Alzheimer’s Disease and Down Syndrome—
Two Sides of the Same Coin

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A Personal Story

Anna and John J. Sie

Sophia Kay Whitten

Never doubt that a small group of thoughtful, committed citizens can change the world; indeed, it's the only thing that ever has.

Margaret Mead
The Down Syndrome and Alzheimer’s Disease Connection

Alzheimer’s Disease

• >5 million patients
• ~$172 billion annual cost
• 10% of people over age 65
• 40-50% of people over age 85

Down Syndrome

• ~400,000 patients
• ~$19 billion annual cost
• 1/691 live births
• 100% acquire features of AD
First Report of Alzheimer’s Disease

I have lost myself

Alois Alzheimer, 1907
BRAIN PATHOLOGY IN ALZHEIMER’S DISEASE

Normal Brain

Alzheimer’s Brain

Plaques and tangles

Glial Inflammation

“The glia showed numerous filaments and fatty vacuoles”
John Langdon Down, 1866
Discovery of Trisomy 21 in Down Syndrome

Jérôme Lejeune, 1958
Normal Brain

Alzheimer’s Brain

Plaques and tangles

George Glenner and Caine Wong 1984

DAEFRHDSGYEVHHQKLVFFAEDVGSNKGAIIGLMVGGVVIA

Aβ Peptide
The Amyloid Precursor Protein Gene Is Encoded on Chromosome 21

The ‘Swedish’ Alzheimer’s Disease Family Inherits a Mutant APP Gene
Mice With Mutant APP Gene Accumulate Amyloid Deposits
RADIAL ARM WATER MAZE START
RADIAL ARM WATER MAZE FINISH LINE
A RECRIPROCAL AD—DS CONNECTION


Review and Hypothesis:
Alzheimer Disease and Down Syndrome—Chromosome 21
Nondisjunction May Underlie Both Disorders

Huntington Potter

Department of Neurobiology, Harvard Medical School, Boston
COUNTING CHROMOSOMES
Trisomy 21 In Alzheimer’s Disease

Skin Cells

Nerve Cells
Potential AD/DS Therapy

1. Suppress the formation or activity of Chr 21

2. Block the toxicity of Aβ
IS ENVIRONMENT IMPORTANT?
Caffeine Consumption of AD Patients Is Half That of Normal Subjects
High Blood Caffeine Levels are Associated With Memory Retention
Getting started:

1) Opening of the Memory and Dementia Clinic

To make an appointment:

- Have your referring physician FAX your medical records, insurance information and demographic information to (720) 848-2106.
- Please allow 2 business days for processing of your paperwork. Then CALL (720) 848-2080 to schedule your appointment.
- Be sure to bring any MRI/CT scans, lab results, and pertinent medical records to your appointment.

We accept all PPO/HMO/POS plans. Please call your insurance carrier to verify benefits and coverage at University of Colorado Hospital. Medicaid and CICP are accepted in available resident’s clinic only for a one-time consultation; wait list may apply.
Getting started:

2) **Global Down Syndrome Foundation/LCI** Support of The Adult Down Syndrome Clinic and AD Center

a. Barry Martin, M.D.; Dennis McGuire, Ph.D.
b. Patient Referral to the Memory And Dementia Clinic
c. Blood biomarker analysis of AD and DS samples
d. Research on the toxicity of Aβ in AD and DS
Getting started:

3) Clinical Trial of Leukine (human GM-CSF) in Alzheimer’s Disease
Getting started:

4) May 14, 2014: Applied for an NIA Grant and Designation as the Rocky Mountain Alzheimer’s Disease Research Center

a. Unique strength—Down Syndrome; Native Americans
b. Community partnership
   University of Colorado
   Global Down Syndrome and other Foundations
   Individual Donors
c. Gubernatorial letter of support
d. $250,000 initial funding allocation from the Legislature
In The End, Everything Will Be OK.
If It’s Not OK, It’s Not The End.

Unknown