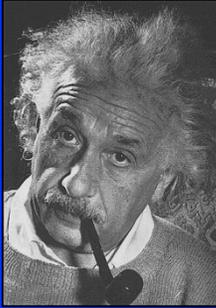


# Biomedical Basics for Trisomy 21

OPTIONS Down Syndrome Track  
Autism One Conference  
May 24, 2012

Norm Schwartz MD  
Milwaukee, Wisconsin

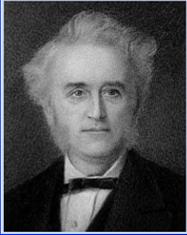
## Opinions about obviousness are to a certain extent a function of time.



Albert Einstein

- The earth is flat.
- Children should be seen and not heard
- Airplanes are impossible
- Women are not logical enough to vote
- Autism caused by bad parents
- “Down’s is not a person”

**John Down 1866**  
“...reversions to a primitive racial type”



**Atlantic Monthly 1968**  
“True guilt arises only from an offense against a person, and a Down’s is not a person.”  
Bard & Fletcher, 1968, pp. 62-64



## History

- 500 BC: First representation of Trisomy 21
- 1838: First description T 21 by Esquirol
- 1846: “the education of idiots” and extended description of T 21 by Séguin
- 1866: John Down describes the phenotype T 21
- 1932: First suggestion chromosomal origin by Waardenburg
- 1959: Extra Chromosome 21 found by Lejeune
- 1961: Geneticists “mongolism” replaced by Down Syndrome or Trisomy 21; Mongolian People’s Republic requests WHO change name
- 1989: Individualization of the Down Critical Region
- 1990s: First trisomic mouse lines
- 1997: Cold Spring Harbor meeting defining DCR-1
- 2000: Sequencing of Chromosome 21 by Hattori et al.

*Genetics IN Medicine • Volume 11, Number 9, September 2009*

“We call on all people of good will to ensure that health protection is grounded in a renewed spirituality : every patient is my brother.”  
Jerome Lejeune MD 1926-1994

1958 T21 Identified



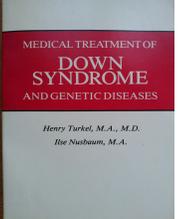
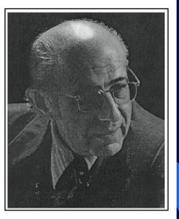

**Jérôme Lejeune**  
FONDATION  
RECHERCHE CURIE GÉNÉTIQUE

JF USA NEWSLETTER  
Volume 7, Number 1

Committed to Finding a Cure for Down Syndrome

I was the doctor who was supposed to cure them and so I leave, I feel I am abandoning them.  
By Jerome Lejeune

The World's Largest Funder of Down Syndrome Research Comes to the United States

Henry Turkel, M.A., M.D.  
The Nashua, N.H.

“Dr. Turkel had the nerve to make his claims when everyone ‘knew’ that children with genetic defects could not possibly be treated successfully.”  
-Abram Hoffer

“I know Dr. Turkel, and I can testify to his sincerity and conviction. The results that he reports are striking. There is evidence that patients would receive significant benefit.”  
-Linus Pauling

**CONTENTS OF MEDICATION COMPRISING THE "U" SERIES OF DRUGS**

	(BREAKFAST)	UPNEOID - B	(AT BEDTIME)
UMORPHOID - A			
Thyroid Globulin	66 mg	is Ulnesoid A but Enteric Coated	
Organic Iodine	66 mg		
UMORPHOID - B	(BREAKFAST)	UPNEOID - C	(P.M.)
Vitamin A	25,000 Units	Nachazolone Hydrochloride	0.05%
Vitamin E	10 mg	Pyrimamine Maleate	0.50%
UNOID - A	(BREAKFAST, LUNCH, DINNER)	Chlorpheniramine Maleate	0.25%
Pantylene Tetrazole	50 mg	Methyl Paraben	0.01%
Glutamic Acid	200 mg	Propyl Paraben	0.02%
Nicotinic Acid	50 mg	UTROPHOID - B	(LUNCH)
UPEPTOID - A	(BREAKFAST, DINNER)	Thiamin Mononitrate (B1)	20 mg
Betaine-Choline Tartrate	100 mg	Riboflavin (B2)	20 mg
Choline-Methionine Tartrate	100 mg	Calcium Pantothenate	20 mg
Inositol	50 mg	Para Amino benzoic Acid	20 mg
Unsaturated Fatty Acids	100 mg	Pyridoxine (B6)	20 mg
Liver Deseccated	75 mg	Niacin	20 mg
UPEPTOID - B	(DINNER)	UTROPHOID - C	(DINNER)
Betaine Hydrochloride	66 mg	Folic Acid	5 mg
Papain	66 mg	Cyanocobalamin (B12)	25 mcg
Pepsin	66 mg	Calcium	30 mg
Pancreatrin	66 mg	Cobalt	0.1 mg
Diastase	3.3 mg	Copper	1 mg
Katocholanic Acid	66 mg	Iodine	0.15 mg
Doposypholic Acid	66 mg	Iron	10 mg
UPNEOID - A	(BREAKFAST, LUNCH, DINNER)	Magnesium	1 mg
Phenylpropanolamine Hydrochloride	20 mg	Manganese	1.25 mg
Pyrimamine Maleate	25 mg	Molbdenum	0.1 mg
Rutin	20 mg	Zinc	1 mg
Ascorbic Acid	100 mg	Bone Meal (Breakfast, Lunch, Dinner)	200 mg
Amnophylline Magnesium Glycinate	100 mg	Glutamic Acid (Breakfast, Lunch, Dinner)	200 mg

## Medical Amelioration of Down's Syndrome Incorporating the Orthomolecular Approach

Henry Turkel, B.A., M.A. (D.Sc.), M.D.J.

When I treated my first mongoloid patient 34 years ago, the reasons for the accumulations of fats, fluids, and minerals were unknown. But I did know that all living cells require proper nutrients in optimal quantities for the specific cell to develop normally. As a medical student, I elected a course in dietetics. It became obvious to me that nutrients were important therapeutic agents in the treatment of disease, and this line of reasoning extended not only to the physical diseases, like diabetes, arteriosclerosis, and allergies, but also to so-called mental illness including retard-

ORTHOMOLECULAR PSYCHIATRY, VOLUME 4, NUMBER 2, 1975, Pp. 102-115

## Irrational Bias Against Nutrients

**In questions of science, the authority of a thousand is not worth the humble reasoning of a single individual.**

Galileo Galilei 1564-1642

- Medical Profession opposed, for no obvious reason, to Dr. Turkel - to detriment of health and well being of T21 individuals
- Dr. Turkel showed that genetic condition of T21 need not be accepted as inevitably leading to permanent defect and inability to function in society. He provided hope that improvement in functioning is possible

## Down Syndrome or Trisomy 21



The specific disease doctrine is the grand refuge of weak, uncultured, unstable minds such as now rule in the medical profession.

Florence Nightingale 1820-1910

Unwarranted medical skepticism and intransigence has unnecessarily doomed legions of Down Syndrome children to lives greatly diminished by submarginal mental and physical functioning. ...many of them could have benefitted immensely if their poor nutritional status had been taken into account and been treated properly.

Bernard Rimland, Ph.D 1928-2006  
Founder Autism Research Institute

## Parents, Clinicians, Researchers Linking Science, Observations, Evidence, and Clinical Experience for:



# Down Syndrome OPTIONS

Optimizing Potential Through Integrative Opportunities Now

**"It is much more important to know what sort of person has a disease, than what sort of disease a person has."**

Sir William Osler MD "Father of Modern Medicine" 1849-1919



## Principle-base Medicine Integrative Medicine 10:5 Oct/Nov 2011

“A therapeutic intervention is fitting the treatment to the individual. In that sense it is like tailoring ... measuring and trying it on until you get a good fit.... You don't always get it the first time.”

Sid Baker, MD

## Individual Trisomy 21 Treatment Based on physiology, lab tests, research, clinical experience, parent observation

- Reach full potential
- Support growth and development
- Normalize physiology and function
- Improve health, decrease infections
- Assist education
- Maintain health through lifespan

## Trisomy 21- Complex

- Most common genetic abnormality
- Wide range of physical features, congenital malformations, health problems
- Impact of trisomy 21 for each person is INDIVIDUAL
- From profoundly impacted to mild
- In 1930 life expectancy 9 years old, today close to 60
- Multiple genes involved effecting specific proteins, enzymes, metabolic molecules- altering cell biochemistry
- Need systems biology approach to understand\_\_

## Emergence Good and Ill Health

- How our gene patterns are transcribed
- How nutrients and phytochemicals in our food speak to our genes
- How environment influences our genes by epigenetics
- How lifestyle and environment are factors altering gene expression

## Trisomy 21 Complexity Systems Biology

- Whole is greater than sum of parts
- Complex humans cannot be understood by studying individual genes in isolation
- Gene-environment and gene-gene interactions yield emergent properties
- Complex dynamics of living systems, a new field of research -Systems Biology realizes genetic variability does not fully explain diversity of physiology, DNA regulation, or organ function

Human Molecular Genetics, 2009, Vol. 18, Review Issue 1 R75-R83  
doi:10.1093/hmg/ddp010

### Down syndrome—recent progress and future prospects

Frances K. Wiseman<sup>1</sup>\*, Kate A. Alford<sup>2</sup>, Victor L.J. Tybulewicz<sup>2</sup> and Elizabeth M.C. Fisher<sup>1</sup>

<sup>1</sup>Department of Neurodegenerative Disease, Institute of Neurology, Queen Square, London WC1N 3BG, UK and <sup>2</sup>Division of Immune Cell Biology, MRC National Institute for Medical Research, The Ridgeway, Mill Hill, London NW7 1AA, UK

Received December 19, 2008; Revised December 19, 2008; Accepted January 5, 2009

Down syndrome (DS) is caused by trisomy of chromosome 21 (Hsa21) and is associated with a number of deleterious phenotypes, including learning disability, heart defects, early-onset Alzheimer's disease and

DS was once thought to be an intractable condition because of the... have... describe... ing of F... field an... develop... rstand... in this... and to... ie syn...

**Box 1: What is a gene?**

The definition of a gene has shifted over the past 100 years since it was first coined by Wilhelm Johannsen in 1909, based on the ideas of Mendel, de Vries, Correns and Tschermak. Their original theoretical definition of the gene being 'the smallest unit of genetic inheritance' remains the cornerstone of our understanding; however, the definition has grown with our knowledge of molecular

## Medical/Health Problems

- Hypothyroid
- ↓ Decreased growth 3 mos – 3 yrs
- ↓ Immune function
- ↓ Muscle tone
- ↓ Decreased digestive competence
- ↓ Nutrient absorption
- ↓ Stomach acid
- Nutrient deficiencies
- Early onset Alzheimer's
- Gout
- ↑ Celiac disease
- ↑ Allergies
- ↑ Food intolerances
- ↑ Middle ear problems
- ↑ Diabetes type 2
- ↑ Increased chemical sensitivity
- Macrocytic Anemia
- Joint laxity
- Constipation
- Early menopause
- Estrogen deficiency

Henry Osiecki, "The Physician's Handbook of Clinical Nutrition" Seventh Edition, AG Publ

## Biochemical- Physiological

- ↓ Thyroid hormone
- ↓ Serine, methionine,
- ↓ Carnitine
- ↓ Thymus, T cell function
- ↓ IgG, complement C3 C4, interferon, neutrophil function
- ↓ Functional folate, methylation cycle
- ↓ DNA repair
- ↓ A, B12, C, magnesium, choline,
- ↓ Selenium (cofactor glutathione, thyroid)
- ↓ Zinc (cofactor 100's of enzymes)
- ↑ Oxidative stress
- ↑ Thyroid antibodies
- ↑ Copper, cysteine, phenylalanine
- ↑ Purine metabolism
- Antibodies- bile duct, parietal cell (stomach)
- ↑ Sympathetic response= HBP, arrhythmia, CV
- Imbalance cAMP/cGMP cellular 2<sup>nd</sup> messengers= problem w cell communication

Henry Osiecki, "The Physician's Handbook of Clinical Nutrition" Seventh Edition, AG Publ

Proc. Natl. Acad. Sci. USA  
Vol. 78, No. 1, pp. 574-578, January 1981  
Medical Sciences

### Can nutritional supplements help mentally retarded children? An exploratory study

(genotrophic disease/nutrition/Down syndrome/megavitamin)

RUTH F. HARRELL<sup>1</sup>, RUTH H. CAPP<sup>1</sup>, DONALD R. DAVIS<sup>2</sup>, JULIUS PEERLESS<sup>3</sup>, AND LEONARD R. RAVITZ<sup>4</sup>

Vitamin A palmitate	15,000 IU*
Vitamin D (cholecalciferol)	300 IU
Thiamin mononitrate	300 mg
Riboflavin	200 mg

average IQ increase of at least 10.2, a highly significant gain ( $P < 0.001$ ). Three of the five subjects who were given supplements for both periods showed additional IQ gains during the second 4 months. Three of four children with Down syndrome gained between 10 and 25 units in IQ and also showed physical changes toward normal. Other evidence suggests that the supplement im-

Zinc (oxide)	30 mg
Manganese (gluconate)	3 mg
Copper (gluconate)	1.75 mg
Iron (ferrous fumarate)	7.5 mg
Calcium phosphate (CaHPO <sub>4</sub> )	37.5 mg
Iodide (KI)	0.15 mg

### Zinc and Selenium "significantly lowered" Copper, Copper/zinc ration "elevated"

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All rights of any nature whatsoever reserved.  
0896-8610/96/0002-0130

#### Changed Serum Trace Element Profile in Down's Syndrome

JANA KADAROVÁ,<sup>1</sup> ALEXANDER MADARIČ<sup>2</sup>

MARA ŠESTOVÁ, AND EDIL GINTER<sup>2</sup>  
<sup>1</sup>Institute of Preventive and Clinical Medicine, Limbova 14,  
833 01 Bratislava, Slovak Republic

Received August 23, 1995; Accepted September 18, 1995

#### ABSTRACT

Being cofactors of important antioxidant enzymes superoxide dismutase (SOD) and glutathione peroxidase (GPx), which are significantly modified in Down's syndrome (Down's 21), serum levels of micronutrient elements zinc, copper, and selenium and of macroelement magnesium are reported in 18 subjects with Down's syndrome (DS) and their respective well age- and sex-matched controls. Serum zinc and selenium levels were significantly lowered in DS subjects, whereas copper levels were elevated. Consequently, a marked increase (49%) of the copper/zinc ratio in DS patients was observed. There were no differences in serum levels of magnesium between DS and control subjects.

**Index Entries:** Down's syndrome; zinc; copper; copper/zinc ratio; selenium; magnesium; serum levels.

### Is zinc deficiency a cause of subclinical hypothyroidism in DS

- The aim of this study was to evaluate if, in Down Syndrome patients, zinc therapy could improve thyroid function
- 52 patients studied, high incidence of subclinical hypothyroidism - 30%
- More significantly, patients with low zinc levels treated with zinc supplementation improved thyroid function, thus reducing the incidence of subclinical hypothyroidism.

Annals of Genetics 1990;33(1):9-15

The Egyptian Journal of Medical Human Genetics, Vol. 5, (2): 19-26, Nov. 2004

### Endocrinal Deficits in Down Syndrome Patients with Zinc Deficiency

Najat A. Al-Awadi, Rezk L. Al-Naggar, Sadika A. Al-Awadi

Kuwait Medical Genetic Center (KMGC) West Salmiya Health Clinic, Ministry of Health

Overt and subclinical hypothyroidisms are the most common endocrinal deficits in patients with Down syndrome. Hypozincemia in DS patients is related to some endocrinal and immunological functions. Zinc deficiency has been found to impair immune response and growth rate. The aim of this study is to evaluate the role of zinc in impaired thyroid function and GH values ( $p < 0.05$ ). There was an improvement of thyroid function after zinc supplementation for 6 months. TSH levels became  $2.72 \pm 1.64 \text{ mIU/L}$  vs.  $6.2 \pm 4.02 \text{ mIU/L}$  before therapy. Our study shows that zinc deficiency has a remarkable effect on the thyroid function and the growth hormone in patients with DS. We recommend that our patients need further cycles of zinc and normozincemic patients has been found ( $p < 0.001$ ). Low GH values were recorded in hypozincemic patients with OS compared to normozincemic patients. On the other hand IGF-1 levels have been found to be high in hypozincemic patients ( $p < 0.001$ ). There was also a significant association between the low zinc levels and impaired thyroid function and GH values ( $p < 0.05$ ). There was an improvement of

**PEDIATRICS**  
OFFICIAL JOURNAL OF THE AMERICAN ACADEMY OF PEDIATRICS

### Fetal Down Syndrome Brains Exhibit Aberrant Levels of Neurotransmitters Critical for Normal Brain Development

Nigel Whittle, MAppSci<sup>1</sup>, Simone B. Sartori, PhD<sup>2</sup>, Mara Dierssen, MD, PhD<sup>3</sup>, Gert Lubec, MD<sup>4</sup>, Nicolas Singewald, PhD<sup>5</sup>

...nubutyric acid, and taurine) and monoamines (serotonin, noradrenaline, and dopamine) act as developmental signals or regulators. In subjects with Down syndrome, dysfunctional brain development is evident from birth as reduction in ...

**RESULTS:** Fetal Down syndrome brains showed **reductions in the levels of serotonin,  $\gamma$ -aminobutyric acid, taurine, and dopamine in the frontal cortex.** No alteration in the levels of arginine, aspartate, glutamine, glutamate, glycine, histidine, serine, or noradrenaline was observed.

**CONCLUSIONS:** **Serotonin,  $\gamma$ -aminobutyric acid, taurine, and dopamine are critical for the acquisition of brain morphologic features, neuronal and glia proliferation, and synapse formation.** The detected reductions in the levels of these neurotransmitters may indicate potential mechanisms for the observed dysfunctional neuronal gestational age of ~20 weeks versus age-matched control aborted fetuses.

PEDIATRICS Volume 120, Number 6, December 2007

RESEARCH ARTICLE

### Oxidative stress occurs early in Down syndrome pregnancy: A redox proteomics analysis of amniotic fluid

...andra Giorgi<sup>1</sup>, ...idino<sup>2</sup>, ...he ...vo ...is an early ...terious DS ...sm (zinc- ...ion (comple ...the clinical ...ly DS

Trisomy 21  
↓  
Oxidative stress  
↓  
Protein oxidation  
↓  
IMPAIRMENT  
Fe Homeostasis, Acute phase response, Fat metabolism, Coagulation  
↓  
DS phenotype  
Atherosclerosis, Obesity, Neuronal injury, Inflammation, Thrombocytopenia

Proteomics Clin. Appl. 2011, 5, 167-178 DOI 10.1002/prca.201000121

PNAS

### Functional genomic analysis of amniotic fluid cell-free mRNA suggests that oxidative stress is significant in Down syndrome fetuses

Donna K. Slonim<sup>1,2</sup>, Keiko Koide<sup>1</sup>, Kirby L. Johnson<sup>1</sup>, Umadevi Tantravahi<sup>1</sup>, Janet M. Cowarf, Zina Jarraf, and Diana W. Bianchi<sup>1,2</sup>

...supernatant samples. **Functional pathway analysis highlighted the importance of oxidative stress, ion transport, and G protein signaling in the DS fetuses.** Further evidence supporting these results suggests that there are secondary **adverse consequences of DS evident in the second trimester, leading to testable hypotheses about possible antenatal therapy for DS.**

PNAS | June 9, 2009 | vol. 106 | no. 23

PNAS

### Functional genomic analysis of amniotic fluid cell-free mRNA suggests that oxidative stress is significant in Down syndrome fetuses

Donna K. Slonim<sup>1,2</sup>, Keiko Koide<sup>1</sup>, Kirby L. Johnson<sup>1</sup>, Umadevi Tantravahi<sup>1</sup>, Janet M. Cowarf, Zina Jarraf, and Diana W. Bianchi<sup>1,2</sup>

...35, 36), we observe that the **trisomic genes generally showed increased expression in DS, with average up-regulation of nearly 1.5-fold (Fig. 2.4).** However, this effect is **highly variable, with nearly a third of the trisomic genes actually down-regulated on average (but few significantly so).** We see **widespread differential expression of genes from the other diploid chromosomes (Fig. ... expression levels of the 409 individual genes not located on chromosome 21 completely separates the DS samples from the controls (as seen in Fig. 1).** In contrast, Mao et al. (30), who

PNAS | June 9, 2009 | vol. 106 | no. 23

Review Article

### Oxidative Stress and Down Syndrome: A Route toward Alzheimer-Like Dementia

Trisomy 21  
↓  
Early oxidative stress in Down syndrome  
↓  
↑ Cellular stress response → Adaptation, compensatory mechanisms → Increased DS lifespan  
↓  
↑ DNA damage, ↑ protein oxidation, ↑ lipid peroxidation → Accumulation of oxidative damage → Severe DS phenotypes  
↑  
DS phenotypes: AD pathology, Heart defects, Diabetes, Leukemia, Atherosclerosis

Current Gerontology and Geriatrics Research  
Volume 2012, Article ID 724904, 10 pages  
doi:10.1155/2012/724904

CHAPTER 22

### OXIDATIVE STRESS AND MITOCHONDRIAL DYSFUNCTION IN DOWN SYNDROME

Giovanni Pagano\* and Giuseppe Castello

...course of Alzheimer disease (AD), due to DS progression toward dementia and amyloid plaques reminiscent of AD clinical course. Moreover, DS represents one

**As an essential link to oxidative stress, mitochondrial dysfunctions are observed whenever redox imbalances occur, due to the main roles of mitochondria in oxygen**

...well documented hallmark of DS phenotype is represented by a set of immunologic defects encompassing a number of B and T-cell functions and cytokine production, together prompting a proinflammatory state. In turn, this condition can be directly interrelated with an in vivo prooxidant state.

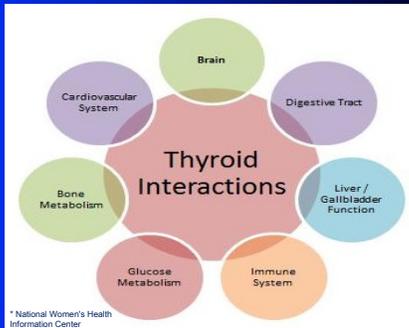
...mice, to be reviewed in this chapter. Together, **in vivo alterations of mitochondrial function are consistent with a prooxidant state as a phenotypic hallmark in DS.**

Neurodegenerative Diseases, edited by Shamim I. Ahmad.  
©2012 Landes Bioscience and Springer Science+Business Media



## Thyroid Gland: Metabolic Powerhouse

Treat the person – not the numbers

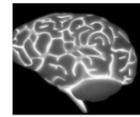


Thyroid hormones regulate stem cells, neurogenesis, migration, neuronal interconnections, synaptogenesis & brain plasticity

### Thyroid-Brain Crosstalk

#### Thyroid Hormones Support

- Neurogenesis
- Precursor cell proliferation
- neuronal migration
- Dendritic and axonal growth
- Myelination
- Synaptogenesis



Promote Plasticity

Promote Healthy Brain Function

Decrease Neurodegeneration

## Hypothyroidism:

Slow reflexes & heart rate,  
enlarged neck,  
puffy face,  
swollen hands & feet



39

## Hypothyroidism: low basal temp

<http://www.stoophthyroidmadness.com/temperature/>

constipation,  
dry skin,  
slow maturation  
slow thinking

40

**DS/T21 Thyroid dysfunction\***  
**N=136**, "overall hypothyroidism...  
~30-40% & may reach 80-90% in  
early childhood.

Jordan J. C. et al. Thyroid disorders in Down's syndrome. *Int J Down Syndrome*. 2005; 9(3): 34-39.  
Rooney S, Walsh E. Prevalence of abnormal thyroid function in a Down's syndrome population. *Ir J Med Sci* 1997; 166: 80-82.  
Toledo C, et al. Anomalies of thyroid function in children with Down's syndrome. *Arch Pediatr* 1997; 4: 116-120.

**Treat the person not the lab numbers.**

41

## Basic Good Diet

- Eat as nature intended-real food not food like, processed food, remember 4 rules:
  - Eat food that will rot, before they do
  - Eat foods available > 2,000 years ago
  - Eat low on food chain
  - Don't eat what you can't pronounce
- Choose Organic when possible
- Try Juicing, Blending
- Gluten Free Casein free, consider strict elimination
- Avoid fast & processed foods, sugar, additives
- Oil Change to healthy fats- olive, grape seed, coconut



## Nutrients

- Adequate amounts essential to support the body's complex metabolic pathways
- Deficiency in micronutrients predisposes to toxicity by decrease ability to eliminate toxins
- Sufficiency of buffering minerals, primarily magnesium & zinc, facilitate toxic metal excretion and block absorption
- First morning urine pH in the 6.5-7.5 range suggesting adequate cellular buffering capacity

## Alkaline Diet

Food & Chemical Effects on Acid / Alkaline Body Chemical Balance-									
Most Alkaline	More Alkaline	Low Alkaline	Lowest Alkaline	Food Category	Most Acid	Low Acid	More Acid	Most Acid	Food Category
Leafy Greens	Green Vegetables	Green Vegetables	Green Vegetables	Leafy Greens	Meat	Meat	Meat	Meat	Meat
Broccoli	Spinach	Kale	Chard	Leafy Greens	Beef	Pork	Chicken	Turkey	Meat
Asparagus	Cauliflower	Brussels Sprouts	Cabbage	Leafy Greens	Wheat	Rice	Grains	Grains	Grains
Green Beans	Peas	Lentils	Beans	Leafy Greens	Apples	Oranges	Lemons	Limes	Fruit
Avocado	Olives	Almonds	Walnuts	Leafy Greens	Wheat	Rice	Grains	Grains	Grains
... (many more rows) ...	...	...	...	...	...	...	...	...	...

## Industrial and Agricultural Chemicals

- Have increased at an almost exponential rate for the past 50 years, ~ 10 new chemicals, introduced each day
- EPA estimates that 87,000 chemicals are in use today
- Plastics industry has grown at the rate of 6-12% per year since mid-1940s
- Annual production in U.S. reaching 85 billion pounds > 338 pounds per person, per year

Colburn Environ. Health Perspec. 2004; 112(9)

## Increased Vulnerability Children

- Increased due to both rapid development and incomplete defense systems:
  - A developing child's chemical exposures are greater pound-for-pound than those of adults
  - An immature, porous blood-brain barrier allows greater chemical exposures to the developing brain
  - lower levels of chemical-binding proteins, allowing more chemicals to reach "target organs"
- Rapidly developing organ systems- more vulnerable to damage from chemical exposures
- Detox capacity not fully developed

Environmental Working Group July 2005

## Synergistic Toxicity

- Pb and stress
- Pesticides
- Paraquat and maneb - relative risk of Parkinson's Disease
- Polybrominated diphenylethers (PBDEs) and PCBs
- Heavy metals

Cory-Slechta DA Neurotoxicology 2005 Feb.  
Eriksson et al Toxicol Sci 2006 Dec; 94(2): 302-9

## Toxic Metal Sources

- Lead: toys, vinyl, food, lead paint before 70's
- Mercury- amalgams, vaccines, breathing- power plants incinerators
- Aluminum- cookware, deodorants, baking soda, cans
- Arsenic- chicken, treated wood
- Fluoride- toothpaste, water, treatments

## Plastics

- No phthalates, bisphenols, pvc wraps, microwaving
- 1,2,4,5 best choices, 1&5 can contain phthalates
- Glass always preferable
- [www.healthobservatory.org/library.cfm?refid=77083](http://www.healthobservatory.org/library.cfm?refid=77083) info on safer plastics

## Biomedical Interventions Support Detox

- **Micronutrients**
- **Copper/Zinc**
- **GI Support**
- **Antioxidants**
- **Sulfur**
- **Methylation Support**

## Treatment of High Cu/Zinc Ratios

- Zinc supplement, need to monitor with blood levels, sometime need lots
- Adequate Selenium, molybdenum
- Support Glutathione
- B6/Magnesium
- Optimize Vitamin C dose
- Avoid Sources of Copper
  - Tap water (Cu pipes)
  - Swimming pools and hot tubs (Cu algacide)
  - Chocolate, Carob, Soy, Shellfish, Liver
- Avoid red/yellow dyes and MSG deplete Zn

## Antioxidants

- Vitamins A,C, and E, carotenoids, selenium
- High dose C to bowel tolerance, potential benefits:
  - toxic metal & pollutant elimination
  - improved immune function
  - increased bile flow
  - increased carnitine production
  - adrenal support
  - improved intestinal ecology
  - stabilization of tetrahydrobiopterin
  - support neurotransmitter production
  - increases glutathione levels
  - keeps folate in active form
  - increases iron absorption
  - safe and effective laxative at high dose

## Gastrointestinal Support

- Treat Constipation/diarrhea
- Digestive enzymes
- Pre and probiotics
- Treatment guided by symptoms and lab
- Antifungals, antibacterials, antiparasite treatment- prescription meds can have advantage
- Specific Carbohydrate, body ecology diet, elimination diet

## Maintain Methylation Pathways

- Critical for detoxification
- Impaired by toxicants
- Supportive nutrients:
  - B-12- hydroxy, methyl, adenosyl
  - TMG
  - DMG
  - Folinic acid, methylfolate
  - Glutathione
  - Ascorbate (vitamin C)
  - N-acetyl cysteine.

## Detoxification Requires:

- Energy (ATP), energetic burden
- Nutritional support, fiber, sulfur sources
- Antioxidants
- Optimal Copper/Zinc balance
- Vitamin /minerals, co-factors
- Healthy gastrointestinal lining
- Soft stool 2x/day optimal essential to treat constipation (if it is present)
- Good liver function
- Methylation support
- Sauna, epsom salt baths (support)

## Curcumin Protective Against Lead Toxicity

- Curcumin treated animals had more glutathione and less oxidized proteins in the hippocampus compared to animal with similar lead exposure w/o curcumin
- Retained spatial reference memory (i.e., water maze)
- Curcumin is neuroprotective against heavy metal induced neurotoxicity

Dairam et al J Agric Food Chem. 2007; 55(3), 1039-1044\

## Clean Up Environment

- **Water**
- **Air**
- **Personal Care Products**
- **Clothing/Laundry**
- **House and Garden**
- **Toxic metals**
- **Plastics**

### Pure Water

- Pure water is essential; dehydration hinders the body's ability to eliminate waste and keep resilient; pesticide levels, heavy metals, hormone residues, volatile organic compounds fluoride more important than bacteria

### Clean Air

- Fresh air, outdoors
- Use fresh flowers, natural oils
- **Avoid** air fresheners, sprays, perfumes, cleaning agents, new paints, new carpets, flea treatments, insecticides, furniture chemicals
- Use HEPA and furnace filters
- Check for CO output in gas appliances
- Be aware humidity (optimal 45-50%) mold

## Personal Care Products

- Nearly all deodorants contain aluminum, which is readily absorbed
- Perfumes and cosmetics can contain multiple potential toxins.
- Organic cotton clothes, most fabrics have chemicals
- Be aware fungicides, flamer retardants bedding, mattress

### Clothing and Laundry

- Detergents and fabric conditioners are common allergens, contain multiple toxins.
- Choose natural products.

## House and Garden

- Cleaning products- natural, biodegradeable
- Minimal perfumes, chemicals
- Flame retardants in clothing, fungicides in bedding
- Persistent organic pollutants in carpets
- Herbicide & pesticides free
- Do not allow children or pets on treated lawns for at least 3 weeks after spraying
- Arsenic in treated wood
- Chlorine from pools

## Toxic Metal Sources

- Lead: toys, vinyl, food, lead paint before 70's
- Mercury- amalgams, vaccines, breathing- power plants incinerators
- Aluminum- cookware, deodorants, baking soda, cans, foil
- Fluoride- tootpaste, water, treatments

### Plastics

- No phthalates, bisphenols, pvc wraps, microwaving
- 1,2,4,5 best choices, 1&5 can contain phthalates
- Glass always preferable
- [www.healthobservatory.org/library.cfm?refid=77083](http://www.healthobservatory.org/library.cfm?refid=77083) info on safer plastics

## My Approach to Treatment

- Comprehensive, individualized functional, integrative
- Medical therapy based on predictive, sensitive, specific lab testing
- Monitor clinical outcome
- Understand biochemical imbalances to prioritize low risk, high gain treatments.
- Evaluate environmental toxicity
- Engage family, "Doctor Mom & Dad" as colleagues to determine what works best
- Not a single cause,-complex, need to think deeply about causes and consequences in Trisomy 21

## Multidimensional Treatment Individualized to

- Assess and correct biochemical imbalance
- Assess amino acid and neurotransmitter sufficiency and provide repletion
- Support vitamin, mineral, and enzyme metabolism
- Support detoxification
- Repair and optimize intestinal function
- Provide immune augmentation
- Provide hormone replacement therapy, including vitamin D, when necessary.
- Physical, occupational therapy
- Education on additional supportive therapies

## Options for Initial Lab Testing

- Thyroid:
  - Free T4 T3, reverse T3
  - Antibodies TPO, thyroglobulin
- Basal Temp
- CBC
- Iron, iron binding
- Ferritin
- Vitamin D
- Vitamin A
- Copper/zinc
- Uric Acid
- Selenium
- Iodine
- Lipid panel
- Amino acids
- Organic acids which
- Essential Fatty Acids
- Heavy metals
- Celiac
- Stool analysis
- Immune function
- Oxidative stress markers
- Ammonia

## Biomedical Treatment Options

- Multivitamin w B complex, trace min
- Omega w > DHA/EPA
- Buffered Ascorbate (vit c)
- Vitamin D
- CoQ10
- B6
- Magnesium
- Folate
- Flavanoids/flavinols
- Choline
- Carnitine
- TMG/DMG
- EGCG
- Targeted amino acids
- Zinc
- Selenium
- Probiotics
- Digestive enzymes
- Glutamine
- Alpha keto glutarate
- Uric acid
- Homocysteine
- Curcumin
- Ginkgo Biloba
- Alpha lipoic acid
- Melatonin

## Potential Benefits Biomedical Rx

- Promote cognitive function
- Promote growth and development
- Improve thyroid function
- Balance neurotransmitters
- Improve health and immune function
- Support Methylation, folic acid cycle
- Increase glutathione
- Decrease oxidative stress
- Decrease total heavy metal burden
- Decrease pesticides, pollutants
- Maintain function, decrease disability through lifespan

J Neurodev Disord (2009) 1:150–157  
DOI 10.1007/s11689-009-9018-7

### Adult reversal of cognitive phenotypes in neurodevelopmental disorders

Akino J. Silva · Dan Ehlinger

#### A ray of hope: reversing cognitive deficits in adults

There are now a number of compelling examples, in mice, of **rescue of neurocognitive deficits associated with developmental disorders in adults**, including NFI, TSC, Down syndrome, Rubinstein-Taybi syndrome (RTS), Fragile X syndrome (FXS), and Rett syndrome.

**possible to reverse neurodevelopmental disorders in adults;**

**adults could, in special cases, dramatically improve the mental disorders, including autism, since they suggest that it may be possible to treat or even cure them in adults.**

## Suggestions

- Be Grateful
- Love your children like the whole world depended on it.
- Usually, the sooner you begin, the better the results.
- Ask yourself: Should I wait for orthodox medicine to find a solution?
- Work in progress, don't stop looking for answers and asking questions
- Keep the faith many families see significant benefit

If you want to go fast go alone. If you want to go far, go together.

African Proverb

Together, we know enough to begin individualized treatment for trisomy 21.  
If not now, when?

Thank you!!!

Norm Schwartz MD  
10602 N Pt Washington Rd  
Suite 101  
Mequon WI 53092  
262 240-0133

## Trisomy 21 Biomedical Resources

[Blog, Down Syndrome: A Day to Day Guide](#)  
[Nuper Down Syndrome](#)  
[gotdownsyndrome.blogspot.com](#)  
[riverbendds.org/index.htm](#)  
[changingmindsfoundation.org/documents/links.html](#)

## Environmental Groups

- Save our kids, heal our planet [www.sokhop.com](http://www.sokhop.com)
- Environmental Working Group [www.ewg.org](http://www.ewg.org)
- Environmental Health Perspectives Journal [www.ehponline.org](http://www.ehponline.org)
- Collaborative on Health and the Environment: [www.cheforhealth.org](http://www.cheforhealth.org)
- Toxicology Encyclopedia [www.toxipedia.org](http://www.toxipedia.org)
- Institute for Children's Environmental Health: [www.iceh.org](http://www.iceh.org)
- We Can Solve the Climate Crisis: [www.wecansolveit.org](http://www.wecansolveit.org)
- Environmental Health News: [www.environmentalhealthnews.org](http://www.environmentalhealthnews.org)
- Our Stolen Future: [www.ourstolenfuture.org](http://www.ourstolenfuture.org)
- Pesticide Action Network: [www.panna.org](http://www.panna.org)
- Coming Clean: [www.chemicalbodyburden.org](http://www.chemicalbodyburden.org)
- Natural Resources Defense Council: [www.nrdc.org/health](http://www.nrdc.org/health)
- Beyond Pesticides: [www.beyondpesticides.org](http://www.beyondpesticides.org)
- Greenpeace Chemical Kitchen: [www.greenpeace.org](http://www.greenpeace.org)
- Alliance for a Healthy Tomorrow [www.healthytomorrow.org](http://www.healthytomorrow.org)

The information on the following slides provided courtesy of Stu Freedenfeld MD  
Please see his excellent web site: [www.StocktonFP.com](http://www.StocktonFP.com)  
for more details and information

## Household and Personal Care

[www.ewg.org/reports/skindeep2/index.php](http://www.ewg.org/reports/skindeep2/index.php) Searchable index for product safety information  
[www.lookinggoodorganics.com](http://www.lookinggoodorganics.com) Personal and house care  
[www.Earthessentials.com](http://www.Earthessentials.com) Skin , personal care  
[www.ecos.com](http://www.ecos.com) [www.ecover.com](http://www.ecover.com) Cleaning products  
[www.seventhgeneration.com](http://www.seventhgeneration.com) Paper, cleaning products  
[www.needs.com](http://www.needs.com) Household and personal care  
[www.acmynet.org](http://www.acmynet.org) Source of non-toxic art supplies  
<http://www.thesofflending.com> Baby bottles, sippy cups  
<http://www.onestepahead.com> Toys, child needs  
<http://www.greenhome.com> Clothing and bedding  
[www.organicmattressstore.com](http://www.organicmattressstore.com) Bedding and mattresses

## Green Lawns- Safe Neighborhoods

Insect control: Insecticidal soap, diatomaceous earth, and neem products. Insects are vital and killing agents are not selective.

Weed control: Corn gluten, hot water and vinegar, pull the weeds, or just leave them be and relax

Fungicide alternatives: Sulfur, baking soda, certain copper products and avoid over watering.

## Green Lawns-Safe Neighborhoods

[www.gardensalive.com](http://www.gardensalive.com) Lawn and garden products Also a resource for garden questions.

[www.WHY.org/91fm/vbyg](http://www.WHY.org/91fm/vbyg) non-toxic solutions to garden and indoor plant questions.

[www.pesticides.org](http://www.pesticides.org) Information on dangers of pesticides and safe alternatives

<http://www.beyondpesticides.org/gateway/index.htm> Safety information of specific pesticides

## Natural Insect Repellants

Neem oil can be taken orally as a mosquito repellent. (Ayush Herbs, [www.ayush.com](http://www.ayush.com) )

Thiamin 100mg/d may deter mosquitoes.

Topicals: 2% soybean oil, Vick's Vapo Rub, pure vanilla extract (1:1 with water), cinnamon oil, oil of lemon eucalyptus.

Enzyme shampoos to get rid of fleas and lice and spray house with 50/50 vinegar and water

[www.LiceBgone.com](http://www.LiceBgone.com) enzyme products for lice, scabies and crabs

## Natural Insect Repellants

Cockroaches: spread bay leaves and sprinkle a fine dusting of baking soda and boric acid

Ants: Sprinkle black pepper, chili powder or chalk and brush lavender and olive oil on trails.

Moths: Cedar chips and lavender oil

Mice and rats: Lavender oil, cedar oil or camphor will repel them.

Trade products include Ambermin's Natural Source Bug Repellent, Gone (insect detergent spray), All

Terrain Herbal Armor Insect Repellent, Buzz Away, BiteBlock and Outdoor Herbal Spray

## Environmentally Friendly Products

[www.hangersdrycleaners.com](http://www.hangersdrycleaners.com) green dry cleaning

[www.greenearthcleaning.com](http://www.greenearthcleaning.com) green dry cleaning

[www.earth911.org](http://www.earth911.org) local recycling centers including fluorescents

[www.lamprecycling.com](http://www.lamprecycling.com) info mailing used fluorescent bulbs

Note: Home Depot will recycle fluorescent bulbs

## Whole Foods

[www.ams.usda.gov/farmersmarkets](http://www.ams.usda.gov/farmersmarkets) national listing of farmers' markets.

[www.localharvest.org](http://www.localharvest.org) local sources of sustainably grown food

[www.catwellguide.org](http://www.catwellguide.org) online directory of sustainably raised meat, poultry, dairy, and eggs, US & Canada

<http://www.nal.usda.gov/afsic/csa/> Community Supported Agriculture programs (CSA's)

[www.westonaprice.org](http://www.westonaprice.org) Weston A. Price Foundation

[www.foodroutes.org](http://www.foodroutes.org) interactive web site to find local farmers, markets, CSA's near you

## Whole Foods

[www.bostonveg.org](http://www.bostonveg.org) vegetarian basics  
[www.realmilk.com](http://www.realmilk.com) info on raw milk:  
[www.westonaprice.org](http://www.westonaprice.org)  
[www.eatwellguide.org](http://www.eatwellguide.org) healthy food markets,  
farms and restaurants by zip code  
[www.localharvest.org](http://www.localharvest.org) local organic farms  
farmers markets and restaurants  
[www.ams.usda.gov/farmersmarkets](http://www.ams.usda.gov/farmersmarkets)  
[www.foodnews.org](http://www.foodnews.org) guide to Pesticides in  
Produce

## Water

EPA Safe Drinking Water Hotline (800-426-4791)  
Information water safety, contaminants, qualified labs  
<http://www.nrdc.org/water/drinking/uscities/contents.asp>  
reports contaminants in major cities  
[www.safe-drinkingwater.org/pdf/makesense.pdf](http://www.safe-drinkingwater.org/pdf/makesense.pdf) fact sheets  
on common contaminants  
[http://www.psr.org/site/DocServer/  
Drinking\\_Water\\_and\\_Disease\\_Primer.pdf?docID=559](http://www.psr.org/site/DocServer/Drinking_Water_and_Disease_Primer.pdf?docID=559)  
Physicians for Social Responsibility info water and health  
Laboratory testing:  
<http://www.doctorsdata.com>  
<http://www.h2otest.com/other.html>  
<http://www.ntllabs.com/homeowner/index.html>

## Resources

[http://www.care2.com/greenliving/make-your-own-  
non-toxic-cleaning-kit.html](http://www.care2.com/greenliving/make-your-own-non-toxic-cleaning-kit.html) non-toxic cleaning kit  
[http://www.care2.com/greenliving/make-your-own-  
non-toxic-cleaning-kit.html](http://www.care2.com/greenliving/make-your-own-non-toxic-cleaning-kit.html) home made cleaning  
products  
[www.nsf.org](http://www.nsf.org) tests on water filter's performance  
[www.nrdc.org/water/drinking/gfilters.asp](http://www.nrdc.org/water/drinking/gfilters.asp) guide to  
common water filters  
[www.searchawarf.org](http://www.searchawarf.org) info on filtration  
technologies  
[www.nrdc.org/water/drinking/bw/appa.asp](http://www.nrdc.org/water/drinking/bw/appa.asp) tests of  
several hundred bottled water brands

## Resources

[www.ucsusa.org/publications/greentips/](http://www.ucsusa.org/publications/greentips/) Union of Concerned  
Scientists  
[www.environmentalobservatory.org/library/cfm?refid=77083](http://www.environmentalobservatory.org/library/cfm?refid=77083)  
info on plastics  
[www.ozoneaction.org](http://www.ozoneaction.org) Info persistent organic pollutants  
[www.greenbuildingsupply.com](http://www.greenbuildingsupply.com) [www.greenhomeguide.com](http://www.greenhomeguide.com)  
[www.hardwoodinstaller.com/](http://www.hardwoodinstaller.com/) [www.hardwoodscene.com/](http://www.hardwoodscene.com/)  
[www.hardwoodinstaller.com/finishes-water.htm](http://www.hardwoodinstaller.com/finishes-water.htm) info on green  
building supplies  
[www.nurturedfamily.com/catfuzzibunz.aspx](http://www.nurturedfamily.com/catfuzzibunz.aspx)  
[www.diaperpin.com](http://www.diaperpin.com) reusable diapers  
[www.leadcheck.com](http://www.leadcheck.com) test kits for lead, cadmium, mercury,  
arsenic, nickel and chromium