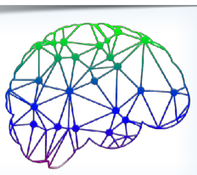
SYNGAP1
NEUROLOGY 101
THE BRAIN
INFOGRAPHIC

Syngap1 is a protein used in the brain



Brain is composed of around 100 billions neurons

Neurons process and transmit information through electrical and chemical signals

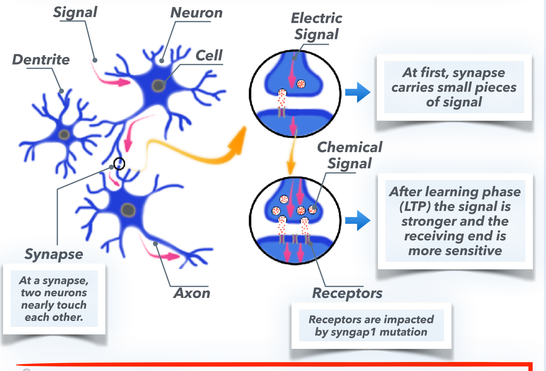
Neurons are what is called "grey matter" and compose 60% of the brain

SYNGAP1 stands for "Synaptic Ras GTPase-activating protein 1"
This is part of a family of proteins which are involved in transmitting signals between cells.

Neurons communicate between themselves through Synapses

There are between 1000 and 10000 synapses per neuron

-Axons convert electrical information to chemical information vessels
-Dentrites receive chemical information and convert it back to electricity
Axons and dendrites are called "white matter" (40% of the brain content)



At a synapse, two neurons nearly touch each other.

At first, synapse carries small pieces of signal

After learning phase (LTP) the signal is stronger and the receiving end is more sensitive

Receptors are impacted by syngap1 mutation

Day to day Analogy:

- Axons are like a speaker and send information
- Dentrites are like microphones and receive what is sent by axons
- The more you use the pathway, the more the speaker is loud and the more the microphone is sensitive

Brain neuroplasticity

The brain is plastic and is able to change based on experience
This is called neuroplasticity

New thoughts and skills shape out new pathways

Repetition and practice strengthen these pathways forming new habits

This learning phase is called: Long Term Potentiation

Old pathways are used less and weaken

This unlearning phase is called: Long Term Depression

Neural pathways connect different areas of the brain
Each pathway is associated with a particular behavior or action

Every time we think, feel or do something, we strengthen this pathway

Habits are well travelled pathways
Our brain finds these things easy to do.

Day to day Analogy 1:

- If you cross for the first time a wheat field, you will leave a small path
- The more you use the same path, the larger it become
- If you stop using the path, the wheat will grow again and will reduce the path size

Day to day Analogy 2:

- Pathway looks like dirt road at first
- If many cars use it, the city will put tar and change it to street, then road and eventually highway.
- If it is not used anymore and not maintained, the tar will go away and it will be a dirt road again

Impact of Syngap1 Mutation

One role of Syngap1 protein is to control conveying and binding of new receptors to the synapse

Regulation of receptors binding is crucial to have proper long term potentiation. Syngap1 mutation impacts the regulation of new receptors creation

Syngap1 mutation impacts as well brain plasticity by accelerating the maturation of the dendrites too fast (size of the reception area)

Impacted neuronal growth, functional maturation, and establishment of functional brain circuits are linked to Syngap1 syndrome

Day to day Analogy:

- Syngap protein job description looks like the sound engineer of a music band
- Engineer role is to put the right number of microphones to the right synapses spots and adjust sound volume and balance (up or down)
- Not having the right balance does not prevent to listen to music but you potentially need to listen multiple times to fully hear all sounds and voices
- With Syngap1 mutation, the stage is too big and there are too many microphones used

References, sources and further reading

Sources used:

- Richard Huganir lab homepage
  - http://neuroscience.bs.jhmi.edu/huganir/
- Gavin Rumbaugh lab homepage
  - https://www.scripps.edu/rumbaugh/
- Epilepsy Research Centre, Australia
  - http://www.epilepsyresearch.org.au/
- Genetic and neurodevelopmental spectrum of SYNGAP1-associated intellectual disability and epilepsy
  - Mignot C, von Stülpnagel C, Nava C, et al. J Med Genet Published Online First: March 17, 2016 doi:10.1136/jmedgenet- 2015-103451
- SYNGAP1 encephalopathy: A distinctive generalized developmental and epileptic encephalopathy
  - Neurology 92(2):10.1212/WNL.0000000000006729 · December 2018
- https://www.khanacademy.org/science/health-and-medicine/executive-systems-of-the-brain/modal/v/long-term-potentiation-and-synaptic-plasticity
- https://opentextbc.ca/anatomyandphysiology/chapter/12-5-communication-between-neurons/

More information on:
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Note : information page was written by Syngap parents, not medical professionals. Medical professional review has been performed. This is based on interpretation of current research paper related to Syngap1 protein

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