BEAT AUTISM NOW (BAN)

LOGICALLY, EFFECTIVELY AND INEXPENSIVELY

BOOKLET 1

GENERAL INFORMATION AND

THE GENETIC CONNECTION: WHY SOME INDIVIDUALS ARE MORE AFFECTED BY TOXINS THAN OTHERS

E.BLAUROCK-BUSCH PHD
The Genetic Connection: Why some individuals are more affected by toxins than others .......................... 1
Introduction .................................................................................................................................................. 3
General Information about ASD (Autism spectrum disorder) ................................................................. 5
  International Statistics ................................................................................................................................. 5
  US Autism Statistics ................................................................................................................................. 6
Genetics ..................................................................................................................................................... 6
Toxins and pollutants .................................................................................................................................. 6
Vaccination and viruses ............................................................................................................................... 6
Contributing Factors: .................................................................................................................................. 7
How is autism diagnosed? ............................................................................................................................ 7
More Literature .......................................................................................................................................... 8
The Genetic Connection: ............................................................................................................................ 10
Why some individuals are more affected by toxins than others ............................................................... 10
The detoxification pathway .......................................................................................................................... 11
Phase 1 ....................................................................................................................................................... 11
  The Cytochrome P450, specifically the CYT 450 1A1 ............................................................................... 11
  Superoxide Dismutase 1 (SOD1) ............................................................................................................ 11
  Superoxide Dismutase2 (SOD2) ............................................................................................................. 12
Phase 2 Enzymes ....................................................................................................................................... 12
  Glutathion-S-Transferase M1 (GSTM1) .................................................................................................... 12
  Glutathion-S-Transferase T1 (GSTT1) ....................................................................................................... 12
  Glutathion S-Transferase P1 (GSTP1) ....................................................................................................... 13
  N-Acetyltransferase 2 (NAT2) .................................................................................................................... 13
  Apolipopotein E (Apo E) .......................................................................................................................... 13
  Apo E Research ....................................................................................................................................... 14
Summary: Why genetic testing .................................................................................................................... 14
How to test? .................................................................................................................................................. 15
More Literature .......................................................................................................................................... 15
About the author: ........................................................................................................................................ 17
INTRODUCTION

While attending some workshops and meetings, attended by doctors and other therapists specializing in the treatment of autism, I listened to speeches by ‘experts’. Most had good intentions, but some were so clearly commerce-motivated and provided distorted information, I knew I had to speak up. I did then and I do now with this informational handbook.

Some say I have commercial interests as well, and they are partially right. While I continue to work for the laboratory I founded in 1975, I am no longer the owner. My daughter is, and I support her as much as I can, engaging myself into research projects, publishing papers, lecturing to professional groups and occasionally to lay people such as the DAN group in Barcelona or Tenerife.

Money has never been a motivating force in my life. Excellence of service, devotion to good health and honesty is.

Most of all, I am a mother and grandmother, and I am thankful for having a healthy family. As Schopenhauer said, ‘health is not everything, but without health everything is nothing.”

My daughter, now 39 years of age, might have turned autistic or sick in other ways had I continued with thiomersal-containing vaccinations. I am convinced of that. My husband and I were educated enough to see the danger when she reacted violently after her first vaccination. We did not know then that I had passed on mercury in utero, unknowingly intoxicating my unborn with all the mercury I had stored as a result of exposure at the workplace and overzealous dental treatments. A mineral analysis of hair taken during our daughter’s first year clearly indicates a severe mercury overexposure. It explained the strange symptoms she displayed at times, the anxiety attacks, the reversed drug reactions, the thyroid problems (mercury is easily stored in thyroid tissue) and food sensitivities. During her younger year, we were heavily into nutritional and chiropractic treatments, all of which helped. Later, after my involvement with IBCMT (International Board of Clinical Metal Toxicology), we stepped up treatment and I must credit Dr. Peter van der Schaar, chairman of IBCMT for his involvement and help. His detoxification treatments reduced her remarkable mercury load. Before she attempted pregnancy, we stepped up the process, hoping to intercept the passing on of mercury from mother to child. We largely succeeded, and I am grateful.

Abortion, pregnancy and the delivery of a child is an effective process of detoxification; the woman detoxifies her system at the expense of the unborn or born.

That I met (and learned to trust) Dr. Van der Schaar changed my life and the life of my family in many ways. I will always be thankful for that.

I am grateful that my personal experiences did not turn into tragedy. I now understand parents of ill children. Their feeling of despair is not foreign to me. It
upsets me when commerce takes advantage of families who are challenged with disadvantaged children. If I don’t stand up for them who does?

The purpose of this first booklet, which will be followed by more to come, is to:

- provide scientific evidence (as it exists at this time) to the cause of Autism
- provide logical explanation regarding the diagnosis of Autism i.e. what are useful diagnostics and what tests are practically useless or meaningless
- how diagnostic results can lead to treatment success

In short, I hope to prevent parents from spending money for useless diagnostics and treatments, give them confidence and protect them from financial vultures. I want them to spend the saved money on education for the child, books or pets, or a holiday that is fun for the entire family.

Since this is an ongoing writing process, based on present knowledge and research, I appreciate any feedback, positive or negative. I will pay attention to comments, consider inclusion but am not interested in commercialism.

I do stand behind the analytical and medical information provided here, not because of its commercial impact but because of its validity as known at this time, and if the information given here improves the well-being of only one child, the effort in compiling it was well worth it.

Last but not least, I am not the ‘inventor’ of new diagnostics or treatments, but I do claim to have analyzed and sorted available information to the best of my knowledge, always having in mind the wellbeing of families who are burdened and blessed with an autistic individual.

Most sincerely,

Eleonore Blaurock-Busch PhD
GENERAL INFORMATION ABOUT ASD (AUTISM SPECTRUM DISORDER)

First identified more than 50 years ago, autism affects half a million people in the UK – tens of millions worldwide - and is considered as one of the most common developmental disorders.

Autism is considered a life-long brain disorder that is normally diagnosed in early childhood.

Autism is a spectrum disorder varying in severity and impact from individual to individual, ranging from those with no speech and severe learning disabilities to people with IQs in the average range who are able to hold down a job or start a family. People with autism may also have unusual patterns of language development, narrow interests and engage in repetitive and sometimes challenging behaviours.

Asperger's Syndrome is a form of autism in which speech development and IQ are normal, but in which social disability can be compounded by depression and mental health problems.

Some people with autism demonstrate significantly challenging behaviours; most need specialist support and care.

This and more information is provided by http://www.autistica.org.uk/about_autism/index.php

INTERNATIONAL STATISTICS

About 1 in 88 children has been identified with an autism spectrum disorder (ASD) according to estimates from CDC's Autism and Developmental Disabilities Monitoring (ADDM) Network.

ASDs are reported to occur in all racial, ethnic, and socioeconomic groups

ASDs are 4 to 5 times more common among boys (1 in 54) than among girls (1 in 252).

Studies in Asia, Europe, and North America have identified individuals with an ASD with an average prevalence of about 1%. A recent study in South Korea reported a prevalence of 2.6%.
US AUTISM STATISTICS

Studies indicate that 1 in 6 children in the U.S. had a developmental disability in 2006-2008, ranging from mild disabilities such as speech and language impairments to serious developmental disabilities, such as intellectual disabilities, cerebral palsy, and autism.

- Recent documents estimate that one out of every 150 infants is becoming autistic.
- 4 out of 5 are male
- 3 out of 4 are thought to be mentally retarded
- 1/3 of those with autism suffer from epilepsy
- Most of the autistic are institutionalized by age 13
- Occurrence has increased 556% during the 1990s, possibly due to an increase in awareness

This and more information is provided by http://www.autism-society.org/about-autism/facts-and-statistics.html

GENETICS

- Children are 25x more likely to have autism if another sibling is autistic
- In identical twins, 75% of the autistic have an affected twin
- A clear genetic predisposition is present, but no consistent chromosomal link
- It is unknown what triggers those that are predisposed

TOXINS AND POLLUTANTS

Brick Township, N.J., a working class town with a well-known toxic landfill was found to have three times the normal autistic occurrence.

Of the toxins involved, mercury seems to be the most common problem.

VACCINATION AND VIRUSES

More evidence points towards childhood vaccinations as a trigger, and the most common vaccine which seems to trigger autism is the MMR (measles, mumps and rubella) vaccine. The reaction is not immediate. The child begins to become autistic
about one month after being vaccinated. Other vaccines which appear to cause problems are the *triple - diphtheria, tetanus, pertussis (whooping cough)*. Also, children have been reported to become autistic after *chicken pox* or other viruses. These particular children tend to have many infections and viruses in their early years, for example, tonsillitis and ear infections.

**CONTRIBUTING FACTORS:**

Nutritional inadequacies

Metabolic problems such as pyroluria

Food and chemical sensitivities

Digestive dysfunctions or genetically based intolerances such as lactose or gluten intolerances.

**HOW IS AUTISM DIAGNOSED?**

The following information is provided by the National Institute of Neurological Disorders and Stroke, contact the Institute's Brain Resources and Information Network (BRAIN) at:

BRAIN
P.O. Box 5801
Bethesda, MD 20824
(800) 352-9424
http://www.ninds.nih.gov

ASD varies widely in severity and symptoms and may go unrecognized, especially in mildly affected children or when it is masked by more debilitating handicaps. Very early indicators that require evaluation by an expert include:

- no babbling or pointing by age 1
- no single words by 16 months or two-word phrases by age 2
- no response to name
- loss of language or social skills
- poor eye contact
- excessive lining up of toys or objects
- no smiling or social responsiveness.

Later indicators include:

- impaired ability to make friends with peers
- impaired ability to initiate or sustain a conversation with others
• absence or impairment of imaginative and social play
• stereotyped, repetitive, or unusual use of language
• restricted patterns of interest that are abnormal in intensity or focus
• preoccupation with certain objects or subjects
• inflexible adherence to specific routines or rituals.

Health care providers will often use a questionnaire or other screening instrument to gather information about a child’s development and behavior. Some screening instruments rely solely on parent observations, while others rely on a combination of parent and doctor observations. If screening instruments indicate the possibility of an ASD, a more comprehensive evaluation is usually indicated.

A comprehensive evaluation requires a multidisciplinary team, including a psychologist, neurologist, psychiatrist, speech therapist, and other professionals who diagnose children with ASDs. The team members will conduct a thorough neurological assessment and in-depth cognitive and language testing. Because hearing problems can cause behaviours that could be mistaken for an ASD, children with delayed speech development should also have their hearing tested.

Children with some symptoms of an ASD but not enough to be diagnosed with classical autism are often diagnosed with PDD-NOS. Children with autistic behaviours but well-developed language skills are often diagnosed with Asperger syndrome. Much rarer are children who may be diagnosed with childhood disintegrative disorder, in which they develop normally and then suddenly deteriorate between the ages of 3 to 10 years and show marked autistic behaviours.

For more information, contact

MORE LITERATURE

PEDIATRICS, October 5, 2009, based on a National Children’s Health Survey done with 78,000 parents in 2007.


THE GENETIC CONNECTION:

WHY SOME INDIVIDUALS ARE MORE AFFECTED BY TOXINS THAN OTHERS

The human body is exposed to a wide array of xenobiotics in one's lifetime, and complex enzymatic mechanisms are genetically available to detoxify these substances. A variety of mechanisms support or impair the body's natural ability to detoxify and scientific literature suggests an association between impaired detoxification and certain diseases.

Missing or nonfunctioning enzyme systems impair biotransformation systems, consequently increasing the need for lifestyle changes, including the avoidance of certain toxins.

An individual’s ability to tolerate toxins depends on how quickly the body can eliminate the toxic burden, and this important biological detoxification mechanism depends on enzyme functions. Certainly, the human body contains multiple enzyme systems involved in the detoxification process, but when one or more important enzymes are missing or are functioning improperly, the body’s ability to eliminate the excess burden is affected. Normal detoxification is impaired.

To put it simply: if a child is missing one or two enzyme systems, the immunization with its significant mercury exposure can overwhelm the young developing brain tissues, resulting in nerve damage. Had all enzyme systems functioned properly, the body would have more quickly eliminated excess mercury, preventing some or most of the damage.

In the United States, case-control studies have reported that an important detoxification enzyme is missing in 23% -41% for those of African descent; 32%-53% for those of Asian descent, 40% -53% for those of Hispanic descent, and 35% -62% for those of European descent. Several population studies have reported the deletion polymorphism among U.S. Caucasi ans as ranging from 48%-57%. Other countries have reported varying frequencies of the deletion polymorphism, and an Iranian study showed that in 31 percent to 38 percent of the population the GSTM1 enzyme was missing. Groups such as Pacific Islanders and Malaysians have a reported frequency of 62%-100%. Other Asian populations have high-reported frequencies of the deletion genotype ranging from 48% -50% for Japanese and 35%-63% for Chinese. A population-based study conducted among Chinese reported a frequency of 51% for the GSTM1 deletion genotype. Two Korean case-control studies found frequencies of 53% and 56% for the GSTM1 deletion genotype.

The above statistics demonstrate that missing enzyme systems are playing a large role in most populations. Since the genetic make-up is inherited, it would make sense to have expecting parents and/or newborns tested, particularly before the child is subjected to metal-containing vaccines or environmental toxins. Genetic testing is relatively inexpensive and in most cases, it has to be done only once in a lifetime.
THE DETOXIFICATION PATHWAY

PHASE 1

While much is known about the role of Phase I enzymes in the metabolism of pharmaceuticals as well as their activation by environmental toxins, the role of Phase I detoxification in clinical practice has received less consideration than the Phase II enzyme systems.

Enzymes involved in the Phase I metabolism are Cytochrome P450, and the SOD Enzymes.

THE CYTOCHROME P450, SPECIFICALLY THE CYT 450 1A1

These enzymes are involved in the metabolism of drugs or exogenous toxins such as chemical solvents or drugs, including steroids. The amount of the CYP enzymes present in the liver reflects their importance in the detoxification process.

The SUPEROXIDE DISMUTASE (SOD) ENZYMES are present in practically all cells and in extracellular fluids. The SODs are considered free radical scavengers, preventing oxidative damage and thus are considered important to delay the aging process. Genetic polymorphism in SOD enzymes and their altered expressions and activities are associated with oxidative DNA damage and an increased cancer risk. (Khan MA, Tania M, Zhang D, Chen H. Antioxidant enzymes and cancer. Chin J Cancer Res 22(2):87-92. 2010)

SOD enzymes contain metal cofactors which can be copper, zinc, manganese or iron. While all people have an abundance of SOD enzymes, deficiency in any of these metals will lower certain SOD levels and function. In other words, a nutritional deficiency in any of these trace elements potentially impairs SOD enzyme function, leading to a disruption in the detoxification pathway.

SUPEROXIDE DISMUTASE 1 (SOD1)

SOD1 is also called the copper/zinc superoxide dismutase or CuZnSOD. It is present in the cytosol, the nucleus and the mitochondria. Its primary function is to act as an antioxidant enzyme, lowering the steady-state concentration of superoxide. High concentrations are found in liver, brain and testes, but also in red blood cells, pancreas and the lung. Inactivity of an SOD enzyme disturbs the cell metabolism.

A copper or zinc deficiency reduces the function and activity of the SOD1 enzyme.
SUPEROXIDE DISMUTASE2 (SOD2)

This gene, also called MnSOD, is a member of the iron/manganese superoxide dismutase family. Mutations in this gene have been associated with idiopathic cardiomyopathy, premature aging, (IDC) sporadic motor neuron disease, and cancer. (NCBI Report. SOD2 superoxide dismutase 2. upd May2011)

SOD-Gene defects have been associated with diseases such as Amyotrophic lateral sclerosis (ALS) (Banzi et al. SOD1 und amyotrophic lateral sclerosis: mutation and oligomerization. PLoS 3/-/2008. NCBI; Furukawa Y et al. Complete loss of post-translational modifications triggers fibrillar aggregation of SOD1 in familial form of ALS. J. Biol.Chem.283/35/2008)

A reduced Phase I Metabolism reduces the detoxification ability of a variety of xenotoxins including the potentially toxic metals.

PHASE 2 ENZYMES

Phase II reactions follow Phase I reactions. Also known as conjugation reactions (e.g. with glutathion or amino acids or sulfonates), the Phase II system is an important defense mechanism against intake of toxins. The Glutation Transferases and N-Acetyltransferase 2 (NAT2) belong to the group of Phase II Enzymes.

A reduced phase II detoxification leads to the accumulation of toxins. Gene variants in the glutathione S-transferases (GST) may lead to poor management of the extremely radical intermediates from the Phase 1 responses and thereby transmit a predisposition for diseases associated with oxidative stress.

The glutathione S-transferases (GSTM1, GSTT1, etc) are one family of enzymes responsible for the detoxification process, particularly mercury and other toxic metal compounds. These enzymes are also known to play a role in the detoxification of polycyclic aromatic hydrocarbons found in tobacco smoke.

GLUTATHION-S-TRANSFERASE M1 (GSTM1)

GSTM1 is produced in the liver. Through conjugation with glutathion, it functions in the detoxification of environmental toxins and products of oxidative stress, electrophilic compounds, including carcinogens and therapeutic drugs.

Individuals with the GSTM1 */0 Genotype do not have this functioning enzymes and are at greater risk to develop carcinomas.

GLUTATHION-S-TRANSFERASE T1 (GSTT1)
GSTT1 is found in lymphocytes and the liver, and is involved in the detoxification process of a variety of environmental chemicals, such as the ones used in polymer productions. Like all GST Enzymes, GSTT1 detoxifies cancer-causing chemicals as found in cigarette smoke. Approximately 38% of Kaukasions show a complete lack of GTT1 activity. This group with the GSTT1 */0 Genotyp shows a high risk for carcinoma of the lung, breast and larynx.

GLUTATHION S-TRANSFERASE P1 (GSTP1)

GSTP1 is build in blood lymphozytes and tissues such as prostate, lung, breast and brain. It plays an important role in detoxification by catalyzing the conjugation of many drophobic and electrophilic compounds with reduced glutathion.

About 50% of the caucasion population shows complete loss of function, which aids the accumulation of reactive products and thus increases the risk of cancer and neurological diseases.

N-ACETYLTRANSFERASE 2 (NAT2)

The NAT2 functions to both activate and deactivate arylamine and hydrazine drugs and carcinogens. Polymorphisms in this gene are responsible for the N-acetylation process in which humans are segregated into rapid, intermederate or slow acetylator phenotypes.


APOLIPOPOTEIN E (APO E)

APOLIPOPROTEIN E is a apolipoprotein essential for the metabolism of triglyceride-rich lipoprotein consituents. it has been recognized for its importance in lipoprotein metabolism and cardiovascular disease.

ApoE genotyping may help guide lipid treatment when cardiovascular risks are high. It is used as an adjunct test to aid in the diagnosis of dementia and Alzheimer Disease, but an association has not been confirmed.

Abnormalities in the ApoE gene have been found in neonates with brain injuries and/or defects, and may increase the risk for Cerebral Palsy. (Kuroda MM, Weck ME, Sarwark JF, Hamidulla A, Wainwright MS. Association of apolipoprotein E genotype and cerebral palsy in children. Pediatrics 2007;119(2):306-313)
The persistent CNS (central nervous system) effect of lead may be more toxic in individuals who have at least one ApoE-Epsilon4 allele. This study suggests that individuals with ApoE polymorphisms may vary in susceptibility to the long-term effects of lead on the central nervous system.


SUMMARY: WHY GENETIC TESTING

- When we know genetic ‘disabilities’, we are in a better position to protect and support our child’s system
- We can use care in preventing toxic overexposure
- Since we have numerous enzyme systems involved in the detoxification process, we can strengthen our detoxification ability by supporting and strengthening other enzyme systems.
- SOD testing may not be needed, since SOD enzymes are present in everybody’s system. We can test if these enzyme systems function properly, but it would make more sense to first pay attention to potential deficiencies, particularly copper, zinc and manganese before SODs are tested.
- Zinc and manganese deficiencies are not uncommon. Boys are susceptible to zinc deficiency. Blood or hair analyses are an option. While blood tests reveal acute and immediate deficiencies, hair detects if the body has been chronically undersupplied. I would hesitate to have blood drawn from an infant or young child. I consider hair analysis a suitable alternative in locating the problem. If hair analysis results indicate a need for supplementation, it would be logical to supplement for a few weeks before SOD testing is attempted.
- Even in the presence of metabolic or digestive disorders, sufficient nutrients would be absorbed to restore SOD enzyme function to near normal.
- Phase 2 Enzymes are either present or not. When we know that Phase 2 Enzyme systems are missing or non-functional, we also know that we need to support the body’s detoxification potential- because the system cannot detoxify properly on its own. Nearly 50% of the world population misses the GSTM1 enzyme. These people accumulate toxins readily, simply because their body cannot properly detoxify.
- When we know specifics about our individual detoxification potential, we will know how much outside support in the form of natural or synthetic chelation treatment is needed to prevent our body from continuously accumulating the toxins we are exposed to on a daily basis. The sooner we act, the better.
HOW TO TEST?

Genetic testing for detoxification enzymes is simple and inexpensive. The material needed for testing is 1ml of whole blood drawn into an EDTA tube, or 10 drops of whole blood on filter paper, or a gum swap. Test kits are available on request.

For more information: www.microtraceminerals.com

MORE LITERATURE


Probst-Hensch NM, Bell DA, Watson MA, Skipper PL, Tannenbaum SR, Chan KK, Ross RK, Yu MC: N-acetyltransferase 2 phenotype but not NAT1*10 genotype affects aminobiphenyl-hemoglobin adduct levels.


Bell DA, Badawi AF, Lang NP, Illet KF, Kadlubar FF, Hirvonen A: Polymorphism in the N-acetyltransferase 1 (NAT1) polyadenylation signal: association of NAT1*10 allele with higher N-acetylation activity in bladder and colon tissue.

Marcus PM, Vineis P, Rothman N: NAT2 slow acetylation and bladder cancer risk: a meta-analysis of 22 case-control studies conducted in the general population.

Hughes NC, Janezic SA, McQueen KL, Jewett MA, Castranio T, Bell DA, Grant DM: Identification and characterization of variant alleles of human acetyltransferase NAT1 with defective function using p-aminosalicylate as an in-vivo and in-vitro probe.

Katoh T, Inatomi H, Yang M, Kawamoto T, Matsumoto T, Bell DA: Arylamine N-acetyltransferase 1 (NAT1) and 2 (NAT2) genes and risk of urothelial transitional cell carcinoma among Japanese.

Okkels H, Sigsgaard T, Wolf H, Autrup H: Arylamine N-acetyltransferase 1 (NAT1) and 2 (NAT2) polymorphisms in susceptibility to bladder cancer: the influence of smoking.


ABOUT THE AUTHOR:

E. Blaurock-Busch PhD founded the specialty laboratories Micro Trace Minerals of Germany in 1975 and Trace Minerals International, Inc. of Boulder, Colorado in 1984, and continues to be research director of both. She is Scientific Advisor to the International Board of Clinical Metal Toxicologists (IBCMT) and the German Medical Association of Clinical Metal Toxicologists (KMT). She has lectured in universities of countries and to medical groups around the world and received the IBCMT Award for Outstanding Services in 2005. She has written numerous articles and several books in English and German, and is a member of the British Society of Ecological Medicine and the European Academy for Environmental Medicine.