

Down Syndrome – Hidden Miracles

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Down Syndrome John Langdon Down 1/800 births All groups. 3rd 21st chromosome 1-22 plus XX or XY plus extra chromosome material from 21: 90+% of the time there is an extra chromosome 21 from either egg or sperm. 2-4% of cases are mosaic. 3-4% of cases have 46 chromosomes (translocation) – additional chromosome 21 material on another chromosome (14th). 88% of trisomy 21 are maternal, 8% paternal and 2% mitotic errors.

Signs and Symptoms of Down Syndrome

- Flat face with an upward slant to the eye, short neck, and abnormally shaped ears
- Small hands and feet
- Deep crease in the palm of the hand
- White spots on the iris of the eye
- Poor muscle tone, loose ligaments

The Occurrence of Down Syndrome Not attributable to any known parental behaviors or environmental factors. Subsequent probabilities after a child with Down syndrome is 1% greater than baseline maternal age. Balanced translocation errors may not be exhibited in carrier parent.

Down syndrome and maternal age 35+ yo have 9% of babies but 25% of Down syndrome. Younger have 75% of Down babies because of higher birth rate yet lower incidence. So maternal age alone will not detect 75%

Prenatal Screening for Down Syndrome

- Nuchal translucency (US test)
- Triple or quadruple screen (looking for maternal blood markers)

Diagnostic testing for Down

- Chorionic villus sampling (9-11 wks) 1-2% risk of miscarriage
- Amniocentesis (14-18 wks) fetal cells from amnionic fluid
- Percutaneous umbilical blood sampling (PUBS) (20+ wks) greatest risk of miscarriage, most accurate

Down Syndrome Diagnosis

- Genetic testing after birth

Associated Medical Disorders

- Hypotonia (low muscle tone)
- Celiac (and food sensitivities) screen with IgA. Egg and dairy sensitivities also.
- Intestinal abnormalities/ constipation/ Hirschsprung disease
- Respiratory problems/ ear infections
- Osteoporosis
- Cardiac anomalies (evaluate with echocardiogram)

- Hypothyroidism
- Amblyopia/near/far sightedness, cataracts
- Hearing loss
- Slower growth and milestones' reached
- Increased susceptibility to infection and leukemias (tonsillitis, OM, pneumonia, chronic respiratory infections)
- Myelodysplasia – defective development of the spinal cord (esp the lower cord)
- Seizure disorders (10x)
- Atlantoaxial instability (transverse ligament)
- SOD overproduction with resultant oxidation status increase

Newborns Need Pediatric Cardiologist (50% of children with DS have abnormalities)

- Pulmonary hypertension (VSD or AVSD with no symptoms of heart failure)
- Congenital heart defects

Infants and Preschool Children

- Hearing examination
- Growth/weight charting until 18 yo and head circumference up to 36 months old
- Vision examination
 1. Amblyopia
 2. Cataracts
 3. myopia/hyperopia (near/far sightedness)

Early Intervention and Education special programs and other resources

Adolescence with Down Syndrome males are typically sterile but females menstruate and can carry a child, increased risk of baby with Down Syndrome.

Adults with Down Syndrome Americans with Disabilities Act (ADA) if have more than 15 employees

Supplementation (all are in dispute and the “waters” murky)

Biochemically compensate for genetic handicap. Roger Williams, discoverer of pantothenic acid, called this “genetotrophic concept”. Amelioration of genetic pattern by the increased supply of one or more nutrients.

August 2003 National Down Syndrome Society's position statement on vitamin therapy: “Despite the large sums of money which concerned parents have spent for such treatments in the hope that the conditions of their child with Down syndrome would be bettered, there is no evidence that any such benefit has been produced”.

- Thiamine Dr. Ruth Flynn Harrell 1981 learning disabled children IQ increase 10-15 points *Proceedings of the National Academy of Sciences*. 1946 *Journal of Nutrition* “a liberal thiamine intake improved a number of mental and physical skills of orphanage children”. “A number of well publicized studies conducted to “replicate” her work could not do so when they refused to use incomplete or inadequate doses. In spite of obvious bias, negative replication studies using incomplete or low doses are the ones that have been accepted, and Dr. Harrel's work shelved.” *Orthomolecular Medicine for Everyone* p278
- Vit E Delayed onset of dementia in Ts65Dn mouse (mouse model for DS) Antioxidants very important for children with Down's. Trisomy 21 leads to SOD overproduction without corresponding catalase or glutathione peroxidase increase

- Selenium as a thyroid cofactor, immunodeficiency, cofactor in glutathione peroxidase (free radical scavenger)
- Vit C helps with immune support. Antioxidant. Not produced by humans.
- Zinc helps with thyroid production and in immunodeficiency. Pregnant mouse models with ethanol toxicity had reduced abnormalities and spatial memory impairments with Zn supplementation.
- Acetyl-L-Carnitine (ALCAR) potentially increase visual memory and attention
- Co Q-10 Necessary component for mitochondrial energy production in heart
- Free Radical Control via antioxidants: Vit E, Vit C, Se, CoQ10
- Folic Acid for SAM cycle and methylation pathways. Reduces AGE's which affect pre and post natal brain development. Same pathway for diabetes and possibly Alzheimer's
- Multivitamin/multimineral poor digestion and dubious food quality is benefited from supplementation
- Vit A An important antioxidant and immune support stimulant
- Bioflavonoids, Quercetin, Taurine, and Lutein/Zeaxanthin for cataract support
- Digestive Enzymes Potentially children with DS need a more complete digestion of their food
- Vit B12 methyl B12 helps with clearance of toxins
- Tryptophan Precursor to serotonin, which people with DS are deficient in. 5 HTP also may be of benefit
- Arginine and Ornithine May help control the excess production of lysine found in DS
- Essential Fatty Acids (EFA) DHA and EPA necessary in myelination of brain and as an anti-inflammatory
- Choline Phosphatidyl choline and phosphatidyl serine are brain components. May help reduce dementia
- Probiotics improve gut health and reduce toxic burden. Fiber also.
- Sicca Cell Therapy (cell therapy)
- Piracetam a GABA derivative that may help with mental acuity
- DMSO The by-product is MSM which is used in sulfation phase II pathways in the body

Lifestyle Changes

- Malabsorption Balanced diet!!! Possibly liquid vitamins. These are so very important for the child with Down syndrome
- Celiac Disease diet
- Hearing Aids
- Physical Training (mindful of cardiac conditions)
- Stimulating environment
- Physical Therapy (atlanto-axial instability – transverse ligament loss)
- Realistic goals
- Other children in the family
- Love as a drug