Down Syndrome (Trisomy 21) and Dementia

Overview of Down Syndrome

The most common form of Down syndrome is **Trisomy 21**, which accounts for 95% of individuals with Down syndrome. Trisomy 21 occurs when a person is born with an extra copy of **chromosome 21**, one of the 23 human chromosomes. Each chromosome occurs in pairs, with one copy inherited from the mother and one from the father. In individuals with Down syndrome, all three copies of chromosome 21 are present instead of the usual two. Scientists believe this extra copy results from a random occurrence during cell division that produces eggs and sperm.

Human chromosomes contain approximately 30,000 genes, which carry the biological blueprint for the body. Genes instruct the body on how to build proteins — molecules essential for all the body's structures and functions. Researchers have identified over 400 genes on chromosome 21 and expect more to be discovered.

The extra copies of genes in Down syndrome, though all three copies carry protein codes, lead to developmental disabilities and various health issues. Down syndrome typically impacts learning, language, and memory, although the severity of the impact varies from person to person. Other common health problems experienced by individuals with Down syndrome include congenital heart conditions, issues affecting bones and muscles, and vision and hearing impairments. A key goal of Down syndrome research is to understand how extra chromosome 21 and its genes contribute to these health challenges.

Increased Life Expectancy and Dementia Risk

With advances in medical care, the average life expectancy of individuals with Down syndrome has increased to **60 years**. However, this longevity brings an additional health risk: a significantly increased likelihood of developing a form of **dementia** that is either identical to or similar to **Alzheimer's disease**. Dementia has now become one of the leading causes of death in people with Down syndrome.

Autopsy studies have shown that by age 40, the brains of almost all individuals with Down syndrome exhibit brain changes typical of Alzheimer's disease, including **beta-amyloid plaques** and **tau tangles**. Despite the presence of these brain changes in virtually all individuals with Down syndrome, not everyone will develop Alzheimer's symptoms. In some cases, individuals may develop these symptoms earlier in life. Researchers are still trying to understand the factors that contribute to this variation.

Prevalence of Dementia in Down Syndrome

As in the general population, advancing age increases the likelihood that an individual with Down syndrome will develop dementia, either identical to or similar to Alzheimer's. Studies indicate that individuals with Down syndrome face a **90% lifetime risk** of developing dementia. For most, this occurs in their **50s**, though the age of onset can vary widely.

Symptoms of Dementia in Individuals with Down Syndrome

Early signs of brain changes leading to dementia may include shifts in personality, cognition, or behavior when compared to an individual's previous functioning. Common symptoms include:

- Difficulty remembering new information
- Decline in attention span
- Trouble finding the right words
- Difficulty following instructions or completing routine tasks
- Taking longer to respond to new situations or information
- Withdrawal from social situations or favorite activities
- Irritability, uncooperative behavior, or aggression
- Changes in walking, coordination, or motor skills
- Mood changes such as sadness, fearfulness, or anxiety
- Increased noisiness or excitability
- Seizures that begin in adulthood

Researchers continue to study the connection between Down syndrome and dementia to better understand these health challenges and improve care for those affected.