

Synapse and autism

In 2003, it was discovered that certain symptoms of autism were linked to synapses.

📌 **What synaptic structures might underlie some of the symptoms associated with autism?**

1. Architecture of a synapse

- Using all the information at your disposal, construct a balance diagram by completing the appendix using legends, cut-and-paste shapes and a title.

Don't forget to use colors!

2. Mutations and consequences on proteins

A mutation is a change in the sequence of nucleotides in DNA.

Depending on the type of change, a distinction is made between substitution, deletion and insertion mutations.

Mutation and consequences on polypeptide chain synthesis:

- Silent mutation: does not affect the protein's primary structure.
- Missense mutation: involves an amino acid change in the protein's primary structure.
- Nonsense mutation: results in the appearance of a protein synthesis stop message

- **Locate the mutations** relative to the reference sequence, by circling them in red, for the following nucleotide sequences (seq1, seq2, seq3) :

Seq ref . . . C C T C A T A A A A G T G C T A C C A T C T G T T T T C A A . . .

Seq 1 . . . C C T C A T A A C A G T G C T A C C A T C T G T T T T C A A . . .

Seq 2 . . . C C T C A T A A A A G T G C T A C C A T C T G T T T C C A A . . .

Seq 3 . . . C C T C A T A A A A G T G C T A C C A T C T G A T T T C A A . . .

- **Translate** (using the one-letter nomenclature) each of the sequences (seq ref, seq 1, seq 2, and seq 3) into an amino acid sequence (seq prot.) and **indicate** the changes observed (compared with the reference sequence) by **underlining** them.

- **Specify** the mutation involved for each sequence (seq 1, seq 2, seq 3).

Seq ref . . . CCTCATAAAAGTGCTACCATCTGTTTTCAA . . .

Seq prot.ref

Seq 1 . . . CCTCAT AACAGTGCTACCATCTGTTTTCAA . . .

Seq prot.1

Mutation

Seq 2 . . . CCTCATAAAAGTGCTACCATCTGTTTTCAA . . .

Seq prot.2

Mutation

Seq 3 . . . CCTCATAAAAGTGCTACCATCTGATTTCAA . . .

Seq prot.3

Mutation

- **Write a report** of no more than twenty lines, answering the following question:

In your opinion, what structural anomaly(ies) could there be in a synapse that would explain certain autistic symptoms, and what would be their origin?