

Patient's date of birth:

(Day-Month-Year)

_____ - _____ - _____

Has a standardized IQ/EQ test ever been performed?

- yes
- no
- unknown

if yes:

Name of the IQ/EQ test:

Result of the IQ/EQ test:

Degree of the patient's disability:

- normal and age-appropriate
- mildly delayed
- moderately delayed
- strongly delayed
- profoundly / massively delayed

Degree of the patient's disability:

- degree: _____
- unknown

Has this severity been medically confirmed?

- yes
- no
- unknown

Questions regarding pregnancy

Unless otherwise stated, please tick the most appropriate answer.

Did the pregnancy occur spontaneously?

(i.e. without the aid of an assisted reproduction method such as artificial insemination)

- yes
 no
 unknown

How old was the mother when the patient was born?

- Age: _____ years
 unknown

How old was the father when the patient was born?

- Age: _____ years
 unknown

Questions about birth

Unless otherwise stated, please tick the most appropriate answer.

In which week of pregnancy was the delivery?

- _____ week
 unknown

Please enter the following information:

Birth weight (in g): _____ g

Birth length (in cm): _____ cm

Head circumference at birth (in cm): _____ cm

Questions about the physical development

Unless otherwise stated, please tick the most appropriate answer.

Have there been or are there currently any feeding problems:

(You can select more than one.)

drinking weakness / feeding disorder

if yes:

At what age did the symptoms begin?

- Age: _____ month
 _____ years
 unknown

At what other age did the symptoms begin?

- yes
 no
 unknown

If applicable: At what age did the symptoms end?

- Age: _____ month
 _____ years
 unknown

problems with weight gain

if yes:

At what age did the symptoms begin?

Age: _____ month
_____ years

unknown

At what other age did the symptoms begin?

yes
 no
 unknown

If applicable: At what age did the symptoms end?

Age: _____ month
_____ years

unknown

How severe are the symptoms?

mild
 moderate
 severe
 unclear

reflux

if yes:

At what age did the symptoms begin?

Age: _____ month
_____ years

unknown

At what other age did the symptoms begin?

yes
 no
 unknown

If applicable: At what age did the symptoms end?

Age: _____ month
_____ years

unknown

How severe are the symptoms?

mild
 moderate
 severe
 unclear

obstipation

if yes:

At what age did the symptoms begin?

Age: _____ month
_____ years

unknown

At what other age did the symptoms begin?

yes
 no
 unknown

If applicable: At what age did the symptoms end?

Age: _____ month
_____ years

unknown

How severe are the symptoms?

mild
 moderate
 severe
 unclear

(suspected) abdominal pain

if yes:

At what age did the symptoms begin?

Age: _____ month
_____ years

unknown

At what other age did the symptoms begin?

yes
 no
 unknown

If applicable: At what age did the symptoms end?

Age: _____ month
_____ years

unknown

How severe are the symptoms?

mild
 moderate
 severe
 unclear

diarrhea

if yes:

At what age did the symptoms begin?

Age: _____ month
_____ years

unknown

At what other age did the symptoms begin?

yes
 no
 unknown

If applicable: At what age did the symptoms end?

Age: _____ month
_____ years

unknown

How severe are the symptoms?

mild
 moderate
 severe
 unclear

tube feeding (e.g. nasopharyngeal or PEG)

if yes:

At what age did the symptoms begin?

Age: _____ month
_____ years

unknown

At what other age did the symptoms begin?

yes
 no
 unknown

If applicable: At what age did the symptoms end?

Age: _____ month
_____ years

unknown

How severe are the symptoms?

mild
 moderate
 severe
 unclear

Is there scoliosis (curvature of the spine)?

- yes
- no
- unknown

if yes:

Specify the degree of severity:

- mild
- moderate
- severe
- unclear

Questions on gross-motor development

Unless otherwise stated, please tick the most appropriate answer.

Please evaluate the muscle tonus (muscle tension) of the patient.

1) trunk musculature:

- hypotone (reduced muscle tension)
- normotone (normal muscle tension)
- hypertone (increased muscle tension/spasticity)
- unknown

Specify the degree of severity:

- mild
- moderate
- severe
- unclear

2) arm musculature:

- hypotone (reduced muscle tension)
- normotone (normal muscle tension)
- hypertone (increased muscle tension/spasticity)
- unknown

Specify the degree of severity:

- mild
- moderate
- severe
- unclear

3) leg musculature:

- hypotone (reduced muscle tension)
- normotone (normal muscle tension)
- hypertone (increased muscle tension/spasticity)
- unknown

Specify the degree of severity:

- mild
- moderate
- severe
- unclear

Is a wheelchair necessary?

- yes
 sometimes
 no
 unknown

if yes:

At what age was a wheelchair necessary? _____ months _____ years

Please describe the development of the patient's gross-motor functions.

The patient's gross motor development continues.
(the patient learns new skills or improves already learned skills)

- not applicable
 applicable
 unknown

Since the following age the patients' gross motor development stops.

- not applicable
 applicable, at age: _____ month _____ years
 unknown

Since the following age the patient loses already learned gross motor skills (regression):

- not applicable
 applicable, at age: _____ month _____ years
 unknown

Questions on fine-motor development

Unless otherwise stated, please tick the most appropriate answer.

What are the patient's fine-motor functions:

Select all applicable development steps and indicate the age from which this was possible.

Grasping objects

- Age: _____ month
 _____ years
 unknown

Objects can be guided from hand to mouth

- Age: _____ month
 _____ years
 unknown

Objects can be passed from one hand to the other

- Age: _____ month
 _____ years
 unknown

Pincer grasp

(grasping items with thumb and forefinger)

- Age: _____ month
 _____ years
 unknown

- Can eat with the fingers
 - Age: _____ month
_____ years
 - unknown

- Correct use of objects
(e.g. cutlery, pens, etc.)
 - Age: _____ month
_____ years
 - unknown

- None of the above skills are possible
 - Age: _____ month
_____ years
 - unknown

Please describe the development of the patient's fine-motor functions:

- The patient's fine motor development continues.
(the patient learns new skills or improves already learned skills)
 - not applicable
 - applicable
 - unknown

- Since the following age the patients' fine-motor development stops:
 - not applicable
 - applicable, at age: _____ month _____ years
 - unknown

- Since the following age the patient loses already learned fine-motor skills (regression):
 - not applicable
 - applicable, at age: _____ month _____ years
 - unknown

Questions about movement

Unless otherwise stated, please tick the most appropriate answer.

Which remarkable movement patterns are present?

Select all applicable movement patterns and indicate the degree of severity.

- dystonia (stiffening)
 - mild
 - moderate
 - severe
 - unclear

- dystone emotional triggered stiffening
 - mild
 - moderate
 - severe
 - unclear

- dyskinesia (disturbed movement pattern)
 - mild
 - moderate
 - severe
 - unclear

- hyperkinesia (involuntary extra movements)
- mild
 moderate
 severe
 unclear
- ataxia (coordination and/or gait disorder)
- mild
 moderate
 severe
 unclear

Questions on language development and communication

Unless otherwise stated, please tick the most appropriate answer.

- What is the highest level of speech development?
- no expressive language
 sounds (vowels)
 babbles (vowels + consonants)
 speaks single words
 speaks two-word sentences
 speaks multi-word sentences
 normal speech
 unknown

- At what age the patient starts to speak first words:
- _____ month _____ years
 unknown

Please describe the patient's speech development:

- The patient's speech development continues.
(the patient learns new skills or improves already learned skills)
- not applicable
 applicable
 unknown
- Since the following age the patient's speech development stops:
- not applicable
 applicable, at age: _____ month _____ years
 unknown
- Since the following age the patient loses already learned speech skills:
- not applicable
 applicable, at age: _____ month _____ years
 unknown

How does the patient communicates mostly?

- sounds
- words
- sentences
- signing, gestures
- facial expression
- images
- tablet, PC
- turn of eyes

At what age did social smiles first appear?

- did not occur so far
- age: _____ month _____ years
- unknown

Question on the continence development

Unless otherwise stated, please tick the most appropriate answer.

Does the patient have gained daytime bladder control?

- yes, always
- yes, partially
- yes, almost
- no
- unknown

Does the patient have gained bladder control during sleep?

- yes, always
- yes, partially
- yes, almost
- no
- unknown

Is the patient stool-continent (clean) during the day?

- yes, always
- yes, partially
- yes, almost
- no
- unknown

Is the patient stool-continent (clean) at night?

- yes, always
- yes, partially
- yes, almost
- no
- unknown

Questions about behavior

Unless otherwise stated, please tick the most appropriate answer.

Does the patient shows repetitive (recurring) behaviour?

- yes, always
- yes, partially
- yes, almost
- no
- unknown

Does the patient show repetitive (recurring) hand movements?

- yes, always
- yes, partially
- yes, almost
- no
- unknown

Does the patient show repetitive (recurring) movements of the whole body?

- yes, always
- yes, partially
- yes, almost
- no
- unknown

Is the patient suffering from pica syndrome? This means that the patient eats uneatable substances or objects.

- yes, always
- yes, partially
- yes, almost
- no
- unknown

Is the patient afraid of new situations?

- yes, always
- yes, partially
- yes, almost
- no
- unknown

Does the patient easily make new social contacts?

- yes, always
- yes, partially
- yes, almost
- no
- unknown

Does the patient show aggressive behaviour?

- yes, always
- yes, partially
- yes, almost
- no
- unknown

Against whom are aggressive behavior patterns intended?

- directed against itself
- directed against others
- directed against itself and others
- unclear / unknown

Zeigt der Patient/die Patientin hyperaktives Verhalten?

- yes, always
- yes, partially
- yes, almost
- no
- unknown

Does the patient show hyperactive behavior?

- yes, always
- yes, partially
- yes, almost
- no
- unknown

Is the patient unable to concentrate?

- yes, always
- yes, partially
- yes, almost
- no
- unknown

Is the patient fascinated by water?

- yes, always
- yes, partially
- yes, almost
- no
- unknown

Is the patient fascinated by plastic?

- yes, always
- yes, partially
- yes, almost
- no
- unknown

Does the patient often seem joyful?

- often appears cheerful
- smiles often without any apparent reason
- laughs often without any apparent reason
- has laughing episodes
- laughs in sleep
- no / unknown

Does the patient show other behavioral features?

Is the patient's behavior or behavioral specifics influenced by medication or dietary supplements?

- yes
- no
- unknown

if yes:

How is the patient's behavior or behavioral characteristics affected by medications or dietary supplements?

Questions about sleep behavior

Unless otherwise stated, please tick the most appropriate answer.

At what age did the patient sleep through the night for the first time?

- _____ months _____ years
- unknown

Does the patient have a regularly sleeping rhythm?

- yes, always
- yes, mostly
- yes, partially
- rarely
- no
- unknown

Does the patient have problems falling asleep?

- yes, always
- yes, mostly
- yes, partially
- rarely
- no
- unknown

Does the patient have problems sleeping through the night?
(sleep through the night: at least 4-5 hours at a time)

- yes, always
- yes, mostly
- yes, partially
- rarely
- no
- unknown

Are there any further additions/remarks on sleep behavior?

Questions about the EEG

Unless otherwise stated, please tick the most appropriate answer.

At what age was the first EEG performed?

- _____ months _____ years
- unknown

Are there were any noticeable abnormalities in background activity?

- yes
- no
- unknown

What abnormalities in background activity did this initial EEG show?

- focal slowing
- generalized slowing
- missing sleep patterns
- other

What other abnormalities in background activity did this initial EEG show?

Did the initial EEG show epilepsy related abnormalities?

- yes
- no
- unknown

Which epilepsy typical abnormalities did the initial EEG show?

- Focal
- Multi-focal
- Generalized

Did one or more of the following characteristics ever appear on EEG?

Deterioration of the EEG background

if yes:

At what age did the deterioration of the EEG background begin?

- _____ months _____ years
- unknown

Does the deterioration of the EEG background persists?

- yes
- no
- unknown

If applicable: When did the deterioration of the EEG background end?

- _____ months _____ years
- unknown

Hypsarrhythmia

if yes:

At what age did the hypsarrhythmia start?

- _____ months _____ years
- unknown

Does the hypsarrhythmia persists?

- yes
- no
- unknown

If applicable: When did the hypsarrhythmia end?

- _____ months _____ years
- unknown

Burst Suppression

if yes:

At what age did the burst suppression start?

- _____ months _____ years
 unknown

Does the burst suppression persists?

- yes
 no
 unknown

If applicable: When did the burst suppression end?

- _____ months _____ years
 unknown

Slow-spike waves

if yes:

At what age did the slow spike-waves begin?

- _____ months _____ years
 unknown

Do the slow spike-waves persist?

- yes
 no
 unknown

If applicable: When did the slow spike-waves end?

- _____ months _____ years
 unknown

ESES/CSWS pattern

(Electrical Status Epilepticus During Slow Sleep/Continuous Spikes and Slow Waves during Slow Sleep)

if yes:

At what age did the ESES/CSWS pattern begin?

- _____ months _____ years
 unknown

Does the ESES/CSWS pattern persists?

- yes
 no
 unknown

If applicable: When did the ESES/CSWS pattern end?

- _____ months _____ years
 unknown

What other characteristics
appeared during the course?

- Are original EEG diagnostic reports available for the study?
- yes
 no
 unknown

You can send us the original EEG findings by regular mail or e-mail, should they be available digitally for the study. Please make personal information, e.g. names, illegible.

Questions about epilepsy

Unless otherwise stated, please tick the most appropriate answer.

- Does the patient has epilepsy?
- yes
 no
 unknown

if yes:

Has the patient one or more of the following seizures?
 (please select all applicable seizure types)

- febrile seizures

if yes:

What was the patient's age at the beginning of the febrile seizures?

_____ month _____ years
 unknown

What was the patient's age at the end of the febrile seizures?

_____ month _____ years
 unknown

What is the maximum seizure frequency at which febrile seizures occur?

Less than five seizures per year
 Between five and ten seizures per year
 Between one and five seizures per month
 Between one and five seizures per week
 Between one and five seizures per day
 More than ten seizures per day

What is the current seizure frequency at which the febrile seizures occurred?

Less than five seizures per year
 Between five and ten seizures per year
 Between one and five seizures per month
 Between one and five seizures per week
 Between one and five seizures per day
 More than ten seizures per day

Are there protecting factors regarding febrile seizures?

Are there factors that promote febrile seizures?

Note:

Absence seizures

if yes:

What kind of absences seizures occur?

- typical
- atypical
- with additional features (e.g. myoclonic, eyelid closure myoclonia etc)

What was the patient's age at the beginning of the absences?

- _____ month _____ years
- unknown

What was the patient's age at the end (if present) of the absences?

- _____ month _____ years
- unknown

What is the maximum seizure frequency at which absences occur?

- Less than five seizures per year
- Between five and ten seizures per year
- Between one and five seizures per month
- Between one and five seizures per week
- Between one and five seizures per day
- More than ten seizures per day

What is the current seizure frequency at which the absences occurred?

- Less than five seizures per year
- Between five and ten seizures per year
- Between one and five seizures per month
- Between one and five seizures per week
- Between one and five seizures per day
- More than ten seizures per day

Are there protecting factors regarding absences?

Are/have there been factors that promote absences?

Note:

Generalized seizures

if yes:

What kind of generalized seizures occur?
(more than one applicable)

- Tonic-clonic
- Myoklonic
- Tonic
- Clonic
- Atonic

What was the patient's age at the onset of the generalized seizures?

- _____ month _____ years
- unknown

What age was the patient at the end (if present) of the generalized seizures?

- _____ month _____ years
- unknown

What is the maximum frequency generalized seizures occur?

- Less than five seizures per year
- Between five and ten seizures per year
- Between one and five seizures per month
- Between one and five seizures per week
- Between one and five seizures per day
- More than ten seizures per day

What is the current frequency generalized seizures occur?

- Less than five seizures per year
- Between five and ten seizures per year
- Between one and five seizures per month
- Between one and five seizures per week
- Between one and five seizures per day
- More than ten seizures per day

Are there/have there been protecting factors regarding absences?

Are there/have there been protecting factors regarding absences?

Note:

Focal seizures

if yes:

What kind of focal seizures occur?
(more than one applicable)

- With clouding / loss of consciousness
- With automatisms (involuntary motion sequences)
- With autonomous features (automatically running internal physical processes e.g. increasing heart rate)
- With speech abnormalities
- With motor abnormalities

What was the patient's age at the onset of the focal seizures?

- _____ month _____ years
- unknown

What was the patient's age at the end (if present) of the focal seizures?

- _____ month _____ years
- unknown

What is the maximum focal seizures frequency?

- Less than five seizures per year
- Between five and ten seizures per year
- Between one and five seizures per month
- Between one and five seizures per week
- Between one and five seizures per day
- More than ten seizures per day

What is the current focal seizures frequency?

- Less than five seizures per year
- Between five and ten seizures per year
- Between one and five seizures per month
- Between one and five seizures per week
- Between one and five seizures per day
- More than ten seizures per day

Are there/have there been protecting factors regarding focal seizures?

Are there/have there been promoting factors regarding focal seizures?

What autonomic features occur during focal seizures?
(more than one applicable)

- Fear
- Vomiting
- Wetting
- Defecating
- Sensation of cold
- Bradycardia (slowed heart rate)
- Flush
- Laughter
- Other

What other autonomic features occur during focal seizures?

What speech abnormalities occur during focal seizures?

- Speaking
- Aphasia (speech loss)
- Dysarthria (unintelligible speech)

What motor abnormalities occur during focal seizures?

- Clonic
- Tonic
- Dystone stiffening
- Hyperkinesia
- Head turning
- eye twisting / nystagmus (eye tremor)
- other

What was the patient's age when epileptic spasms began?

- _____ month _____ years
- unknown

What was the patient's age at the end of spasms (if present)?

- _____ month _____ years
- unknown

What is the epileptic spasms maximum seizure frequency?

- Less than five seizures per year
- Between five and ten seizures per year
- Between one and five seizures per month
- Between one and five seizures per week
- Between one and five seizures per day
- More than ten seizures per day

What is the current epileptic spasms seizure frequency?

- Less than five seizures per year
- Between five and ten seizures per year
- Between one and five seizures per month
- Between one and five seizures per week
- Between one and five seizures per day
- More than ten seizures per day

Are there/have there been protecting factors regarding spasms?

Are there/have there been any promoting factors regarding epileptic spasms?

Note:

Status epilepticus

if yes:

What type of status epilepticus occurs?

What type of status epilepticus occurs?

- Convulsive
- Non-convulsive

What age was the patient at the beginning of status epilepticus?

- _____ month _____ years
- unknown

What was the patient's age at the end (if present) of status epilepticus?

- _____ month _____ years
- unknown

What is the maximum status epileptici seizure frequency?

- Less than five seizures per year
- Between five and ten seizures per year
- Between one and five seizures per month
- Between one and five seizures per week
- Between one and five seizures per day
- More than ten seizures per day

What is the current status epileptici seizure frequency?

- Less than five seizures per year
- Between five and ten seizures per year
- Between one and five seizures per month
- Between one and five seizures per week
- Between one and five seizures per day
- More than ten seizures per day

Are there protecting factors regarding status epileptici?

Are there promoting factors regarding status epileptici?

Note:

Non-epileptic seizures

if yes:

Please explain the type/characteristics of non-epileptic seizures.

Other

if yes:

What other types of seizures occur? Please explain.

Are there postictal abnormalities (after the seizure)?

- yes
- no
- unknown

if yes:

What kind of postictal abnormalities occurred?

- Todd's paresis (temporary paresis)
- Hemianopia (loss of vision)
- Aphasia or dysphasia (speech loss/disorder)
- unknown
- Other

What other postictal abnormalities occurred?

Are/have there been seizure types with different stages?

(e.g. first a focal seizure, followed by a generalized seizure (secondary generalized seizures))

- yes
- no
- unknown

if yes:

What are/have been seizure forms with different stages?

- hypermotoric tonic-spasm sequence
- tonic seizures followed by series of spasms
- other

What other seizure types are/have there been with different stages?

Questions about medication

Unless otherwise stated, please tick the most appropriate answer.

Which of the following antiepileptic drugs were/are used?

ACTH (Synacthene®)

if yes:

What effect does the medicament have/had?

- good
- moderate
- unchanged
- deterioration of the seizure situation

To which type of seizure is/was the effect particularly related?

- febrile seizures
- absences
- generalized seizures
- focal seizures
- epileptic spasms (involuntary muscle tension)
- status epilepticus
- non-epileptic seizures
- other

What side effects occur/have occurred?

- none
- mild
- severe

What kind of side effects occur/have occurred?

Acetazolamid (Acemit®, Diamox®, Glaupax®)

if yes:

What effect does the medicament have/had?

- good
- moderate
- unchanged
- deterioration of the seizure situation

To which type of seizure is/was the effect particularly related?

- febrile seizures
- absences
- generalized seizures
- focal seizures
- epileptic spasms (involuntary muscle tension)
- status epilepticus
- non-epileptic seizures
- other

What side effects occur/have occurred?

- none
- mild
- severe

What kind of side effects occur/have occurred?

Bromid (Kaliumbromid®)

if yes:

What effect does the medicament have/had?

- good
- moderate
- unchanged
- deterioration of the seizure situation

To which type of seizure is/was the effect particularly related?

- febrile seizures
- absences
- generalized seizures
- focal seizures
- epileptic spasms (involuntary muscle tension)
- status epilepticus
- non-epileptic seizures
- other

What side effects occur/have occurred?

- none
- mild
- severe

What kind of side effects occur/have occurred?

Cannabis (Epidiolex®, CBD®)

What type of cannabis was used?

synthetic CBD

if yes:

What effect does the medicament have/had?

- good
- moderate
- unchanged
- deterioration of the seizure situation

To which type of seizure is/was the effect particularly related?

- febrile seizures
- absences
- generalized seizures
- focal seizures
- epileptic spasms (involuntary muscle tension)
- status epilepticus
- non-epileptic seizures
- other

What side effects occur/have occurred?

- none
- mild
- severe

What kind of side effects occur/have occurred?

natural CBD isolate

if yes:

What effect does the medicament have/had?

- good
- moderate
- unchanged
- deterioration of the seizure situation

To which type of seizure is/was the effect particularly related?

- febrile seizures
- absences
- generalized seizures
- focal seizures
- epileptic spasms (involuntary muscle tension)
- status epilepticus
- non-epileptic seizures
- other

What side effects occur/have occurred?

- none
- mild
- severe

What kind of side effects occur/have occurred?

CBD full extract

if yes:

What effect does the medicament have/had?

- good
- moderate
- unchanged
- deterioration of the seizure situation

To which type of seizure is/was the effect particularly related?

- febrile seizures
- absences
- generalized seizures
- focal seizures
- epileptic spasms (involuntary muscle tension)
- status epilepticus
- non-epileptic seizures
- other

What side effects occur/have occurred?

- none
- mild
- severe

What kind of side effects occur/have occurred?

THC

if yes:

What effect does the medicament have/had?

- good
- moderate
- unchanged
- deterioration of the seizure situation

To which type of seizure is/was the effect particularly related?

- febrile seizures
- absences
- generalized seizures
- focal seizures
- epileptic spasms (involuntary muscle tension)
- status epilepticus
- non-epileptic seizures
- other

What side effects occur/have occurred?

- none
- mild
- severe

What kind of side effects occur/have occurred?

Carbamazepine (Neurotop®, Tegretal®, Tegretol®, Timonil®)

if yes:

What effect does the medicament have/had?

- good
- moderate
- unchanged
- deterioration of the seizure situation

To which type of seizure is/was the effect particularly related?

- febrile seizures
- absences
- generalized seizures
- focal seizures
- epileptic spasms (involuntary muscle tension)
- status epilepticus
- non-epileptic seizures
- other

What side effects occur/have occurred?

- none
- mild
- severe

What kind of side effects occur/have occurred?

Clonazepam (Rivotril®, Antelepsin®)

if yes:

What effect does the medicament have/had?

- good
- moderate
- unchanged
- deterioration of the seizure situation

To which type of seizure is/was the effect particularly related?

- febrile seizures
- absences
- generalized seizures
- focal seizures
- epileptic spasms (involuntary muscle tension)
- status epilepticus
- non-epileptic seizures
- other

What side effects occur/have occurred?

- none
- mild
- severe

What kind of side effects occur/have occurred?

Diazepam (Valium®, Stesolid®)

if yes:

What effect does the medicament have/had?

- good
- moderate
- unchanged
- deterioration of the seizure situation

To which type of seizure is/was the effect particularly related?

- febrile seizures
- absences
- generalized seizures
- focal seizures
- epileptic spasms (involuntary muscle tension)
- status epilepticus
- non-epileptic seizures
- other

What side effects occur/have occurred?

- none
- mild
- severe

What kind of side effects occur/have occurred?

Ethosuximide (Petinimid®, Petnidan®)

if yes:

What effect does the medicament have/had?

- good
- moderate
- unchanged
- deterioration of the seizure situation

To which type of seizure is/was the effect particularly related?

- febrile seizures
- absences
- generalized seizures
- focal seizures
- epileptic spasms (involuntary muscle tension)
- status epilepticus
- non-epileptic seizures
- other

What side effects occur/have occurred?

- none
- mild
- severe

What kind of side effects occur/have occurred?

Felbamate (Taloxa®)

if yes:

What effect does the medicament have/had?

- good
- moderate
- unchanged
- deterioration of the seizure situation

To which type of seizure is/was the effect particularly related?

- febrile seizures
- absences
- generalized seizures
- focal seizures
- epileptic spasms (involuntary muscle tension)
- status epilepticus
- non-epileptic seizures
- other

What side effects occur/have occurred?

- none
- mild
- severe

What kind of side effects occur/have occurred?

Flunarizine (Sibelium®, Flunavert®)

if yes:

What effect does the medicament have/had?

- good
- moderate
- unchanged
- deterioration of the seizure situation

To which type of seizure is/was the effect particularly related?

- febrile seizures
- absences
- generalized seizures
- focal seizures
- epileptic spasms (involuntary muscle tension)
- status epilepticus
- non-epileptic seizures
- other

What side effects occur/have occurred?

- none
- mild
- severe

What kind of side effects occur/have occurred?

Gabapentine (Neurontin®, Gabagamma®, Gabatal®)

if yes:

What effect does the medicament have/had?

- good
- moderate
- unchanged
- deterioration of the seizure situation

To which type of seizure is/was the effect particularly related?

- febrile seizures
- absences
- generalized seizures
- focal seizures
- epileptic spasms (involuntary muscle tension)
- status epilepticus
- non-epileptic seizures
- other

What side effects occur/have occurred?

- none
- mild
- severe

What kind of side effects occur/have occurred?

Ketogenic diet

if yes:

What effect does the medicament have/had?

- good
- moderate
- unchanged
- deterioration of the seizure situation

To which type of seizure is/was the effect particularly related?

- febrile seizures
- absences
- generalized seizures
- focal seizures
- epileptic spasms (involuntary muscle tension)
- status epilepticus
- non-epileptic seizures
- other

What side effects occur/have occurred?

- none
- mild
- severe

What kind of side effects occur/have occurred?

Lacosamide (Vimpat®)

if yes:

What effect does the medicament have/had?

- good
- moderate
- unchanged
- deterioration of the seizure situation

To which type of seizure is/was the effect particularly related?

- febrile seizures
- absences
- generalized seizures
- focal seizures
- epileptic spasms (involuntary muscle tension)
- status epilepticus
- non-epileptic seizures
- other

What side effects occur/have occurred?

- none
- mild
- severe

What kind of side effects occur/have occurred?

Lamotrigine (Lamictal®, Lamotrigin Desitin®)

if yes:

What effect does the medicament have/had?

- good
- moderate
- unchanged
- deterioration of the seizure situation

To which type of seizure is/was the effect particularly related?

- febrile seizures
- absences
- generalized seizures
- focal seizures
- epileptic spasms (involuntary muscle tension)
- status epilepticus
- non-epileptic seizures
- other

What side effects occur/have occurred?

- none
- mild
- severe

What kind of side effects occur/have occurred?

Levetiracetam (Keppra®)

if yes:

What effect does the medicament have/had?

- good
- moderate
- unchanged
- deterioration of the seizure situation

To which type of seizure is/was the effect particularly related?

- febrile seizures
- absences
- generalized seizures
- focal seizures
- epileptic spasms (involuntary muscle tension)
- status epilepticus
- non-epileptic seizures
- other

What side effects occur/have occurred?

- none
- mild
- severe

What kind of side effects occur/have occurred?

Oxcarbazepine (Apydan Extent®, Timox®, Trileptal®)

if yes:

What effect does the medicament have/had?

- good
- moderate
- unchanged
- deterioration of the seizure situation

To which type of seizure is/was the effect particularly related?

- febrile seizures
- absences
- generalized seizures
- focal seizures
- epileptic spasms (involuntary muscle tension)
- status epilepticus
- non-epileptic seizures
- other

What side effects occur/have occurred?

- none
- mild
- severe

What kind of side effects occur/have occurred?

Perampanel (Fycompa®)

if yes:

What effect does the medicament have/had?

- good
- moderate
- unchanged
- deterioration of the seizure situation

To which type of seizure is/was the effect particularly related?

- febrile seizures
- absences
- generalized seizures
- focal seizures
- epileptic spasms (involuntary muscle tension)
- status epilepticus
- non-epileptic seizures
- other

What side effects occur/have occurred?

- none
- mild
- severe

What kind of side effects occur/have occurred?

Prednisone (Decortin®, Ultracorten®)

if yes:

What effect does the medicament have/had?

- good
- moderate
- unchanged
- deterioration of the seizure situation

To which type of seizure is/was the effect particularly related?

- febrile seizures
- absences
- generalized seizures
- focal seizures
- epileptic spasms (involuntary muscle tension)
- status epilepticus
- non-epileptic seizures
- other

What side effects occur/have occurred?

- none
- mild
- severe

What kind of side effects occur/have occurred?

Phenobarbital (Luminal®)

if yes:

What effect does the medicament have/had?

- good
- moderate
- unchanged
- deterioration of the seizure situation

To which type of seizure is/was the effect particularly related?

- febrile seizures
- absences
- generalized seizures
- focal seizures
- epileptic spasms (involuntary muscle tension)
- status epilepticus
- non-epileptic seizures
- other

What side effects occur/have occurred?

- none
- mild
- severe

What kind of side effects occur/have occurred?

Phenytoine (Phenhydantoin®, Zentropil®)

if yes:

What effect does the medication have/had?

- good
- moderate
- unchanged
- deterioration of the seizure situation

To which type of seizure is/was the effect particularly related?

- febrile seizures
- absences
- generalized seizures
- focal seizures
- epileptic spasms (involuntary muscle tension)
- status epilepticus
- non-epileptic seizures
- other

What side effects occur/have occurred?

- none
- mild
- severe

What kind of side effects occur/have occurred?

Rufinamide (Inovelon®)

if yes:

What effect does the medication have/had?

- good
- moderate
- unchanged
- deterioration of the seizure situation

To which type of seizure is/was the effect particularly related?

- febrile seizures
- absences
- generalized seizures
- focal seizures
- epileptic spasms (involuntary muscle tension)
- status epilepticus
- non-epileptic seizures
- other

What side effects occur/have occurred?

- none
- mild
- severe

What kind of side effects occur/have occurred?

Topiramate (Topamax®)

if yes:

What effect does the medicament have/had?

- good
- moderate
- unchanged
- deterioration of the seizure situation

To which type of seizure is/was the effect particularly related?

- febrile seizures
- absences
- generalized seizures
- focal seizures
- epileptic spasms (involuntary muscle tension)
- status epilepticus
- non-epileptic seizures
- other

What side effects occur/have occurred?

- none
- mild
- severe

What kind of side effects occur/have occurred?

Valproic acid (Orfiril®, Valproat®, Depakine®, Convulex®)

if yes:

What effect does the medicament have/had?

- good
- moderate
- unchanged
- deterioration of the seizure situation

To which type of seizure is/was the effect particularly related?

- febrile seizures
- absences
- generalized seizures
- focal seizures
- epileptic spasms (involuntary muscle tension)
- status epilepticus
- non-epileptic seizures
- other

What side effects occur/have occurred?

- none
- mild
- severe

What kind of side effects occur/have occurred?

Vigabatrine (Sabril®)

if yes:

What effect does the medicament have/had?

- good
- moderate
- unchanged
- deterioration of the seizure situation

To which type of seizure is/was the effect particularly related?

- febrile seizures
- absences
- generalized seizures
- focal seizures
- epileptic spasms (involuntary muscle tension)
- status epilepticus
- non-epileptic seizures
- other

What side effects occur/have occurred?

- none
- mild
- severe

What kind of side effects occur/have occurred?

Zonisamide (Zonegran®)

if yes:

What effect does the medicament have/had?

- good
- moderate
- unchanged
- deterioration of the seizure situation

To which type of seizure is/was the effect particularly related?

- febrile seizures
- absences
- generalized seizures
- focal seizures
- epileptic spasms (involuntary muscle tension)
- status epilepticus
- non-epileptic seizures
- other

What side effects occur/have occurred?

- none
- mild
- severe

What kind of side effects occur/have occurred?

Clobazame (Frisium®)

if yes:

What effect does the medicament have/had?

- good
- moderate
- unchanged
- deterioration of the seizure situation

To which type of seizure is/was the effect particularly related?

- febrile seizures
- absences
- generalized seizures
- focal seizures
- epileptic spasms (involuntary muscle tension)
- status epilepticus
- non-epileptic seizures
- other

What side effects occur/have occurred?

- none
- mild
- severe

What kind of side effects occur/have occurred?

Other

if yes:

What other antiepileptic drugs are/have been used?

Other medication

Unless otherwise stated, please tick the most appropriate answer.

Have any other drugs been/are used that are not related to epilepsy?

(e.g. to treat movement disorders or infections)

- yes
- no
- unknown

if yes:

Name of the drug 1

Application purpose

Effect

- good
- moderate
- unchanged
- deterioration of the situation

Comment

Name of the drug 2

Application purpose

Effect

- good
- moderate
- unchanged
- deterioration of the situation

Comment

Name of the drug 3

Application purpose

Effect

- good
- moderate
- unchanged
- deterioration of the situation

Comment

Name of the drug 4

Application purpose _____

- Effect
- good
 - moderate
 - unchanged
 - deterioration of the situation

Comment _____

Name of the drug 5 _____

Application purpose _____

- Effect
- good
 - moderate
 - unchanged
 - deterioration of the situation

Comment _____

Questions about physical characteristics

Unless otherwise stated, please tick the most appropriate answer.

What is the patient's eye color?

- blue
- greyish blue
- green
- brown
- grey
- unclear

What color is the patient's hair color?

- light blond
- dark blond
- black
- brown
- natural red

Is the patient's hair, skin and/or eye color lighter than it would be expected in the family?

- yes
- no
- unknown

Questions about puberty

Unless otherwise stated, please tick the most appropriate answer.

Does the puberty started or happend?

- yes
- no
- unknown

Puberty started:

- too early (female before age 8; male before age 9)
- age appropriate (female age 8-13; male age 9-14)
- delayed (female age > 13; male age > 14)

When did the girl's breast development began?

- not yet
- age: _____ years
- unknown

Questions about diagnostics

Unless otherwise stated, please tick the most appropriate answer.

Has the patient ever had a cranial MRI?
 yes
 no
 unknown

Was the MRI abnormal?
 yes
 no
 unknown

Does the Institute of Human Genetics at Leipzig University Hospital received a copy of the MRI report?
 yes
 no
 unknown

You can send us a copy of the findings by e-mail or regular mail, should they be available digitally for the study.

Please make personal information, e.g. names, illegible.

What was abnormal about the MRI result?
(Please describe the abnormalities)

Other information/abnormalities

Unless otherwise stated, please tick the most appropriate answer.

Are there any sudden or temporary vegetative symptoms?
(automatically occurring inner-body processes)

sudden redness of the skin / face ("flush")
 increased
 decreased
 unknown

excessive sweating
 increased
 decreased
 unknown

increased / decreased heart rate
 increased
 decreased
 unknown

Other (please describe):

 increased
 decreased
 unknown

Other (please describe):

- increased
- decreased
- unknown

Other (please describe):

- increased
- decreased
- unknown

Is there a visual impairment?

- yes
- no
- unknown

if yes:

What kind of visual impairment?

- myopia (near-sightedness)
- hyperopia (long-sightedness)
- strabismus (squint)
- cortical visual impairment (CVI)/cortical blindness
- other

What kind of other visual impairment is present?

Is there a hearing impairment/deafness?

- yes
- no
- unknown

Are there frequent (several times a year) infections?

- yes
- no
- unknown

Does an immunodeficiency was diagnosed?

- yes
- no
- unknown

Does the patient has a psychiatric disorder?

- no
- schizophrenia
- autism
- other

