

# Solving the mystery of Alzheimer's disease: Connections of Down syndrome

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## Abstract

Statement of the problem: Alzheimer's disease (AD), the most common form of dementia is one of the leading causes of death worldwide. Statistics indicate that the number of dementia patients could reach 75 million by 2030. To date, no cure has been found. The world is in desperate need for a solution.

Down syndrome (DS), trisomy 21, is the most common type of mental retardation. All Down syndrome individuals develop the neuropathology of Alzheimer's disease. These include amyloid plaques, neurofibrillary tangles, and enlarged early endosomes. Amyloid precursor protein (APP) gene is on chromosome 21 and overexpressed in DS. However, studies have shown that APP is not enough to manifest AD. The purpose of the study was to investigate if there are other genes that overexpressed in Down syndrome are elevated in Alzheimer's disease patients. Intersectin-1 (ITSN1) and regulator of calcineurin-1 (RCAN1) were the targets of this investigation.

Methodology: We measured the level of two genes overexpressed in DS, ITSN1 and RCAN1 in white blood cells of AD patients and post-mortem brain samples from different types of dementia including AD.

Findings: The results showed that RCAN1 was elevated in AD and Dementia with Lewy body (DLB) brains. No elevation of ITSN1 or RCAN1 was detected in the white blood samples.

Conclusion and significance: The genes that are overexpressed in DS could contribute to the manifestation of AD and DLB. The nature of this contribution needs further investigation. More sensitive detection technology might enable us to use these proteins as biomarkers of the disease

### Biography

Dr Nakisa Malakooti is an emerging scientist in the field of cognitive decline. She has expertise in Down syndrome and Alzheimer's disease. Her dedication to solve the mystery of AD and find a cure led her to look closer into the connection of DS and AD. She has years of experience in research and teaching. She is passionate about her research and finding a cure for the Alzheimer's disease in both DS and non-DS individuals.

### Publications

- 1. Malakooti, N., AIBL Research Group, Fowler, C.J., McLean, C.A., Volitakis, I., Bush, A.I., Pritchard, M.A, Rembach, A., Finkelstein, D.I. and Adlard, P.A. (2019) The Down syndrome-associated protein, Regulator of Calcineurin-1 is altered in Alzheimer's disease and Dementia with Lewy bodies, Journal of Alzheimer's Disease & Parkinsonism 9(2):462-479
- 2. Malakooti, N. (2018) A closer look into Down syndrome: unlocking the mystery of Alzheimer's disease, Preventive Medicine and Community Health, 3 (1):1-3.
- 3. Malakooti, N., Pritchard, M.A., Adlard, P.A. and Finkelstein, D.I. (2014) Role of metal ions in the cognitive decline of Down syndrome, Frontiers in Aging Neuroscience (6):136
- 4. Malakooti, N., Adlard, P.A., Pritchard, M.A., Chen, F., Volitakis, I. and Finkelstein D.I. (2015) Role of ITSN1 in metal ion homeostasis, cognition and motor function in Down syndrome. ANS/ISN Conference, Cairns, Journal of Neurochemistry
- 5. Malakooti, N., Roberts, B., Pritchard, M.A., Volitakis, I., Kim, R.C., Lott, I.T, McLean, C.A., Finkelstein, D.I and Adlard, P.A. (2019) Characterising the metalloproteome in the Down syndrome brain, Metallomics (submitted)



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