NEWS

Genetics: Autism, epilepsy cases share mutations

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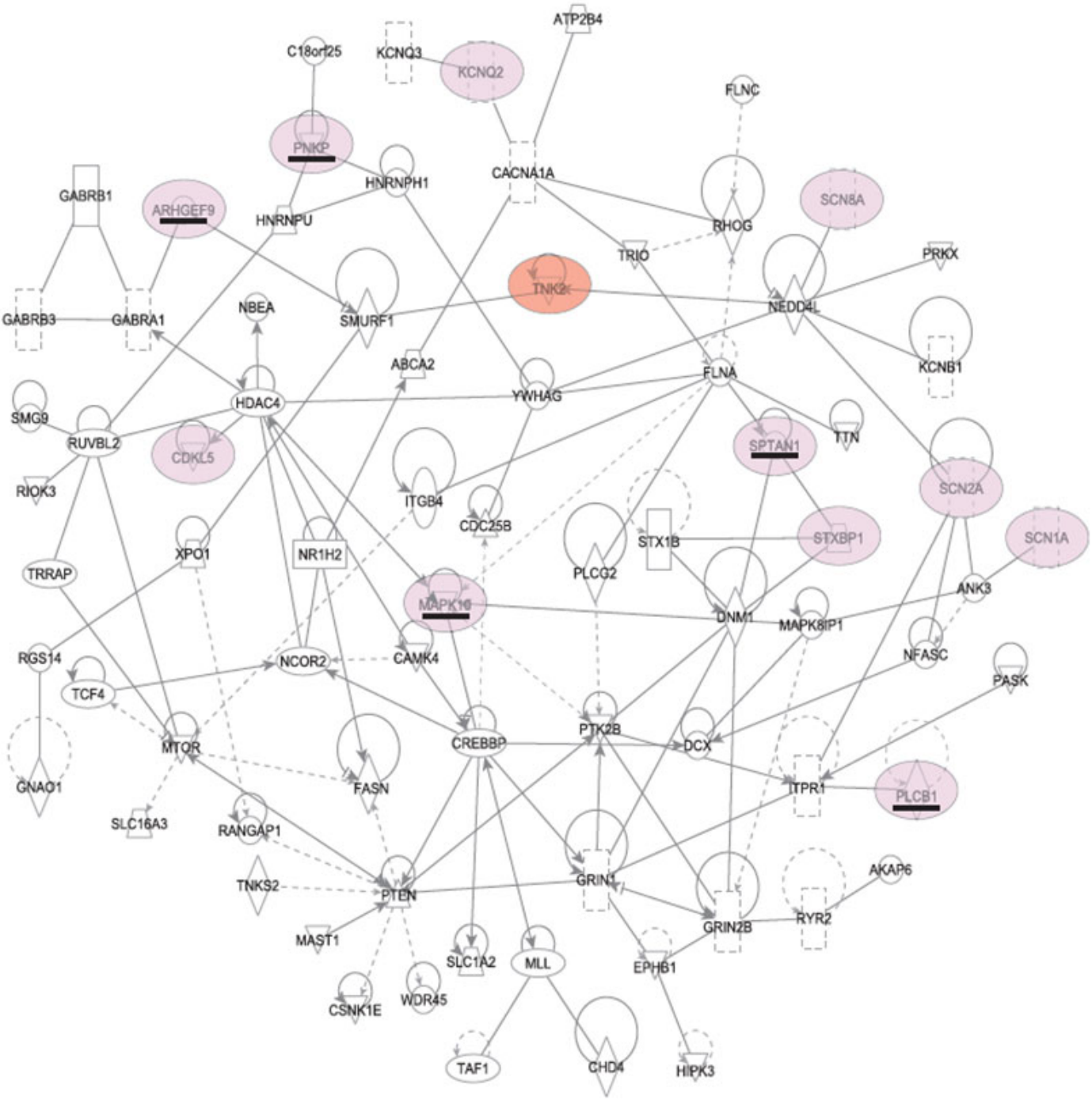

Mutations in GABRB3, a brain receptor linked to autism, are prevalent in severe childhood **epilepsy**, according to a study published 12 September in *Nature*[1].

The study also found that many of the spontaneous, or *de novo,* mutations found in children with epilepsy overlap with those linked to autism and **fragile X syndrome**.

About one-third of people with autism **suffer from epilepsy**. This overlap suggests that the two disorders may have a common origin — a theory borne out by **examples of shared genetics**.

Studies suggest that, biologically, autism is the result of an **imbalance between excitatory and inhibitory** brain signals, which may also underlie epilepsy.

In the new study, researchers sequenced the protein-coding DNA, or **exomes**, of 264 children who suffer from either infantile spasms or Lennox-Gastaut syndrome, and their parents. Both disorders are characterized by multiple seizures during sleep, which may lead to cognitive problems.

The researchers found multiple mutations in nine genes in the children with epilepsy that are not present in their parents. In particular, four children have a mutation in GABRB3, a receptor that transmits calming signals to neurons. None of 610 controls or their parents have a mutation in this gene.

A 2009 study found a **rare variant of GABRB3** that affects neuronal signaling in 17 of 1,152 families with autism and 1 of 292 controls[2].

Overall, the researchers found 277 mutations in the exomes of children with epilepsy that are not present in their parents. Of these, 41 are also **seen in children with autism** and 64 **may be regulated by FMRP**, the protein missing in fragile X syndrome.

References:

1: Epi4K Consortium *et al. Nature* **501**, 217-221 (2013) **PubMed**

2: Delahanty R.J. *et al. Mol. Psychiatry* **16**, 86-96 (2011) **PubMed**