

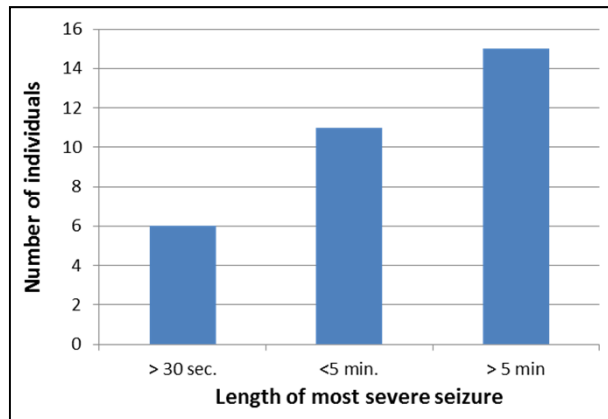
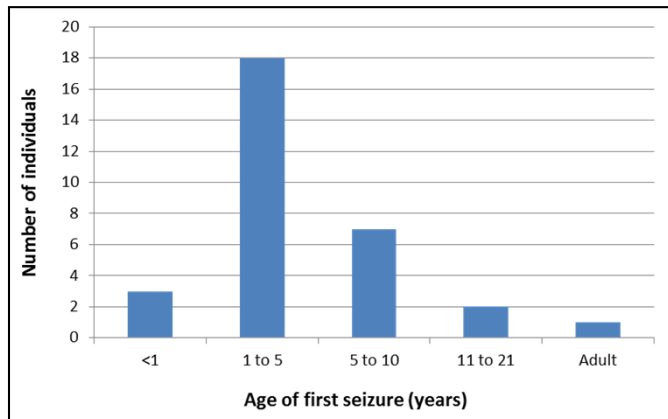
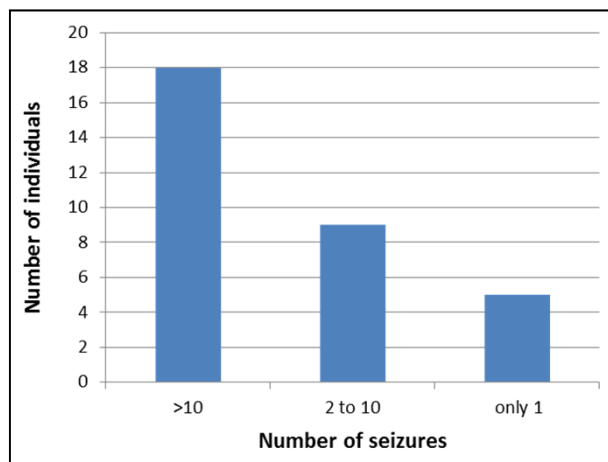
Self-Report Seizure Survey Summary 2017

Tetrasomy 18p

61 responses

33 had at least one seizure = 54%

8 had a seizure in the last year



Medications that work:

Valproate (Depakote)	7
Lamotrigene (Lamictal)	4
Levetiracetam (Keppra)	2
Carbamazepine (Tegretol)	1
Polypharmacy	4

Medications that did not work:

Valproate (Depakote, Epilium)	2
Levetiracetam (Keppra)	2
Zonisamide (Zonegran)	2
Lacosimide (Vimpat)	2
Clobazam (Frisium)	1
Phenobarbital (Luminaletten)	1
Ethosuximide (Zarontin)	1
Rufinamide (Banzel)	1
Amantadine	1
Nitrazepam (Mogadon)	1

Triggers:

Illness/ fever	20
Lack of sleep	6
Flashing lights	5
Medication	2
No apparent trigger	14

18q+

4 responses

2 had seizures with no apparent triggers

Both had more than 10 seizures and both had a seizure in the last year

Duration of the longest seizure varied ; one individual had a seizure lasting more than 5 minutes.

Medications that work:

Levetiracetam (Keppra) 1

Ketogenic Diet +Lacosamide + Lamotrigine + Divalproex Sodium (Depakote) 1

Medications that did not work:

Clobazam (Onfi) 1

Zonisamide, Pyridoxine, Valproic Acid, ACTH therapy, Vigabatrin, Topiramate 1

Ring 18

22 responses

6 had at least 1 seizure = 27%

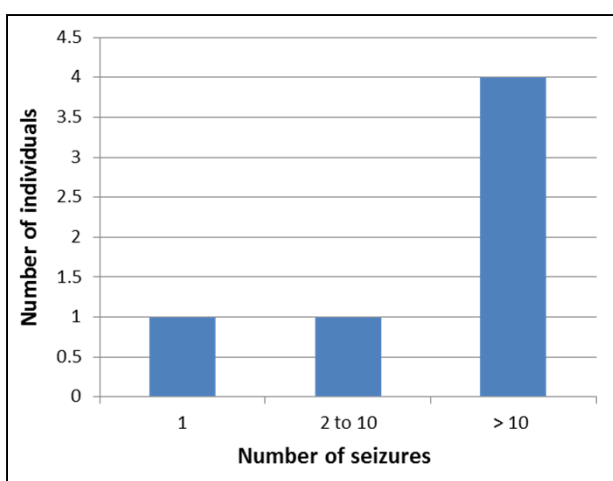
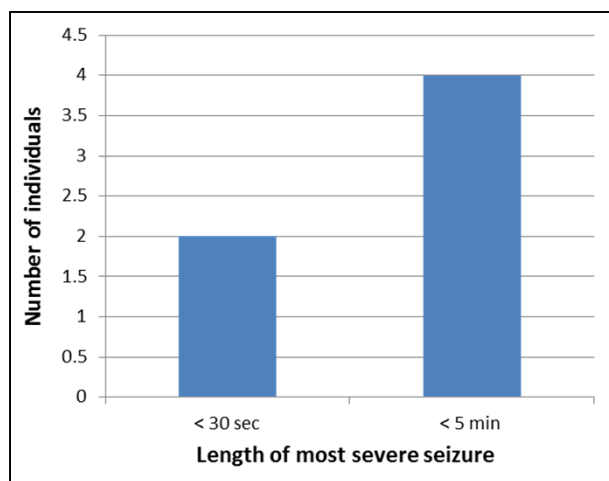
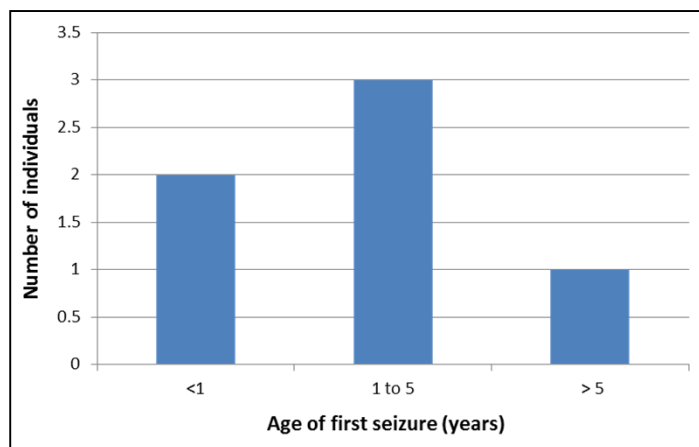
1 had a seizure in the last year

Triggers:

Fever / illness 3

Lack of sleep 2

No apparent trigger 4



Medications that work:

Levetiracetam (Keppra) 2

Topimarate (Topamax) 2

Medications that did not work:

Vigabatrin (Sabil) 1

Trisomy 18

(Some respondents were mosaic or had partial Trisomy 18)

19 responses:

All over 6 years of age

7 had seizures = 37%

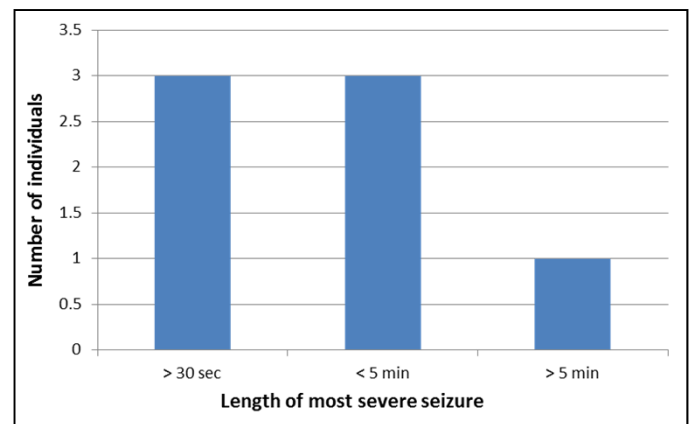
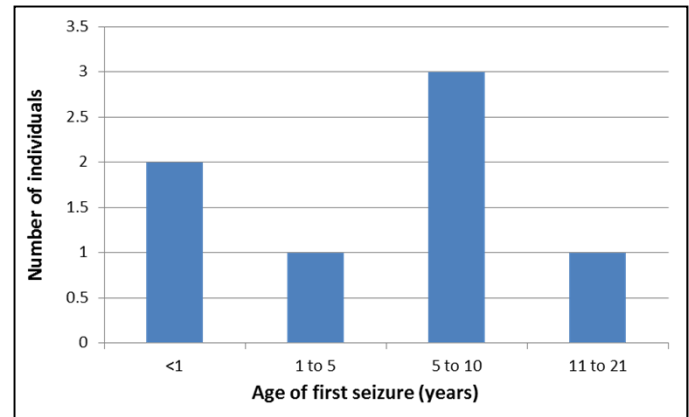
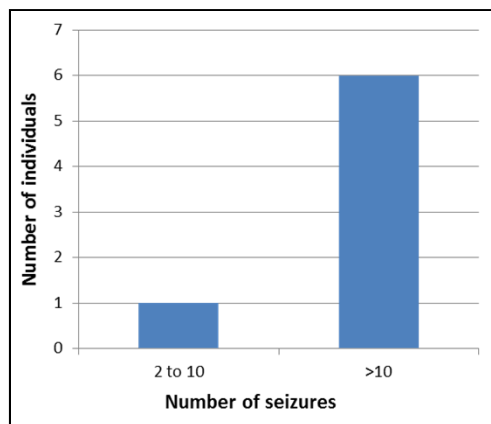
5 had a seizure in the last year

Triggers:

No apparent trigger 4

Fever / illness 2

Lack of sleep 2



Medications that work:

Felbamate (Felbatol) 1

Lamotrigene (Lamictal) 1

Levetiracetam (Keppra) 1

Depakote + Topamax 1

Lamictal + Rufinamide (Banzel) 1

Medications that did not work:

Clobazam (Onfi) 2

Levetiracetam (Keppra) 1

Divalproex Sodium (Depakote) 1

18p+

3 responses

One had one seizure lasting <5 minutes at 2 years of age; this individual is not currently on any seizure medication.

18p-

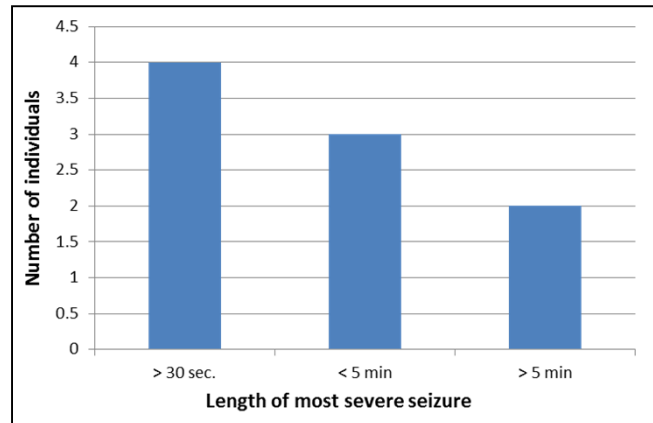
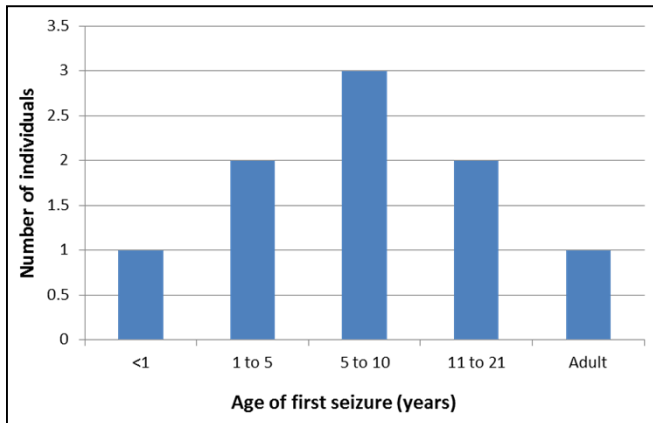
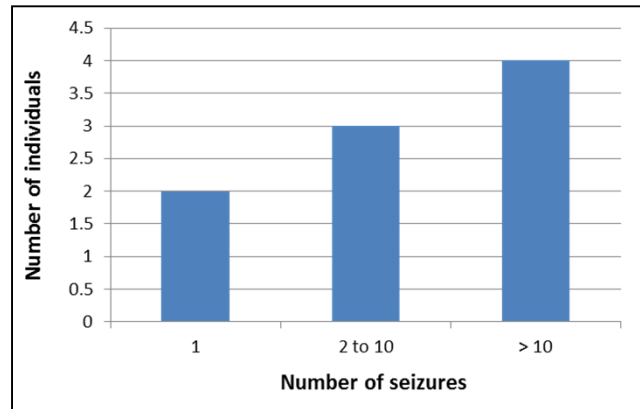
91 responses

9 had at least 1 seizure = 10%

4 had a seizure within the last year

Triggers:

Fever/illness	5
No apparent trigger	5
Lack of sleep	3
Flashing lights	1



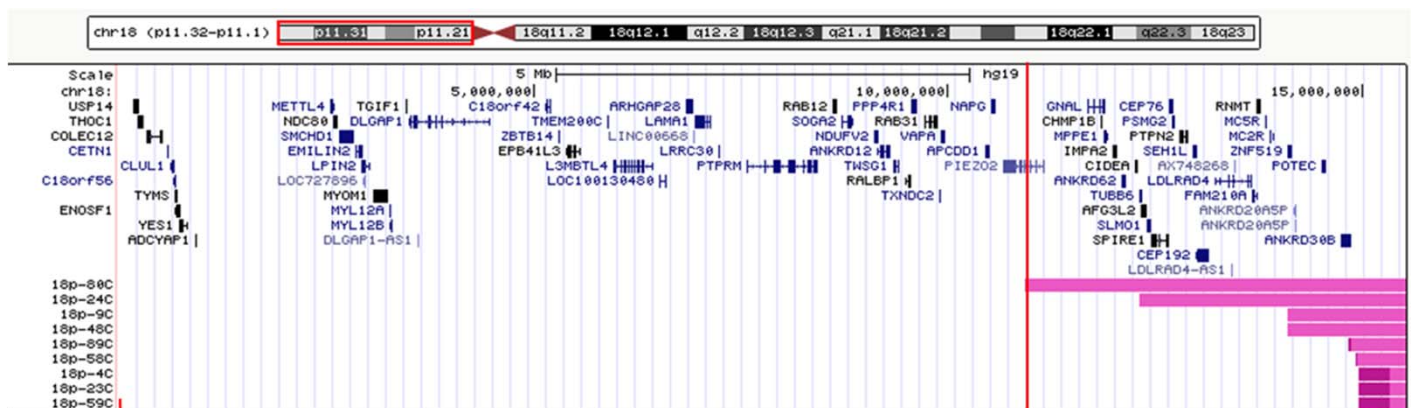
Medications that work:

Topamax + Keppra	1
Trileptal + Depakote	1
Depakote + Lamictal	1
Gabapentin + Lamictal	1
Not on meds	5

Medications that did not work:

Topimarate (Topamax)	1
Levetiracetam (Keppra)	2
Lamotrigine (Lamictal)	1
Rufinamide (Banzel)	1

The percent with seizures is not significantly different between those with a deletion of the whole p arm and those with smaller deletions. The region of 18p that is missing in everyone with 18p- and seizures is shown below.



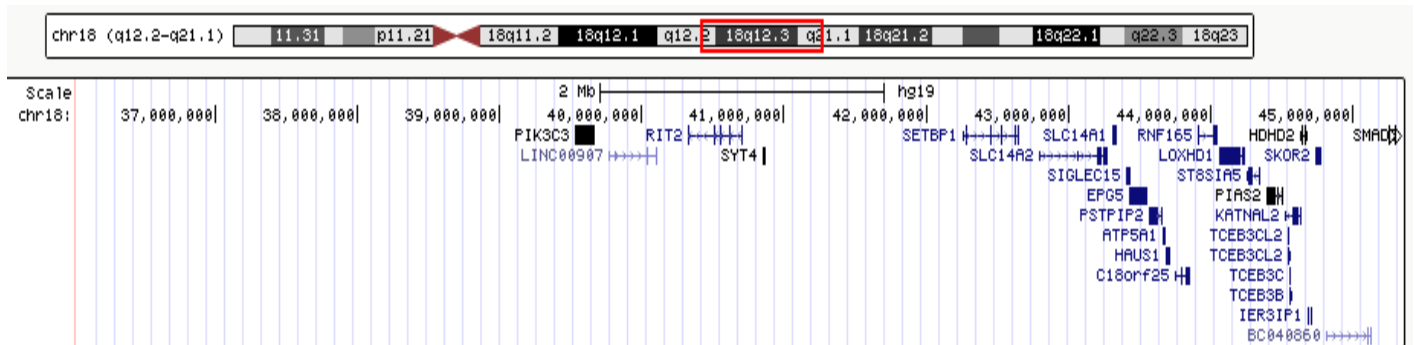
Seizure critical region
1- 10,942,107

Proximal 18q- (an interstitial deletion of 18q between 21 Mb and 47Mb)

7 responses

2 who have had a seizure

- 1 had 1 seizure at age 3 that lasted less than 5 minutes and is not taking seizure medication
- 1 has had more than 10 seizures the longest one lasting more than 5 minutes. Medications included Lamictal and later treated with ACTH and has not had a seizure in the last year. The medication that did not work was Valproate (Depakote).
- Although this family did not participate in the survey, one study participant with proximal 18q- died at age 11 due to seizure complications.
- The region of the chromosome that all 3 children had missing is shown below.



Distal 18q- (deletions that include TCF4)

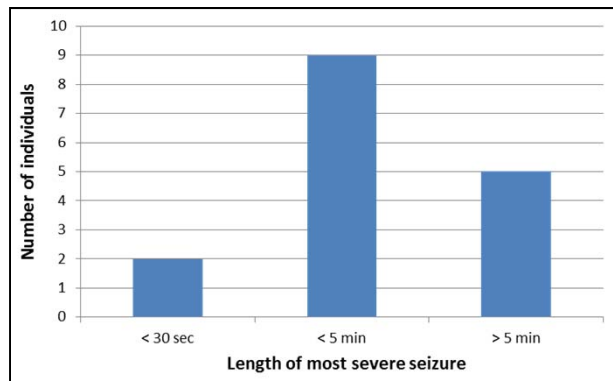
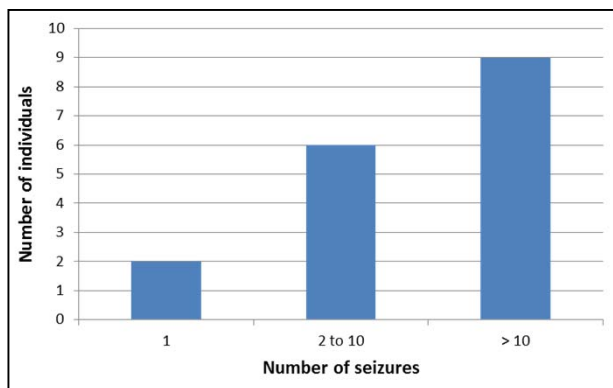
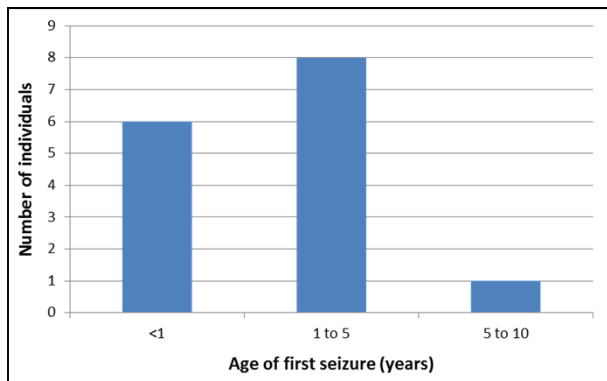
27 responses

16 have had a seizure = 60%

7 have had a seizure in the last year

Triggers:

Fever / illness	6
Lack of sleep	4
No apparent trigger	9



Medications that worked:

Levetiracetam (Keppra)	2
Carbamazepine (Tegretol)	2
Keppra + Valproate (Epilium)	1
Phenobarbital	1
Valproate (Epilium)	1
Keppra + Clobazam (Onfi)	1
Lamotrigene (Lamictal) + Rufinamide (Banzel) + Onfi	1
Keppra + Zonisamide (Zonegran)	1

Medications that did not work:

Carbamazepine (Tegretol)	2
Topiramate (Topamax)	2
Phenytoin (Dilantin)	1
Valproate (Depakote)	1
Levetiracetam (Keppra)	1
Lorazepam (Atavan)	1

TCF4 Mutations (and no deletion of any genes on 18q)

11 responses

3 have had a seizure= 27%

1 had a seizure in the last year

The most severe seizure was longer than 5 minutes

The first seizure was at age 15 months and 4 years

One child had between 2 and 10 seizures and another had more than 10

Triggers were fever/ Illness, a lack of sleep or no apparent trigger

Medications that worked:

Topiramate (Topamax) + Clobazam (Onfi)	1
Levetiracetam (Keppra)	1

Distal 18q- (deletions that do not include the TCF4 gene)

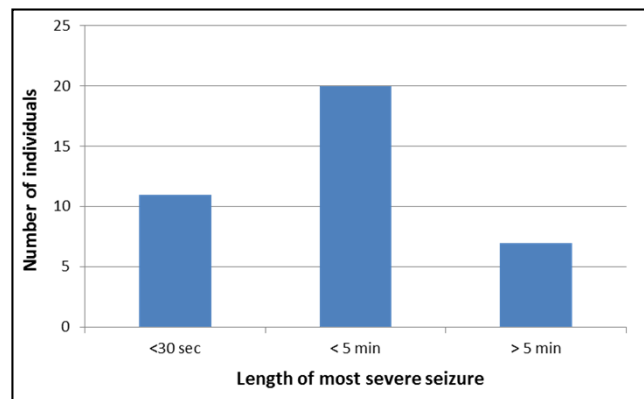
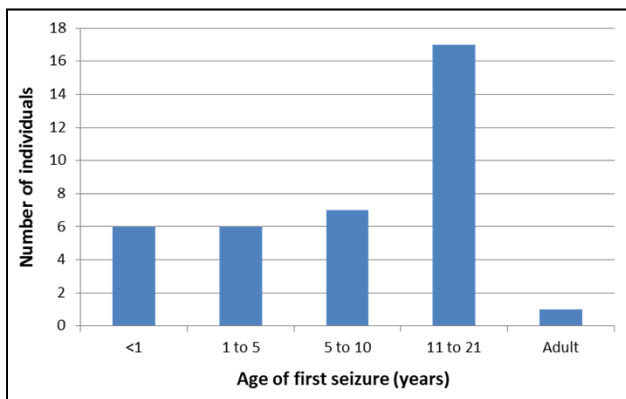
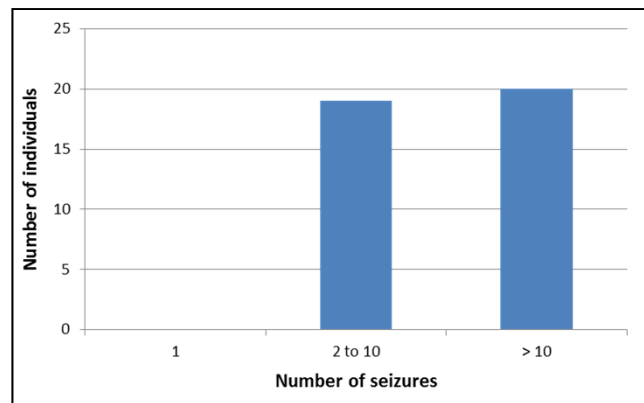
110 responses

40 had seizures = 36%

10 have had a seizure in the last year

Triggers:

Fever / illness	7
Lack of sleep	9
No apparent trigger	29



Medications that work:

Oxcarbazepine (Trileptal)	5
Lamotrigene (Lamictal)	3
Carbamazepine (Tegretol)	3
Valproate (Depakote, Epilim)	3
Levetiracetam (Keppra)	2
Gabapentin	1
Topimarat (Topomax)	1
Zonisamide(Zonegran)	1
Phenytoin (Dilantin)	1
Valproate (Depakote) +Lamictal	1
Valproic acid +Trileptal + Keppra	1
Lactimal + Zonisamide(Zonegran)	1
Lactimal + Topomax	1
Trileptal + Keppra	1
Never been on seizure meds	11

Medications that did not work:

Levetiracetam (Keppra)	4
Carbamazepine (Tegretol)	3
Topimarat (Topomax)	3
Lamotrigene (Lamictal)	2
Valproate (Depakote)	2
Lacosamide (Vimpat)	1
Phenobarbital	1
Lorazepam (Ativan)	1
Oxcarbazepine (Trileptal)	1
Clobazam (Onfi)	1