Seizures and Epilepsy

Introduction: Neurons and Electrical Pulses

Neurons (nerve cells) use electrical pulses when they transmit messages to other neurons and to target cells. Normally, these pulses are produced and travel along in a controlled manner. **Seizures** and **epilepsy** (repeated seizures due to the brain being prone to having abnormal electrical pulses) happen when these electrical pulses are out of control.

What Happens During Seizures

- **Seizures** happen when there are **sudden, uncontrolled bursts of electrical energy** between the cells in one or more areas of the brain.
- When people have seizures, they may:
  - Lose consciousness
  - Have uncontrolled muscle movements, jerking, and increased or loss of muscle tone
  - Stare into space
  - Have changes in hearing, vision, taste, smell
  - Drool or foam at the mouth
  - Have trouble talking and understanding
  - Experience confusion
  - Feel numbness or tingling
  - Feel hot or cold
  - Feel scared or dread
  - Have faster heart rate and breathing

Seizures and 22q Differences

22q11.2 deletion syndrome (22q11.2DS)

- Unprovoked seizures affect up to ~15% of children with 22q11.2DS.
- ~16% of adults with 22q11.2DS experienced seizures. Over 70% of these seizures have known triggers.
- The risk of epilepsy is 4x greater in adults with 22q11.2DS than the general public.

22q11.2 duplication syndrome (22q11.2DupS)

- In a study in Belgium, epilepsy was found in 3/26 (12%) of children with 22q11.2DupS.

Provoked Seizures and 22q11.2DS

- Seizures can be "**provoked**" by many triggers.
- **Keeping a record of the events** leading up to each seizure can help identify the possible triggers.
- Common triggers of seizures in people with 22q11.2DS:
  - **Hypocalcemia** – Not enough calcium in the blood
  - **Hypomagnesemia** – Not enough magnesium in the blood
  - Recent **surgeries**
  - **Fever** and **infections**
  - **Brain abnormalities** (various types)
  - **Antipsychotic medications** – If **Clozapine** is needed to treat schizophrenia, ask your doctor regarding the right dose for you. Anti-seizure medications may be added for people who have had seizures before.

Unprovoked Seizures and 22q11.2DS

- A seizure is said to be "**unprovoked**" if medical professionals cannot find a clear trigger
- Unprovoked seizures have been reported in both children and adults with 22q11.2DS
- After having 1 unprovoked seizure, people may or may not have additional seizures
- Seek medical advice after an unprovoked seizure

Types of seizures

Seizures (whether provoked or not) are classified based on where it **starts** in the brain, level of **awareness**, and the types of movements one has during an event (if present). The types that are common in people with 22q11.2DS are in **red**. For more info, see **Epilepsy** (Cleveland Clinic).

Focal (partial onset) seizures start in one area on one side of the brain. 2 types:

- **Focal aware seizures** – seizures occur, but the person stays conscious and knows what is happening
- **Focal impaired awareness seizure** – seizures occur; the person is unconscious and unaware of what is happening

Generalized seizures affect a wide area of cells on both sides of the brain. 6 types:

- **Absence seizures** – blank stare
- **Atonic seizures** – loss of muscle tone + lose consciousness + fall + bite tongue + lose control of bowel and bladder
- **Tonic seizures** – stiffening of a part of, or the entire body + lose consciousness + fall + bite tongue + lose control of bowel and bladder
- **Clonic seizures** – jerking of part of / entire body
- **Tonic-clonic seizures** (convulsions) – stiff muscles + repeated quick jerking +/- lose consciousness + fall + bite tongue + lose control of bowel and bladder
- **Myoclonic seizures** – brief, lightning-like jerks

Seizures and Epilepsy (continued)

Epilepsy

- **Epilepsy** is a long-term condition that causes **repeated seizures**.
- A person is diagnosed with epilepsy if he/she had:
  - At least 2 unprovoked seizures >1 day apart; OR
  - 1 unprovoked seizure + a known condition that makes the brain prone to develop seizures, or if first seizure happened during sleep
- The cause of epilepsy may not be known. Known causes include brain structural differences or issues, head injuries, brain infections, immune disorders etc.

Diagnosing Epilepsy

- Your healthcare provider will take your medical history
- He/she may ask you to go through tests such as:
  - Electroencephalography (EEG) – checking the electrical activity in your brain
  - Magnetic resonance imaging (MRI) – scanning for anything abnormal in the structure of the brain

Treating Epilepsy

There is no cure for epilepsy, but we can manage it using:

- Anti-seizure **medications**
- **Implants** to regulate electrical activities in the brain
- **Surgery** to remove abnormal section(s) of the brain

Living with Seizures / Epilepsy

1. **Make sure you are safe** if you have epilepsy or are prone to having seizures.
   - Take showers instead of baths
   - Do not drive a car or operate heavy machinery without your doctor's advice
   - Avoid activities at great heights or around open water
   - Tell others how to help before you have a seizure
2. **Decrease the risk** of seizures.
   - Actively manage or avoid your seizure triggers
   - Get enough sleep
   - Avoid alcohol and recreational drugs

What To Do If a Person Has a Seizure

- Stay **calm** – Most seizures only last a few minutes
- Note the **start time** of the seizure and **how long** it lasts
- Remove sharp or hard items (e.g. eyeglasses) to prevent injuries
- Cushion the person's head (e.g. put a pillow or some towels under the head)
- Loosen tight clothing around the neck
- **Do not put anything into the person's mouth or between their teeth – this creates a choking hazard. This is an outdated practice and can cause injury.**
- Do not hold the person down or try to stop their movements
- Do not let people crowd around the person
- Once the seizure stops, turn the person onto their side into a **recovery position** [video]
- Stay with the person until they are fully recovered and aware of their surroundings. The person may be drowsy and disoriented for up to 20 minutes
- Do not give the person anything to eat or drink until they are fully recovered and aware of the surroundings
- Reassure and support the person
- **Call 911** (or your local emergency service) if:
  - The seizure lasts >5 minutes
  - The person has a fever
  - The person has diabetes
  - The person is injured or has trouble breathing after the seizure
  - The person has more seizures without gaining awareness after the first

For more info, please see First Unprovoked Seizure (Nova Scotia Health) and Epilepsy (National Health Service, UK)

Resources

- Epilepsy – Cleveland Clinic, USA
- Epilepsy – Mayo Clinic, USA
- Epilepsy – National Health Service, UK
- First Unprovoked Seizure – Nova Scotia Health, Canada

❖ Seizure and Caring for seizures graphics from Freepik.com

- Neurologic challenges in 22q11.2 deletion syndrome – 2018
- Neurological manifestation of 22q11.2 deletion syndrome – 2022
- Updated clinical practice recommendations for managing [children][adults] with 22q11.2 deletion syndrome – 2023
- Cross-sectional and longitudinal findings in patients with proximal 22q11.2 duplication: A retrospective chart study – 2022

International 22q11.2 Foundation

The mission of the International 22q11.2 Foundation is to improve the quality of life for individuals affected by chromosome 22q11.2 differences through family and professional partnerships.
This information is brought to you by the Foundation for educational purposes only. It is not intended to be taken as medical advice. If you have concerns, please talk to your healthcare provider.

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