GRIN Registry

Institute of Human Genetics

University of Leipzig Medical Center

Languages: Deutsch, English

Study information

In this study, we want to investigate clinical and genetic findings of GRIN-related disorders and collect data of affected individuals in a local 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, pause it at any time and continue later on by using a login and ID. It is possible to enter data directly, but also to upload documents (e.g. molecular genetic findings, electroencephalography, magnetic resonance imaging, clinical reports, etc.), which will be reviewed by a member of our team and transferred pseudonymously into the registry. The study has been approved by the Ethics Committee of the University of Leipzig, Germany. The aims of the study are to establish a registry on GRIN-related disorders, to better understand genotype-phenotype correlations and finally to improve patient care. This online registry is a collaborative effort with our close collaborators Stephen Traynelis (Emory University, Atlanta), Tim Benke (University of Colorado) and CureGRIN.

Patient Information *must provide value

Last name of patient: _____

First name of patient: _____

Contact Person *must provide value

Last name of contact person: _____

First name of contact person: _____

E-Mail of contact person: _____

Please retype E-Mail of contact person (to avoid typing errors):

_____

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).

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 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.

yes

*must provide value

We agree that the clinical and genetic data of our child or guardian may be entered and used in pseudonymized form in the study database "Genotype-phenotype correlations in GRIN disorders" and potentially published.

yes

*must provide value

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

yes

no

*must provide value

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

yes

no

*must provide value

Please enter an alternative e-mail address:

*must provide value

_____

General Information

Data was edited by:	- Parent / Relative / Legal guardian - referring clinician - GRIN-Team Leipzig - other
Who of the GRIN-Team Leipzig?	
Last name of clinician:	_____
First name of clinician:	_____
Please describe who you are:	_____
Data was assessed on:	- Present day - other
Please enter the date of assessment:	dd.mm.yyyy
Patient sex:	- female - male - unknown
Nationality of patient	_____
Date of birth of patient	dd.mm.yyyy
Age of patient	_____ years _____ months
Is the patient still alive?	- yes - no - not assessed /available
Please state date of death:	dd.mm.yyyy

Medical Information

Gene	- GRIN1; NM_007327.3 - GRIN2A, NM_001134407.3 - GRIN2B; NM_000834.4 - GRIN2D; NM_000826.2
Variant DNA change for example: c.1666C>T	_____
Variant protein change for example: p.(Arg217Trp)	_____
Does the patient have a second variant in the same gene? (In case you suspect a recessive inheritance.)	- yes - no
Variant 2 DNA change for example: c.1666C>T	_____
Variant 2 protein change for example: p.(Gln556*)	_____
Variant origin	- de novo - homozygous - compound heterozygous - maternally inherited - paternally inherited - unknown
Further (other than GRIN) genetic findings? for example: pathogenic variant in BRCA1	_____
What was the method of genetic testing?	- Exome-sequencing (trio) - Exome-sequencing (patient) + sanger sequencing of parents - Exome-sequencing (patient only) - Panel (patient) + sanger sequencing of parents - Panel (patient only) - Other
Please describe "other":	_____
Do you know if your child/patient is published?	- yes - no - unknown

In case the data is at hand, please enter any details on publication.	- Year - Journal - Last name of first author - First name of first author - PMID (PubMed-ID) - Patient-ID in Publication

Pregnancy & Birth

Was the prenatal period normal?	- yes - no - not assessed / available
What was abnormal?	_____
Gestational week at birth for example: 41+2	_____+_____
Length at birth (+percentile)	_____ cm
Weight at birth (+percentile)	_____ kg
Occipital Frontal Circumference at birth (+ percentile)	_____ cm
Age at last assessment	_____ years _____ months _____ days
Lenght at last assessment (+ percentile)	_____ cm
Weight at last assessment (+ percentile)	_____ kg
Occipital Frontal Circumference at last assessment (+ percentile)	_____ cm

Family

Parental cognitive level (please make an educated guess):	- above average - average - below average - unknown / not available
Parental consanguinity (parents cousin/cousine)	- yes - no

	- not assessed / available
Other affected family members?	- yes
	- no
	- not assessed / available
Affected Family Members Information (Please describe the relationship of the other affected family members.)	_____ _____

Development

Global developmental delay	- yes
	- no
	- not assessed / available
Severity of developmental delay	- mild
	- moderate
	- severe
	- profound
	- not assessed / available
Head control	- yes
	- no
	- not assessed / available
Head control learned at age	_____ years _____ months
Crawling	- yes
	- no
	- not assessed / available
Crawling learned at age	_____ years _____ months
Sitting	- yes
	- no
	- not assessed / available
Unassisted Sitting learned at age	_____ years _____ months
Walking	- yes
	- no
	- not assessed / available
Unassisted walking learned at age	

	_____ years _____ months
Persistent motor problems during childhood	- yes
	- no
	- not assessed / available
Speech at last exam	- no speech
	- single words
	- sentences
	- not assessed / available
Age of first words	
	_____ years _____ months
Intellecual disability	- yes
	- no
	- not assessed / available
Severity of Intellectual disability	- mild
	- moderate
	- severe
	- profound
	- not assessed / available
IQ (if tested)	
	_____
Dysmorphic facial features	- yes
	- no
	- not assessed / available
Dysmorphic facial features description	
e.g. epicanthus, broad bridge of nose, high forehead etc.	_____
Autism spectrum disorder (ASD)	- yes
	- no
	- not assessed / available
Stagnation/Regression	- yes
(loss of an acquired function)	- no
	- not assessed / available
Stagnation/Regression Description	
(e.g. stagnation and/or regression?, at what age stagnation/regression started?, motor and/or language regression ...)	_____
	_____
	_____

Additional symptoms

Sleeping problems	- yes - no - not assessed / available
Sleeping problems description (e.g. difficulty falling asleep or sleeping through the night)	_____
Behavioral abnormalities	- yes - no - not assessed / available
Behavioral abnormalities description (e.g. stereotypic, aggressive, hyperactive...)	_____
Sensation of pain	- normal - abnormal - not assessed / available
Sensation of pain description (e.g. hyper, hypo, no pain sensation...)	_____
Hypotonia	- yes - no - not assessed / available
Hypotonia description	_____
Spasticity	- yes - no - not assessed / available
Spasticity description	_____
Feeding difficulties in early childhood	- yes - no - not assessed / available
Feeding difficulties description (e.g. due to hypotonia...)	_____
Other gastro-intestinal abnormalities	- yes - no - not assessed / available
Gastro-intestinal abnormalities description (e.g. constipation...)	_____

Movement discorder	- yes - no - not assessed / available
Movement discorder description (e.g. dystonia, dyskinesia, chorea, involuntary orofacial movements...)	_____
Ataxia	- yes - no - not assessed / available
Ataxia description (e.g. at what age...)	_____
Oculogyric crisis (prolonged involuntary upward deviation of the eyes):	- yes - no - not assessed / available
"Neurological Storms" We have been made aware of so called "neurological storms". These are described as paroxysmal sympathetic hyperactivity like agitation, unusually high seizure frequency in short period of time or hyperkinetic movement disorders. Have you seen this in your child/patient?	- yes - no - not assessed / available
"Neurological Storms" description (e.g. how it looks like, how often...)	_____
Other neurological phenotype	- yes - no - not assessed / available
Other neurological phenotype description (e.g. hyperreflexia...)	_____
Scoliosis	- yes - no - not assessed / available
Hearing loss	- yes - no - not assessed / available
Hearing loss description	_____

Cerebral visual impairment / cortical blindness	- yes - no - not assessed / available
Cerebral visual impairment / cortical blindness description	_____
Other ophthalmological abnormalities	- yes - no - not assessed / available
Other ophthalmological abnormalities description	_____

Epilepsy

Seizures	- yes - no - not assessed / available
Age at seizures onset	_____ years _____ months _____ days
Seizure type at onset	- Generalized Seizure - Focal Seizure - Epileptic Spasms - Febrile Seizure - Absence Seizure - Status Epilepticus
Generalized seizure type	- Tonic-clonic - Myoclonic - Tonic - Clonic - Atonic
Focal seizure type	- Loss of conciousness - Without loss of conciousness
Status Epilepticus seizure type	- Convulsive - Non-Convulsive
Seizure frequency at onset	_____ per day / week / year
Further seizure types	- Generalized Seizure - Focal Seizure - Epileptic Spasms - Febrile Seizure

	- Absence Seizure
	- Status Epilepticus
	- none
Further generalized seizure type (6x)	- Tonic-clonic
	- Myoclonic
	- Tonic
	- Clonic
	- Atonic
Further focal seizure type	- Loss of conciousness
	- Without loss of conciousness
Further Status Epilepticus seizure type	- Convulsive
	- Non-Convulsive
Further seizure frequency at onset	_____ per day / week / year
AED (Antiepileptic-drug) previously used Please write down every AED ever used (Valproat - VPA, Sultiame - STM, Levetiracetame - LEV...)	_____
Application of NMDA-receptor modifying drugs (e.g. memantine, dextrometorphan, L-serine, etc.)	- yes - no - not assessed / available
Application of NMDA-receptor modifying drugs description (Please mention drug (memantine, dextrometorphan, L-serine, etc.) and describe response.)	_____
Currently used AED	_____
AED response (Please describe response to certain AED (for example good response to Valproate; no response to Levetiracetame…)	_____
Seizure outcome (Please elaborate: persistant or temporary seizure freedom? Duration of seizure freedom? Duration of longest seizure freedom? Refractory seizures?)	_____
EEG at onset	- normal

	- abnormal
	- not assessed / available
EEG at onset description	
(Please describe EEG phenotype in detail.)	_____
EEG at follow up	- normal
	- abnormal
	- not assessed / available
EEG at follow up description	
	_____
Last EEG	- normal
	- abnormal
	- not assessed / available
Last EEG description	
	_____
Additional Seizure / Epilepsy description	
	_____

MRI

Brain MRI result	- normal
	- abnormal
	- not assessed / available
Age at MRI investigation (normal)?	
	_____ years _____ months
Age at MRI investigation (abnormal)?	
	_____ years _____ months
Brain MRI result: MCD (malformation of cortical development): diffuse bilateral polymicrogyria	- yes - no - not assessed / available
MCD description (please describe: e.g. frontal, persylvian, parietal, temporal)	_____
Brain MRI result: leukoencephalopathy	- yes
	- no
	- not assessed / available
Leukoencephalopathy description (Please describe if possible.)	_____
Brain MRI result: cerebral atrophy	- yes

	- no - not assessed / available
Cerebral atrophy description (Please describe if possible.)	_____
Brain MRI result: reduced white matter volume	- yes - no - not assessed / available
Reduced white matter volume description (Please describe if possible.)	_____
Brain MRI result: thinning of the corpus callosum	- yes - no - not assessed / available
Thinning of the corpus callosum description (Please describe if possible.)	_____
Brain MRI result: abnormal hippocampi	- yes - no - not assessed / available
Abnormal hippocampi description (Please describe if possible.)	_____
Brain MRI result: enlarged lateral ventricles	- yes - no - not assessed / available
Enlarged lateral ventricles description (Please describe if possible.)	_____
Brain MRI result: increased extra-axial spaces	- yes - no - not assessed / available
Increased extra-axial spaces description (Please describe if possible.)	_____
Other brain MRI results: (Please describe other abnormalities or copy the full report if possible.)	_____

Additional Information

Laboratory abnormalities	- yes - no

	- not assessed / available
Laboratory abnormalities description (e.g. high lactate in serum / liquor; low potassium...)	_____
Additional information	- ja - nein - nicht erhoben / verfügbar
Additional information description (e.g. congenital malformations...)	_____