Alzheimer’s disease in Individuals with Down Syndrome

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Dementia

• Umbrella term: Not a single disease- wide range of medical conditions
• Changes thinking, memory, judgment, behavior and feelings
• Can be temporary but often permanent
• Affects learning and language
• Results in significant difficulty in daily function
• Develops most commonly in persons over 80
• Caused by plaques and tangles in brain
• Varies from person to person
• Describes the “what”. Diagnosis gets to the “why”.
• Wellness and Strengths = new
Understanding dementia

Dementia

- Reversible dementias
- Vascular dementia
- Alzheimer's disease
- Frontotemporal dementia
- Lewy body disease
Alcohol, drugs, medication interactions

- Depression, delirium
- Emotional disorders
- Metabolic disorders (e.g., hypothyroidism)
- Eye and ear impairments
- Nutritional (e.g., B12 deficiency)
- Tumors
- Infections
- Alcohol, drugs, medication interactions
Down Syndrome

- Trisomy 21
- 30,000 genes code biological blueprint
- 400 or more genes on chromosome 21
- Affects learning, language, and memory
- Varies person to person
- Gains in well-being and longevity
- By 40 plaques and tangles- role of beta-amyloid fragments
- Increased risk of dementia
- Few with DS would have DIAN
Early Warning Signs of Alzheimer’s disease in persons with Down Syndrome
Change from Baseline

- Reduced Sociability
- Decreased enthusiasm
- Decline in attention
- Sad, fearful, anxious
- Irritable, uncooperative, aggressive
- Restless
- Sleep disturbed
- New seizures
- Decreased coordination and mobility
How the brain works

- There are 100 billion nerve cells, or neurons, creating a branching network.
- Signals traveling through the neuron forest form memories, thoughts and feelings.
- Alzheimer’s disease destroys neurons.
Neurons affected by Alzheimer’s disease

dead cells full of tangles
sparse, damaged cells
amyloid plaques
withered branches
Which functions are affected?

- Language, sense of temperature, touch, pain
- Vision
- Judgment, reasoning
- Memory, language, hearing
- Movement, balance
- Basic functions, including breathing
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- Basic functions, including breathing
- Basic functions, including breathing
Healthy vs. Alzheimer’s brain

Healthy Brain

Severe AD

Ventricles enlarge

Cortex shrivels, especially near hippocampus
Major risk factors

- The primary risk factor is age
- The incidence is higher in women due to women living longer
- Down syndrome is correlated with Alzheimer’s
- Family history can increase risk
- There are two categories of genes

Estimated percentage of Americans over age 71 with dementia
Person Centered Diagnosis= Difficult

• Document baseline by 35- Rocco Assessment
• Reliable informants - observation and report Professional assessment – with cognitive assessments for DS (Don’t use as only benchmark) – SLU, Washington University, Pujols Center
• Rule out impact of other conditions ) thyroid, depression, infections, sleep apnea)
• Imaging
Selecting a doctor

- Doctors can diagnosis Alzheimer’s disease with accuracy
- Choose from:
  - Regular primary care physician
  - Geriatrician
  - Neurologist
  - Psychiatrist
  - Neuropsychologist
Preparing for the doctor’s visit

- Keep a symptom log
  - Write a list of symptoms, be specific
  - Include when, how often and where
  - Develop the list with input from other family members

- List current and previous health problems

- Bring all medications (prescriptions, vitamins, herbal supplements and over the counter medication)
The doctor’s visit

- Medical and family history
- Physical and neurological exam
- Lab tests
- Mental status exam
- May include brain imaging (MRI, CT scan)
- May include neuropsych tests
Challenges

• Shortened lifespan
• No approved medicines in U.S.
• Cholinesterase inhibitors ok’d in U.K.
• Cochrane Review= Memantine no benefit
• Use caution with any research breakthroughs
• Research gaps and lags
• Appropriate residential care
• Trained staff
Plan early

- Be an active partner in your long-term care plan
- Develop a relationship with your healthcare team
- Get legal and financial issues in order
- Grow a support system
- Educate yourself about the disease
Clinical research studies

- Clinical trials fuel progress toward treatments
- Participants receive a high standard of care
Promising information

- Stanford School of Medicine Down Syndrome Research Center
- Implications for treatment: GABA_A receptors in aging, Down syndrome and Alzheimer’s disease, Robert A. Rissman, et al
- A Human Stem Cell Model of Early Alzheimer’s Disease Pathology in Down Syndrome, Yichen Shi¹, et al*
- Personality and behavior changes mark the early stages of Alzheimer's disease in adults with Down's syndrome: findings from a prospective population-based study, Sarah L. Ball¹,*et al
- Mitochondrial and Molecular Medicine and Genetics (MAMMAG), University of California Irvine, Irvine, CA, USA
- Washington University – DIAN study
- Advocacy, Enroll in research
- Alzheimer’s Association and Association on Aging with Developmental Disabilities
- Baseline data
- Education
Thank You

• Questions