

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

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University of Colorado
Anschutz Medical Campus

Department of Neurology



**LINDA CRNIC
INSTITUTE**
FOR DOWN SYNDROME™



**GLOBAL
DOWN SYNDROME
FOUNDATION®**



Children's Hospital Colorado
® Anna and John J. Sie Center for Down Syndrome

A Personal Story



Anna and John J. Sie

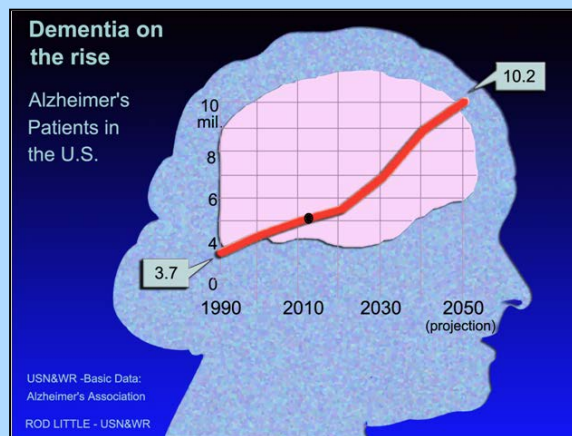


Sophia Kay Whitten

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

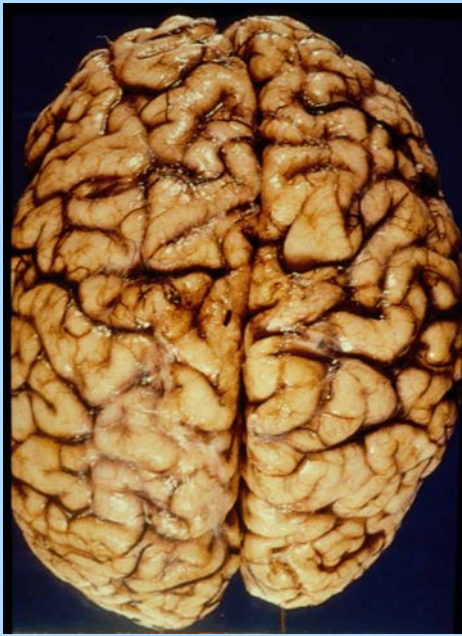


Alois Alzheimer

Alois Alzheimer, 1907

BRAIN PATHOLOGY IN ALZHEIMER'S DISEASE

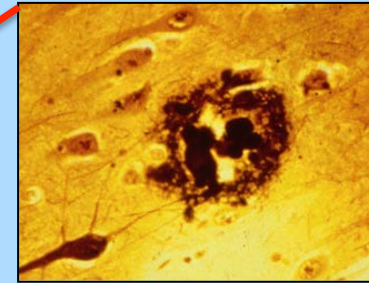
Normal Brain



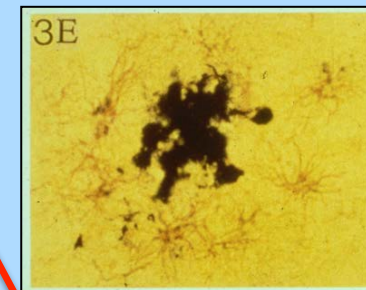
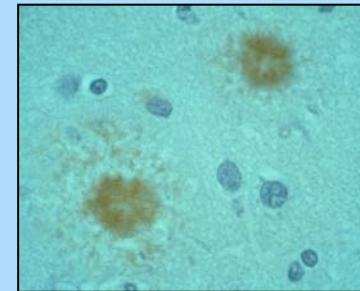
Alzheimer's Brain



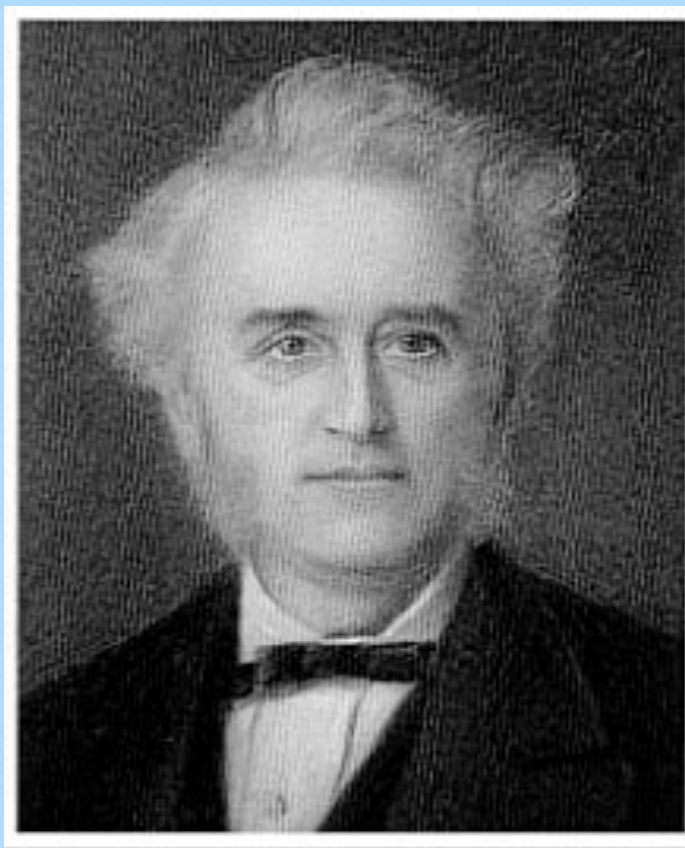
Plaques and tangles



Glial Inflammation

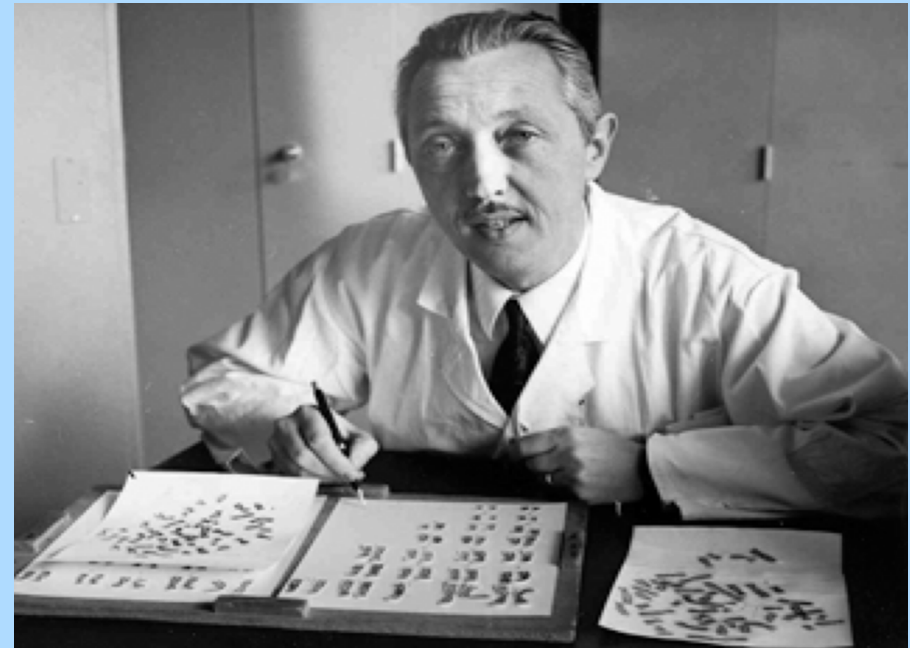
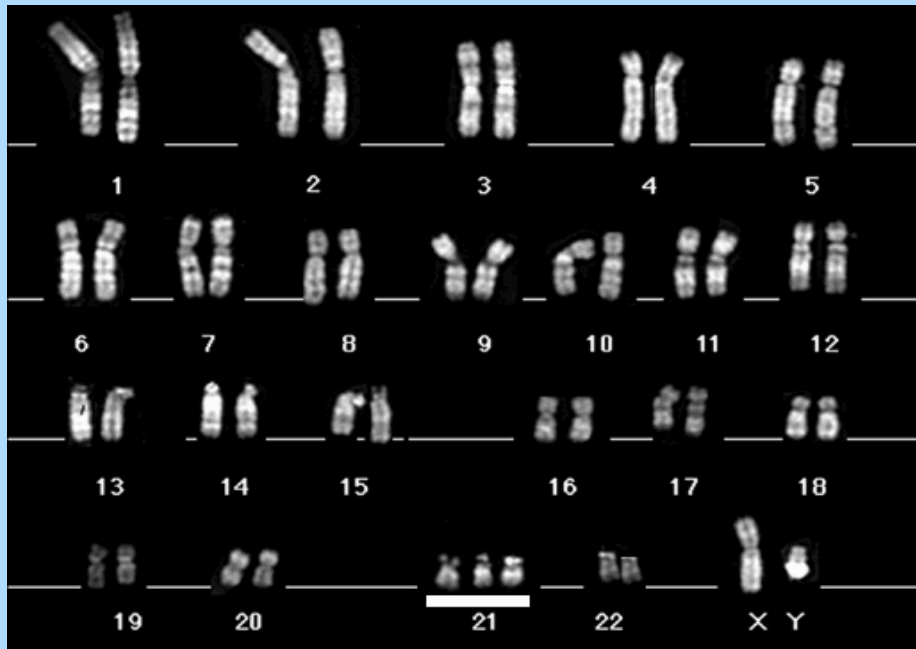


"The glia showed numerous filaments and fatty vacuoles"



John Langdon Down, 1866

Discovery of Trisomy 21 in Down Syndrome

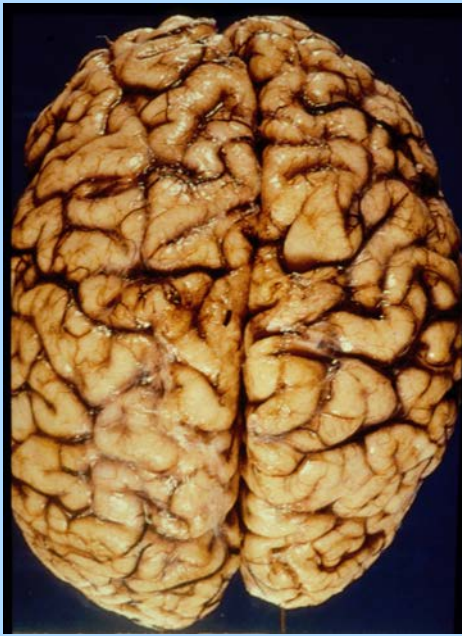


Jérôme Lejeune, 1958

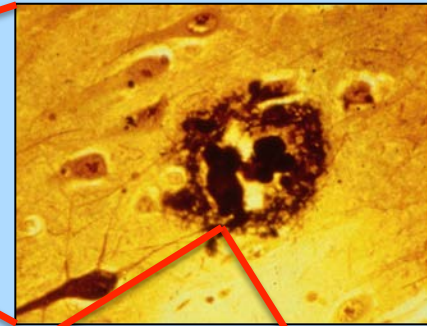
BRAIN PATHOLOGY IN ALZHEIMER'S DISEASE

Normal Brain

Alzheimer's Brain



Plaques and tangles

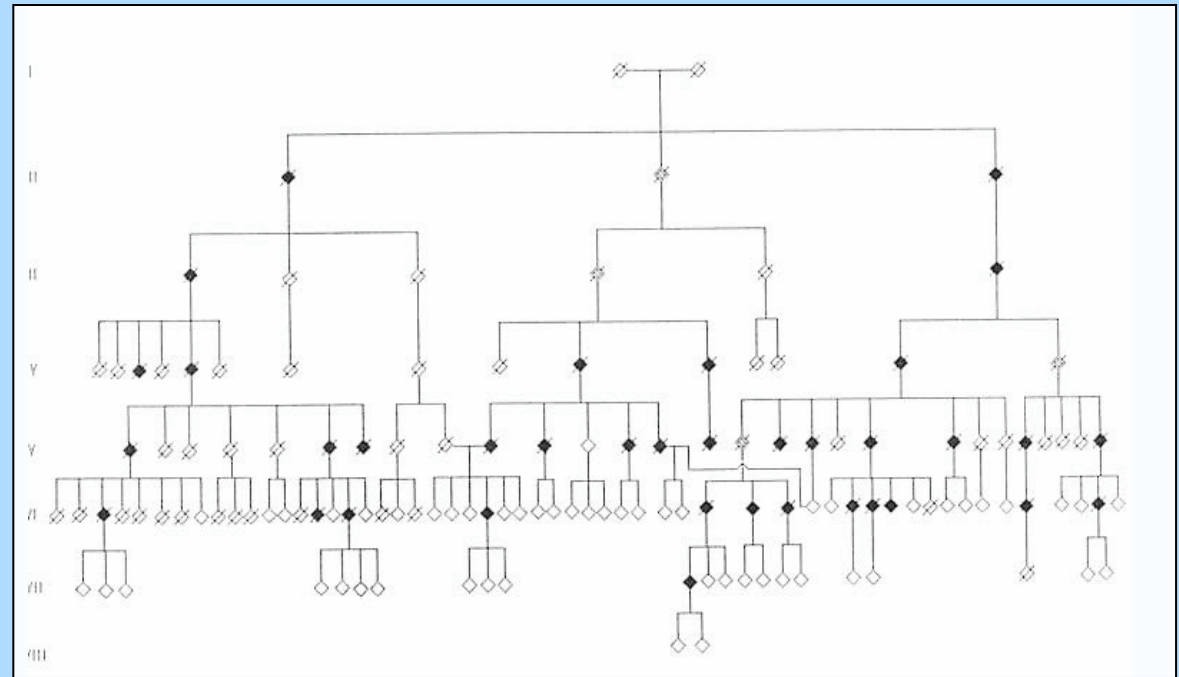
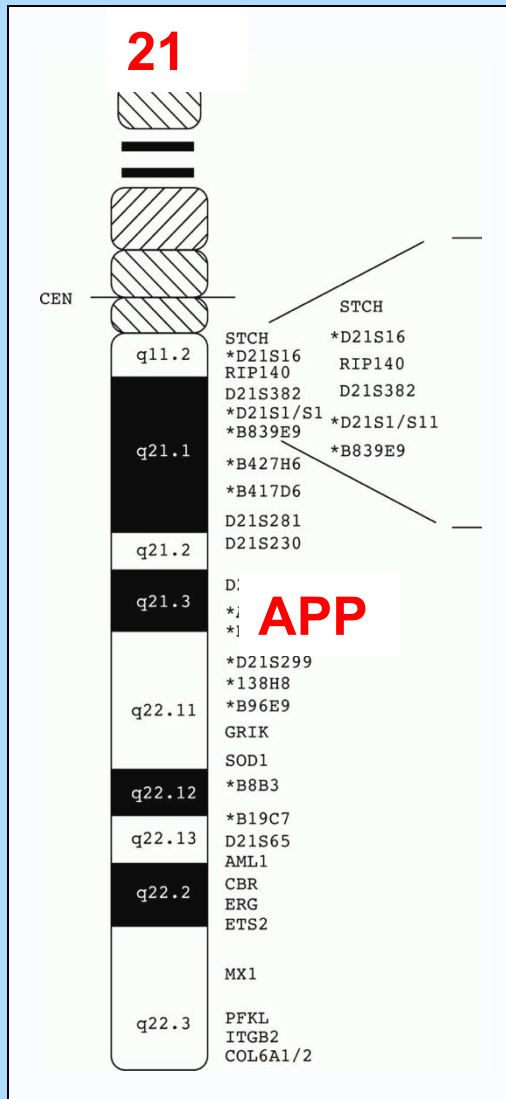


George Glenner and Caine Wong 1984

DAEFRHDSGYEVHHQKLVFFAEDVGSNKGAIIGLMVGGVVIA

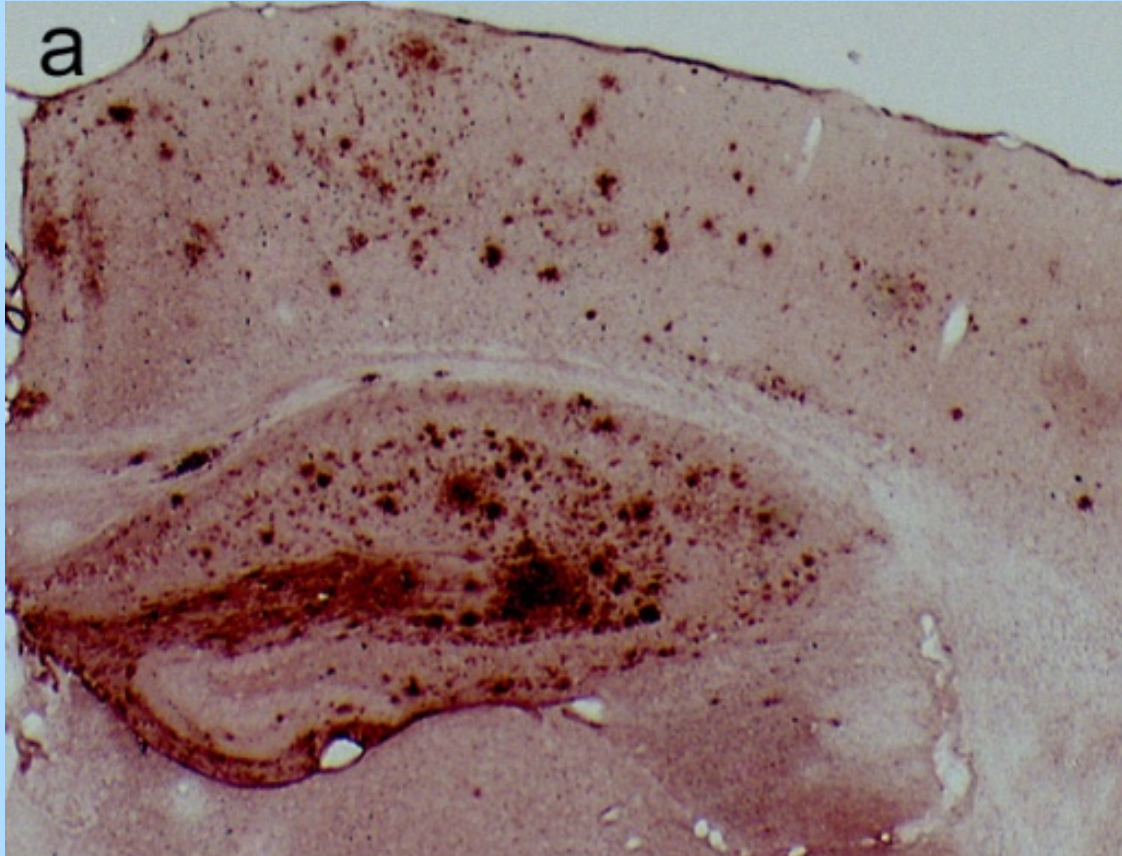
A β Peptide

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 RECIPROCAL AD—DS CONNECTION

Am. J. Hum. Genet. 48:1192–1200, 1991

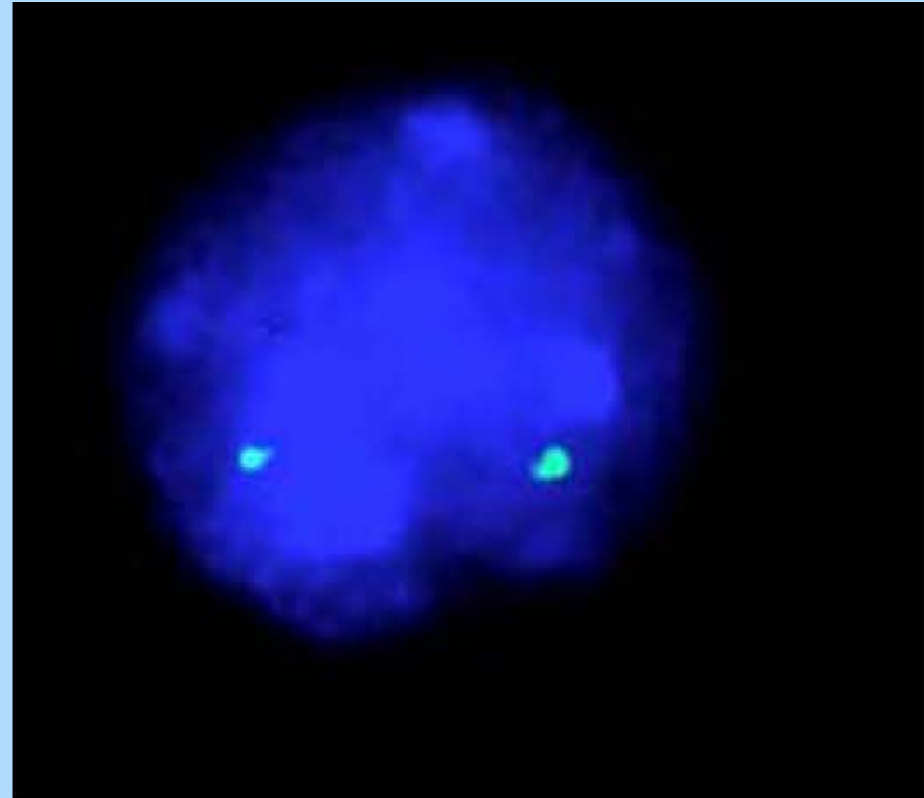
Review and Hypothesis: Alzheimer Disease and Down Syndrome—Chromosome 21 Nondisjunction May Underlie Both Disorders

Huntington Potter

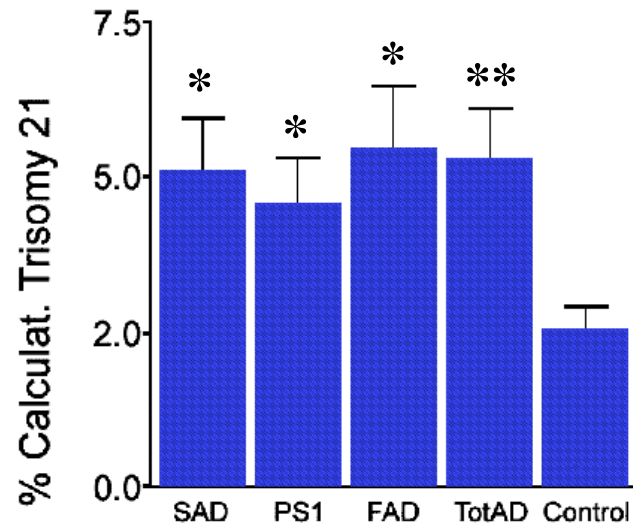
Department of Neurobiology, Harvard Medical School, Boston



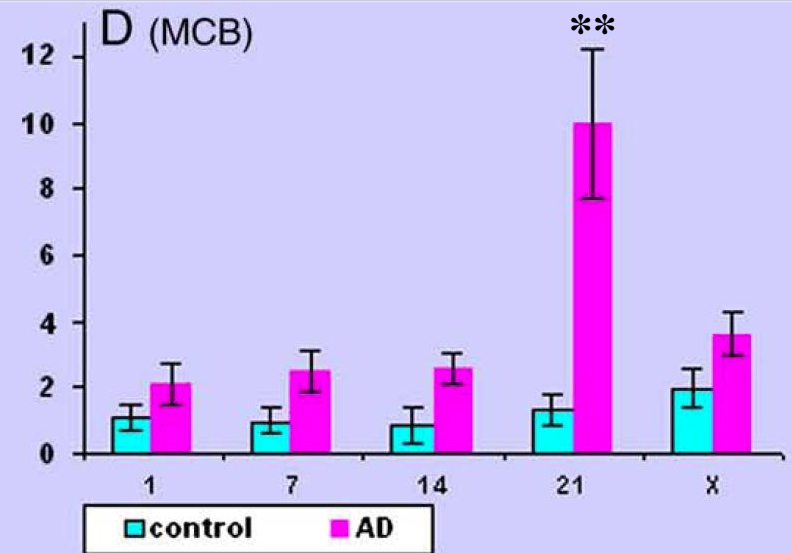
COUNTING CHROMOSOMES



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 $A\beta$

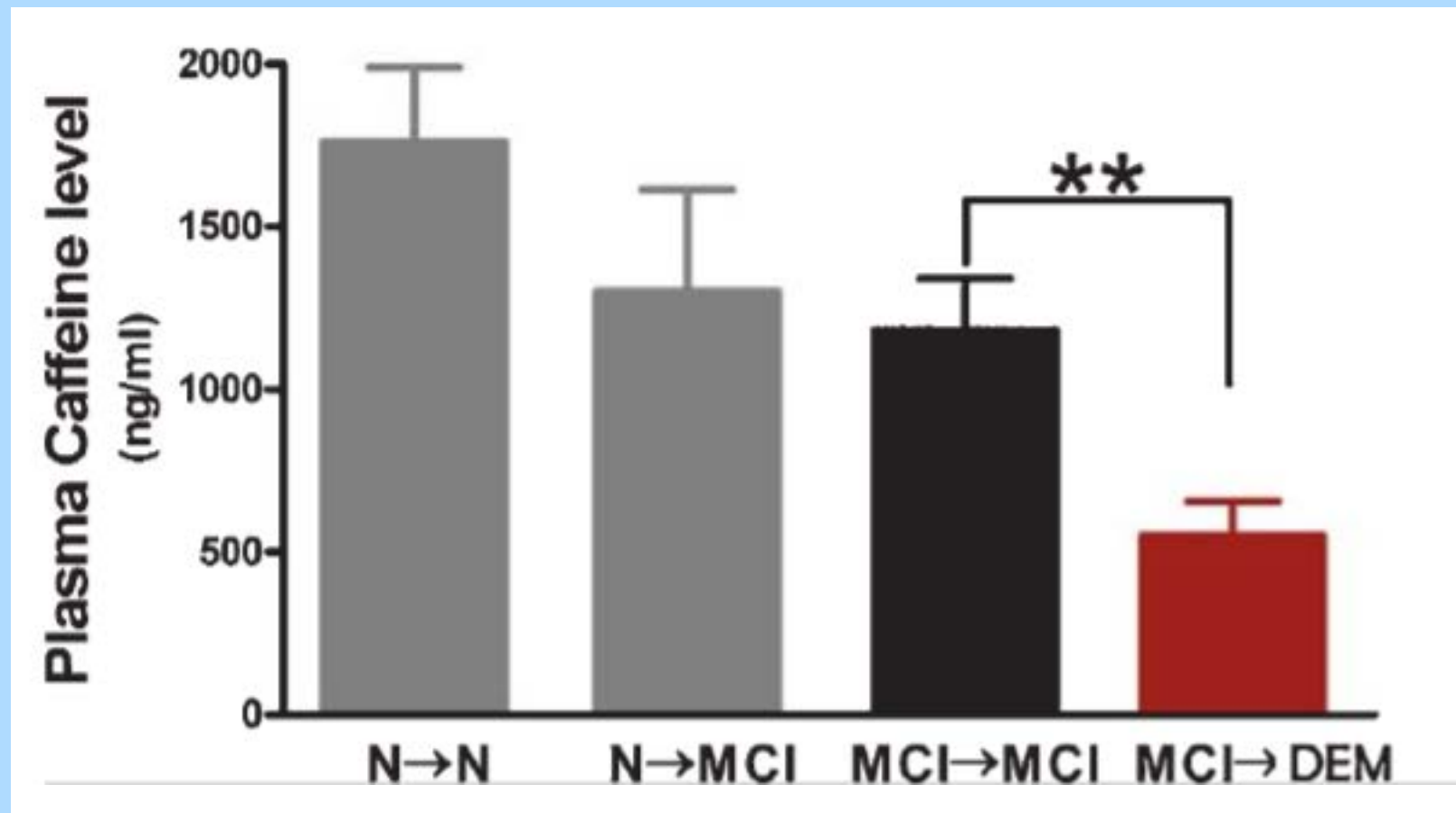
IS ENVIRONMENT IMPORTANT?



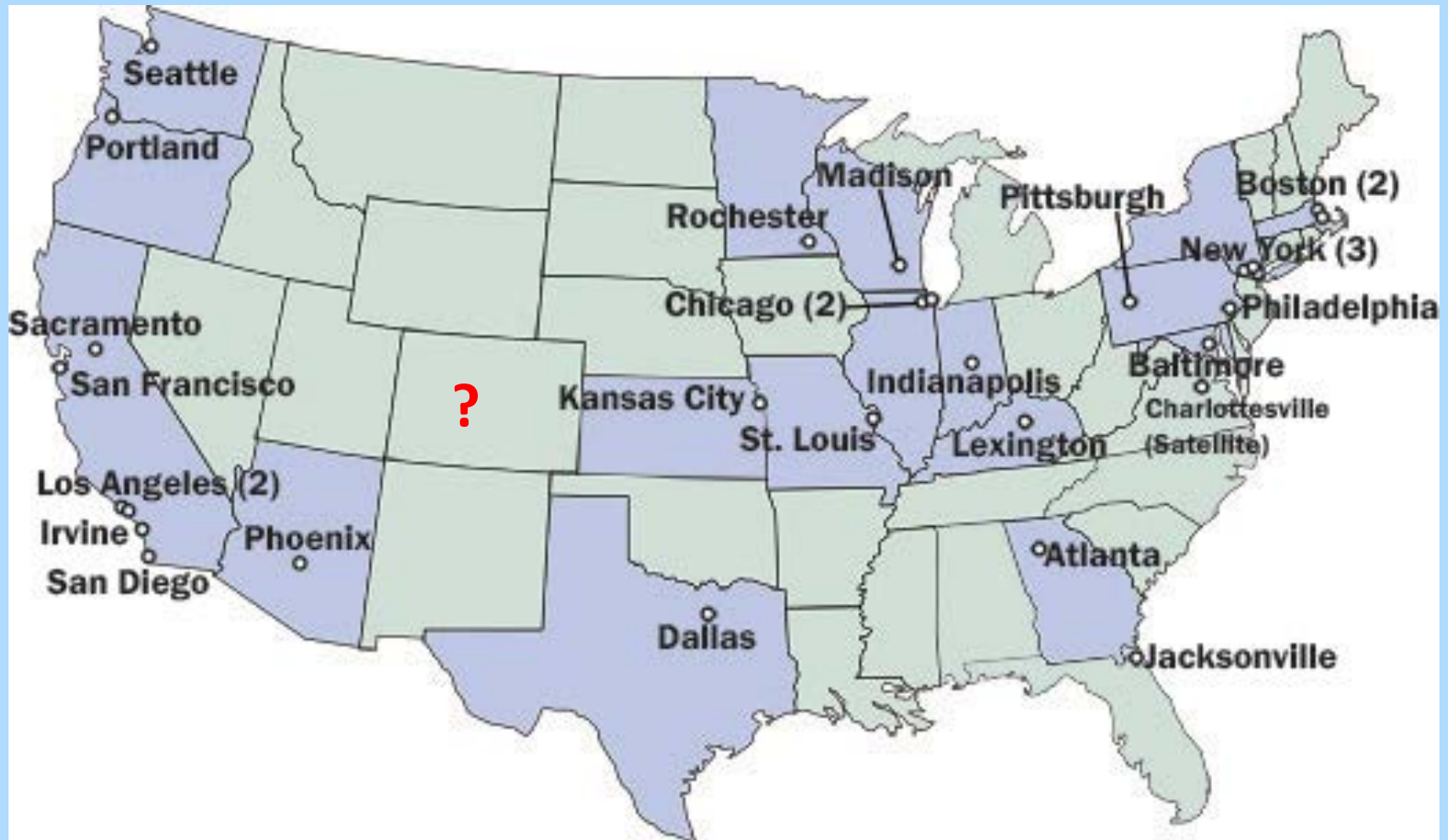
Caffeine Consumption of AD Patients Is Half That of Normal Subjects



High Blood Caffeine Levels are Associated With Memory Retention



NIA ALZHEIMER'S DISEASE CENTERS



Getting started:

1) Opening of the Memory and Dementia Clinic



UNIVERSITY OF COLORADO HOSPITAL
Anschutz Medical Campus

MEMORY AND DEMENTIA CLINIC

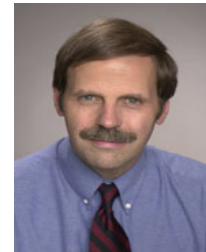
To make an appointment:

- 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.



Jonathan H. Woodcock, MD
Assistant Professor of Neurology
Director, Memory and Dementia Clinic



Christopher M. Filley, MD
Professor of Neurology



Huntington Potter, PhD
Professor of Neurology
Director, Alzheimer's Disease Programs



UNIVERSITY OF COLORADO HOSPITAL
Anschutz Outpatient Pavilion
1635 Aurora Court | Aurora, CO 80045 | 720-848-2080

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 A β 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

