

Genetic defect found to cause severe epilepsy and mental retardation

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A research team at Ben-Gurion University of the Negev and Soroka University Medical Center in Beer-Sheva, Israel has detected a genetic mutation resulting in a progressive disease of severe mental retardation and epilepsy beginning at infancy. The research was just published in the *American Journal of Human Genetics*.

The team, led by BGU Prof. Ohad Birk of the National Institute for Biotechnology in the Negev determined that the defect is associated with the production of the 21st amino acid, selenocysteine (SEC), which leads to progressive [brain atrophy](#).

According to Prof. Birk, "One out of every 40 Jews of both Moroccan and Iraqi ancestry may be carriers of this mutation. As the disease is both severe and common, testing for these mutations is expected to become a routine prenatal genetic screening test in these two populations, enabling prevention of future cases."

It is believed that further research will identify other mutations in the same gene as the cause of [mental retardation](#) with epilepsy in other communities. As the disease is progressive, elucidation of its [molecular mechanisms](#) might open new venues to treatment, preventing disease progression.

The human [genetic code](#), as deciphered some 50 years ago, encodes 20 amino acids which are the building blocks of all proteins in the human body. However, in recent years it became apparent that a 21st amino

acid exists: selenium, entering the body in food, is incorporated in the human tissues into what is known as selenocysteine.

This 21st amino acid is unique in that it is encoded by what is normally a stop codon – that is, a DNA sequence that normally instructs the protein building system to end the chain of amino acids, terminating the generated protein. In contrast with most genes, some 25 genes have a unique component that manipulates the stop codon so that instead of terminating the evolving protein chain, it inserts at that point an SEC building block.

Provided by American Associates, Ben-Gurion University of the Negev

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