

Management of Down Syndrome: A 21st Century Approach

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I. Brief Overview of Down syndrome

- A. What is trisomy 21?
- B. Phenotypic characteristics
- C. Common medical findings and average IQs

II. Current Protocols Utilized for Cognitive Enhancement in Down syndrome

- A. Changing Minds Protocol
 - i. Nutritional supplementation
 - ii. Prozac
 - iii. Focalin
- B. Warner Protocol
 - i. Nutritional supplementation
- C. StemCells21 Protocol
 - i. Umbilical cord blood combined with growth factors & supplements

III. Current Research for Improved Cognition in Down syndrome

- A. Amyloid precursor protein (APP)
 - i. Affects the development of Alzheimer's disease
 - ii. May help predict age of onset of dementia in Down syndrome
- B. GABA-A receptor block
 - i. Decreases inhibitory communication of neurons in mouse models
 - ii. Improves learning in mouse models of Down syndrome
- C. Sonic hedgehog signaling pathway-activator compound
 - i. Restored cerebellar development in mouse models
 - ii. Improved memory and learning in mouse models
- D. L-DOPS
 - i. Corrects norepinephrine deficiency in the hippocampus of mouse models
 - ii. Improved contextual learning and memory in mouse models
- E. Development of cognitive test battery specific for use in Down syndrome
 - i. Targets specific brain regions known to be impaired in Down syndrome
 - ii. Includes non-verbal tests to separate cognition and language skills
 - iii. Sensitive measures will enable determination of therapeutic efficacy of human drug treatment trials

IV. Brief Overview of Common Visual Findings in Down syndrome

- A. High refractive errors
 - i. High hyperopia
 - ii. Lack of emmetropization
 - iii. High magnitude oblique astigmatism

- B. Strabismus
- C. Nystagmus
- D. Reduced best corrected visual acuity
- E. Reduced near visual acuity

V. Research Findings Related to Accommodative Dysfunction and Reduced Near Vision

- A. MEM retinoscopy studies (Woodhouse, et al.)
 - i. Large accommodative lags in persons with Down syndrome
 - ii. Accommodative lag does not improve with age
 - iii. Full distance correction of hyperopia may not reduce lags
- B. Accommodative Amplitude Studies (Woodhouse, et al. & Anderson, et al.)
 - i. Reduced amplitudes compared to age norms in Down syndrome
 - ii. Reduced amplitudes present even in early school years
- C. Objective Measurements of Accommodative Dynamics (Anderson, et al.)
 - i. Reduced reaction times to initiate changes in accommodation
 - ii. Increased fluctuations of steady state accommodative responses
 - iii. Poor performance likely due to sensory deficits, not mechanical deficits
- D. Bifocal Studies (Stewart, et al. & Nandakumar, et al.)
 - i. Bifocals improve near focus
 - ii. Bifocals may encourage improved accommodation once removed
 - iii. Bifocals may result in improvement in literacy skills

VI. Management of Accommodative Dysfunction in Down syndrome Patients

- A. Measure accommodation on all patients with Down syndrome
 - i. MEM retinoscopy
 - ii. Open field autorefraction
- B. Measure near acuity on patients with Down syndrome
 - i. Near LEA chart
 - ii. Measure through tentative add prior to dilation
- C. Prescribing guidelines
 - i. Full hyperopic corrections
 - ii. Bifocals fit high
 - iii. Glasses with appropriate fit
 - a. Cable temples
 - b. Specs for Us (designed specifically for Down syndrome)