

70 years old and going strong with Down syndrome and no dementia

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In the world of Down syndrome, 'Mr. C' is a rarity. A real person whose progress has been tracked for the past 16 years, at seventy, 'Mr. C' has well surpassed the average life expectancy of a person with Down syndrome, currently in the late fifties, but in the teens when 'Mr. C' was born. Further, 'Mr. C' does not exhibit clinical symptoms of Alzheimer's disease, which is almost a given for people with typical Down syndrome over 65 years of age.

'Mr. C', while remaining nameless, puts an optimistic face on the future of aging for people with Down Syndrome, as scientists ask the critical question: What is it about 'Mr. C's individual characteristics and experiences that have made him not only live longer, but also age successfully despite having Down syndrome? The case of 'Mr. C', including descriptions of comprehensive cognitive, behavioral, and genetic analyses and implications for research is published in the June 2008 issue of the journal, *Intellectual and Developmental Disabilities*. To read "Successful Aging in a 70-Year-Old Man With Down Syndrome: A Case Study" by Sharon J. Krinsky-McHale et al., click [here](#).

"'Mr. C' paints an optimistic picture for people with Down syndrome who are aging, and says that an ordinary person with Down syndrome ought to be able to make it to seventy, once you find 'Mr. C's secret,'" explains Dr. Sharon J. Krinsky-McHale, lead author of the study and Research Scientist at the New York State Institute for Basic Research in Developmental Disabilities.

People with Down syndrome are born with an extra copy of Chromosome 21, and these third copies of genes on Chromosome 21 lead to an overproduction of certain proteins that cause the phenotype of Down syndrome, including familiar facial features, congenital heart diseases, cognitive impairments, and other conditions. Previous studies of people with Down syndrome over 75

years of age have found that they usually do not have typical Down syndrome, which 'Mr. C' does. That is, these previous cases did not have three complete copies of Chromosome 21 throughout all the cells in their bodies, but had an atypical Down syndrome genotype, explaining why they may have been less severely affected and why they had a higher life expectancy. However, 'Mr. C' is aging successfully despite having complete Trisomy 21, making this the first case of its kind reported in scientific literature.

"The understanding in the field [until] very recently was that if you had Down syndrome and were lucky enough to make it to 70 years of age, then you would have to have Alzheimer's disease," explains Wayne Silverman, the study's co-author and Director of Intellectual Disabilities Research at the Kennedy Krieger Institute in Baltimore. "But now, we have 'Mr. C', who clearly contradicts this longstanding assumption and offers a new benchmark and tremendous hope for all people with Down Syndrome to live longer and healthier lives."

The processes regulating aging and dementia are complex and it is unlikely that any single mechanism can fully explain the array of changes that occur with aging. While future research could take several directions, including studies of risk factors like cholesterol or the bio availability of estrogen, Silverman explains that the area of gene expression offers promise at this time. While an extra copy of Chromosome 21 in a person causes an overabundance of some proteins, one possible explanation for 'Mr. C's successful aging and increased life expectancy is that, for one reason or another, he does not actually have an overexpression of all the genetic material that comes along with his extra copy of Chromosome 21.

Source: American Association on Intellectual and Developmental Disabilities

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