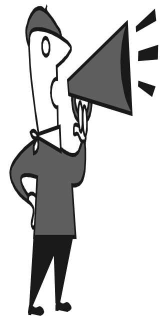
THE NOONAN CONNECTION

The Noonan Syndrome Support Group, Inc. May 2003

HELLO FROM THE PRESIDENT

Hello, so many things have happened in the last several months that I want to share with you all.

The Towson University Department of Electronic Media and Film, Corporate Video class selected TNSSG's video proposal for production during the Spring semester. Thank you to the people and families in MD and the family from NJ who volunteered to be part of this venture.



Even though the students worked in a different time zone (Collage Time) they recorded hours of video, in our homes that resulted in a great 10 minute video about the group.

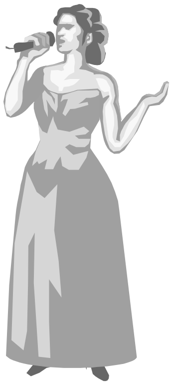
Darcie and I flew to Minnesota on February 13th, to attend a fund raiser hosted by the Minnesota chapter (see page 10 for details). Darcie was asked to share her story after dinner. Imagine my fear, excitement, pride and tears as my 16 year old daughter walked up to the microphone. With candor and grace she spoke with such conviction, telling the audience about her life. Darcie talked about school, church, singing, acting, driving and friends. She then went on to list all of her 'issues' including her learning ability. Here is an excerpt from her talk.

" I have learned that I am Darcie! Period! I also happen to have Noonan syndrome, I am not the syndrome.

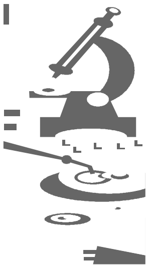
Because of the support from friends and family and people I haven't known for long, and people I don't even know I am able to live my life knowing I have people behind me 100% of the time. I have support, as you can tell I use that word a lot. That's because of what the basis of the group is to me.

Thank you to everyone involved in this fund raiser tonight, because that shows how much you support what we do.

Peggy and her family have meant a great deal to me and I know that with out them and the rest of the team we would not be where we are today. Again Thank you."

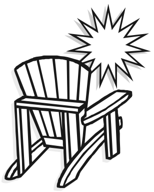


Having been raised in Bermuda, I was some what reluctant to travel to MN in February. Imagine my surprise when blizzard conditions in Maryland caused Darcie and I to stay in MN an additional 3 days! We are very grateful to the Edwards family for there outstanding hospitality, thank you.



An invitation to tour a research facility and hospitals resulted in a trip to Boston, MA in May. The proposed research looks very promising, and as more information is made available I will pass it along.

Wanda H. Robinson



THE NOONAN SYNDROME SUPPORT GROUP, Inc.
A Non-Profit Organization
We reach around the world!
***Founder and President**:* Wanda Robinson
Vice President: Dave Robinson
Secretary: Pamela Hauck
Medical Advisor: Dr. J. Noonan

The Noonan Syndrome Support Group , Inc. was founded in June 1996, by Wanda Robinson.

We offer information and support to those whose lives are touched by Noonan syndrome. The group is self funded. Contributions are gratefully accepted and will help the next family to receive information about Noonan syndrome.

You can reach us at:
E-mail: **wandar@bellatlantic.net**
Or:
info@noonansyndrome.org
Web page:
http://www.noonansyndrome.org
Mail:
TNSSG, Inc. PO Box 145
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Telecommunication service is Funded by the Minnesota Chapter of TNSSG, Inc.
1-888-686-2224 within the United States or
1-410-374-5245 outside of the United States

This newsletter is intended to provide basic information about Noonan syndrome. It is not intended to, nor does it, constitute medical or other advice. Readers are warned not to take any action with regard to medical treatment or otherwise based on the information in this brochure without first consulting a physician. The Noonan Syndrome Support Group, Inc. does not promote or recommend any treatment, therapy, institution or health care plan. The information contained in this brochure is intended to be for your general education and information only and not for use in pursuing treatment or course of action. Ultimately, the course of action in treating a given patient must be individualized after a thorough discussion with the patient's physician's.

You are more than welcome to plagiarize, as you see fit!

Did you know these characteristics are common in Noonan syndrome?

Clinical Characteristics

Short stature (2 SD below mean)
Triangular shape of face
Prominent brow
Hypertelorism
Epicanthus
Antimongoloid palpebral slant
Ptosis
Depressed nasal bridge
Broad apex nasi
Low-set and/or malformed ears
High-arched palate
Normal karyotype

Neck

Short
Webbing
Low hairline

Chest

Shield-like
Wide-spaced nipples
Pectus exacavatum

Cardiac Abnormalities

Pulmonic stenosis (PS)
PS and ventricular septal defect
Atrial septal defect (ASD)
ASD with anomalous pulmonary venous return
Endocardial cushion defect (ECD)
ECD + patent ductus arteriosus and mitral insufficiency
Both PS and ASD
Patent ductus arteriosus (PDA)
Undiagnosed heart disease

Extremities

Cubitus valgus
Gracile fingers
Short stubby fingers
Lymphedema
Dystrophic nails
Shortened fourth metacarpal(s)
Clinodactyly of fifth finger(s)
Palmar simian crease
Undescended testes
Delayed puberty
Skeletal retardation

Cognitive Development

Learning disability
Cognitive disability

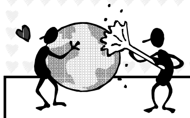
WE NEED YOUR HELP. IF YOU LIVE OUTSIDE THE USA?

Would you be willing to photo copy the Noonan Connection and mail it to other people in your country? It has become to expensive to mail each edition of The Noonan Connection out of country.

An edited edition of The Noonan Connection is available online. 28 people in the UK will receive this edition thanks to the efforts of **Michelle Ellis**, we would also like to thank (Simon Bland of) East Surrey College, Redhill, Surrey, U.K for kindly photocopying this newsletter.

Contact: wandar@bellatlantic.net if you can help

FROM THE HEART



One Mans Story…

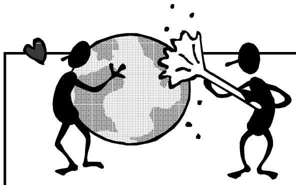
I first "found out" that I had Noonan's Syndrome in 1984 during a counseling session my mom wanted me to go to because I was becoming highly anxious at the time. Being anxious and prone to worry had been a part of my life but it was getting worse at the time. Anyway, since it was her idea I wanted her to see the counselor with me, and it was then that she said I had this thing.

That is not to say I hadn't known that something wasn't "quite right" but I had no idea of any specific birth defect. I knew I was short and looked a little physically different, but not as different as I actually am. Since I realized I have this and the more I find out about it the more questions that get answered. I was probably told that I had NS and there is some history regarding it in my past but I guess I either didn't remember being told or I put it out of my mind.

I cannot honestly say that I thought people looked at me differently, but as I look back I do now see that people did treat me differently because it was clear that I did

(Continued on page 3)

FROM THE HEART



(Continued from page 2)

have some physical disabilities and limitations. I clearly was never chosen first for the mandatory sports teams that physical education teachers imposed on us. In fact, I was chosen last. I was sometimes given somewhat special treatment and seen as needing some type of "special education." These were things that I resented at the time but were probably extremely necessary. It was just that I did not specifically know that I had such a thing as NS and that having this did make me different and that being treated differently was in order.

I lived under the facade that I was like everyone else and that I'd be taller some day and I would live the good old American domestic life as everyone else if not more so. In fact, I wanted to believe that I was somehow destined to be a big shot in the business world and make a lot of money. They were dreams based completely on unrealistic thoughts. The tea leaves were in place to indicate that absolutely nothing of the sort was going to happen. The height of this silliness came in wanting to be a dentist since they make a lot of money, and yet I resented being given special education to deal with my poor muscular coordination. Imagine how silly it is that someone who can't even do fifth grade art projects somehow expects to put mercury in people's mouths because it commands a high salary!

Then again I had no idea about supply and demand. When my father tried to teach me in his normal impatient way about economics he asked me how a grocery store was put where it was to which I responded that "they" put it there. That would have made perfect sense if I was in the USSR but not in the USA. Oh well, I guess I might have been ideal for Mao's Great Leap Forward into disaster.

The most important development that has resulted from having NS is the continuous problem of swelling in my legs. It effects how well and how fast I can walk and requires constant care and vigilance. It started in the fall of 1983 and precipitated numerous visits to different doctors to find out the cause and, hopefully, a solution. What was eventually found out was by an article our family physician read in the New England Journal of Medicine was that some people with NS get edema in the legs with the advent of puberty. In other words, it was a genetic time bomb waiting to go off and so it did. Of all the things that go along with NS, this is the most troubling and requires the most attention. It is also the most debilitating. I can live with looking differently and being a joke in the dating world. I've long since stopped caring about that, but a threat to my mobility is like a threat to national security.

In closing, I have a few thoughts on what would have been beneficial to me in growing up and might be helpful to parents with NS children. First of all, I would have preferred to have been told that I had NS and that certain expectations were out of the question.

The physical therapy and special education things were for my own good, but I didn't want to believe I had a problem since I didn't know that there really was something genetically wrong.

A good stern "look son" lecture might have helped. I might have approached the special education to work on my coordination differently.

It does need to work both ways, however. Special education teachers, I think, need to stop the humiliating "happy smiley" pandering that sometimes come with it. People resent being treated differently like that because it makes them feel subhuman. I can imagine the humiliation that the elderly in nursing homes feel when they are treated like children because they can no longer do certain functions. These are people who built careers, fought the most catastrophic war in history and created the nation's true golden age. They deserve respect not humiliation.

Too often, we treat the disabled in a pandering manner and almost as if they are problems that "get in the way." Special education should not be warehousing children with physical and mental limitations but hard core remedial education that challenges them. If the "cut and paste" special education drills I was expected to do was given with a stern realization that told me, "You have physical limitations that other people don't have. You have to work harder than they do to achieve the same results. This is not to put you down, this is to raise you up. This isn't to humiliate you but to discipline you and with the right amount of discipline you will be able to achieve many things. Some things you will not be able to achieve. I can help put you at the average level of coordination but there is no way it will be above average. Don't concentrate and what you will never be able to do. Focus on what you can do."

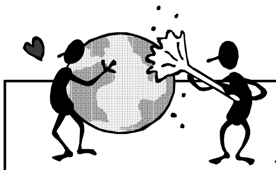
Physical education teachers too often emphasize sports and this has a very detrimental effect. The ones who need physical education the most are the ones who do the most poorly in athletics. People with NS or any other physical problem need good physical therapy and exercise. It is something that I have come to realize, thanks to my fathers continuous lecturing. It is small wonder that when in my high school sophomore year when we were given a choice in what type of PE class we were to take that I chose weight conditioning because you weren't in competition nor were you expected to have certain physical abilities. Anyone with limbs can make use of weight training and any physical educational program should be made so that it benefits everyone, not just those whom are already blessed with good or normal physical abilities.

Sincerely,

Frank Jacobson

MORE THE HEART

DISABILITIES AND DISCRIMINATION



To Tell, or not to Tell?

When I finished University in 2001, I embarked on finding a job. Within weeks I had an interview with Company X. I had talked to my job agency about whether I should tell Company X that I have NS. I was advised that it would be in my interests to tell them as I could be in trouble in the future if I didn't tell them.

On both the application form, medical questionnaire and the interview, I was open when they asked me about NS. On my first day, I even gave my manager a large file of information on NS as she had asked for more information. A couple of days later my manager asked me whether I was 'mentally retarded'. I was shocked by this, as this was only one paper that had suggested that there may be learning difficulties with NS - I hadn't had any special needs in respect of education at all.

Many months later it was mentioned that I may not be doing my job as effectively as I could be. After a year of assessments, meetings etc, I was asked to leave, in effect due to my disability. Of course there is much more to say with what happened in that year, but for sake of this article, all that is relevant is that they asked me to leave in effect due to my disability.

I had worked many temp jobs in the past (I never told them I had any disability) and no-one complained about my work – in fact many companies wanted me to stay on as a permanent member of staff, but due to study commitments, I could not stay. I had never been discriminated in the workplace before, and never thought I would be, as although I have a disability, I have never classified myself as disabled, and in any case, feel that I would only go into a job that I could 'do'. Although I am unable to do some things, which in any case are the more physical activities, I do not see myself as disadvantaged at all, my philosophy being 'everyone has problems, and many are worse off than myself'. I never, not even for one-second think that I would be treated by any employer differently (less favorably) due to my disability.

Looking back, the obvious mistake I made when joining Company X was to tell them that I had NS. They then had this 'unknown' to deal with, and I believe that they were worried, not even the company doctor had heard of NS, and although I gave them the information they requested, this could have impacted the situation; a little knowledge is a bad thing. I believe that throughout that year did everything they could (including 2 neuropsychological assessments) so that they could dismiss me, maybe they were panicking, maybe I was too 'high maintenance' (though I do not see how I would be).

The question is 'Should we tell?' I for one, will not ever tell any future employer I have NS, but I will tell them that amongst other things, I have multiple joint pains and need reasonable adjustments in the form of special chair, footrest etc.

However, this question does not just apply to employment, but everything – should we be obliged to refrain from telling people that we have a disability, because we are scared that we would be discriminated against? I am not suggesting that we want to tell everyone we meet, in fact far from it, personally, I will tell anyone that asks me (mainly doctors) and that's all, but definitely err on the side of preferring not to tell people. We are no less capable than anyone else, and I am sure that my experiences when I worked on temp jobs were not a co-incidence, and that comforts me to know that I am in fact capable of working, capable of doing a good job. I am confident that because I told Company X that I had NS, this although not immediately disadvantaged me, did so eventually.

? There is of course no easy answer to this question, maybe through time people's perception may change, and I am confident that as we continue to grow, we can help make this happen through what we do as a group. ?

Michelle Ellis
London, UK
TNSSG U.K Rep.

Fear causes Discrimination in Church?

After our latest trip to the cardiologist Jacob was put on Beta blockers and we were told by the Cardiologist that he is now to be limited in sports and strenuous activity as it will put a strain on his heart that he can not handle. Ok we will deal with this.

So , Jacob who is home schooled with his other siblings...enjoys playing with his friends at Church....well we were recently told that unless I could stay with him that Jacob could not be part of the Sunday school class...they felt that the teachers (3 in the room) could not, and should not have to handle Jacob because of his recent diagnosis. Imagine our hurt and concern from this, our own church saying this to us ..we were so upset...and tried to explain that the only difference with Jacob today and the day before is that we know have a different diagnosis.

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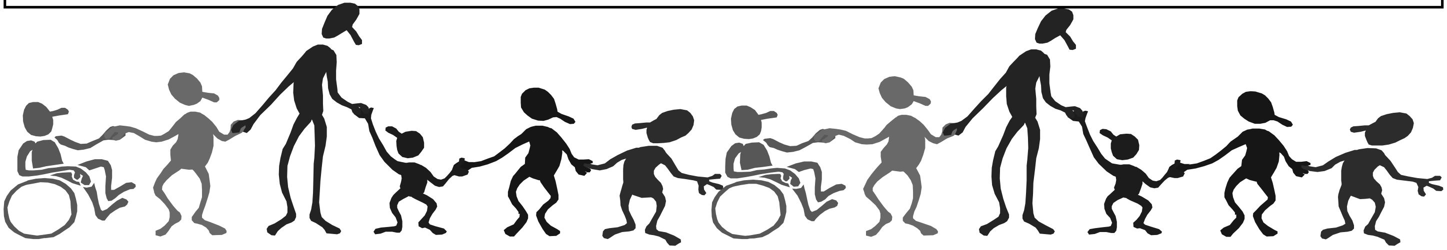
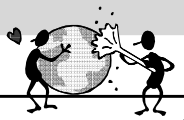




MORE FROM THE HEART

(Continued from page 4)

Needless to say I stay with him in order for him to play with his friends but I was so disappointed...My husband and I have been in Ministry for over 20 and have to say that in all honesty have never come across this attitude (with in the church) before.

I understand what it is like to be afraid of something you do not know about . and we have had the stares and people make comment about Jacob (we even went through that with out other son when he had Cancer and lost his hair and could not walk etc., you learn to live with it) .but I believe that we have been really good at explaining and educating.

Thank God Jacob does not understand what had happened ...just does not like the fact Mom is now in the room with all his friends (seen but not heard) .

Blessings,
Sandra Gardner
Mom to Jacob 5 with NS and Hannah 12 with no NS and Mckenzie 11 no NS but a Cancer survivor.
British Columbia, Canada

Fear Causes discrimination in school?

As a parent with a child that has Noonan Syndrome I do feel that he is discriminated against and not totally accepted. At school he is always teased by his peers because of his size, facial features and learning disabilities. He is also discriminated against by the teachers when it comes to his health issues and learning disabilities.

Time after time, four years to be exact I have brought written materials from Doctors and about Noonan Syndrome to aid them in helping Dezmond. Yet I have to constantly remind them that he is not supposed to be playing sports (due to his heart condition (HCM) and remind them of his IEP. I remind them that he is supposed to be getting more help during class time.

I'm always told "He looks so normal, we just forget sometimes". To me that is discrimination! What is looking normal?

Dana Timmons
Mother to Dezmond Wingate (NS) age 9
Pineville, North Carolina

Discrimination is unfair...

Justin is five years old and was diagnosed with Noonan Syndrome when he was two. I was told the day he was born about numerous anomalies that he had, but I was never told that these anomalies were all related, and might mean that he had any congenital problem. From the beginning, Justin has been discriminated against. Not on purpose, but due to a lack of knowledge about Noonan Syndrome and the law. To end discrimination, we must communicate, and educate, and advocate for our children.

Not until Justin was diagnosed with Failure to Thrive, did any doctor look at Justin as a whole person. Doctors did not listen to me when I would explain Justin's symptoms. When I tried to get Justin into physical therapy, I was asked by a doctor if I might be taking on too much responsibility.

My baby was nine months old, had not gained 7 pounds since birth, and was not yet sitting with assistance. He was looking at numerous surgeries in his future, to correct what anomalies could be corrected. He was a person, a human being, and I was going to make sure that he got all the help that he deserved. I don't think asking for a referral to P/T was unreasonable.

Failure to communicate, and be heard, causes discrimination too. Justin had abdominal surgery to bring down an undescended testicle. After the surgery proved more difficult than expected, Justin stayed overnight in the hospital. Since his surgery was usually performed on an outpatient basis, we were put in a room farthest away from the nurse's station and were not checked on regularly. We were even questioned by a nurse as to why we were staying overnight for such a simple procedure. After returning home, Justin became inconsolable.

Two days later I called the specialist, who said I was over reacting, that nothing could possibly be wrong with the surgery, and that I was simply imagining things. Two days later, Justin was back in the hospital for ten days, nearly dieing of scarlitina. I could go on and on, about not communicating and not being heard. Remember to listen, and remember to find someone to listen to you.

Sheer ignorance on the public's part also causes discrimination. Many believe that if they can't see a disability, that there isn't one. Due to Justin's low muscle tone, by age four he was still not potty trained. I tried to put Justin into a summer day camp with his older brother, but they refused him on the grounds of his potty training. I had to go to the director of the camp and explain Noonan Syndrome and low muscle tone and how this affected Justin's potty training. Eventually, Justin was accepted and thrived in the program.

A lack of knowledge about Noonan Syndrome causes discrimination, and so does a lack of knowledge about the law where you live. I am currently fighting with the school system to keep Justin coded with a disability, to ensure him an education that will help him to reach his fullest potential. You see, because he is affected minimally, and has come so far with the services he has already received, the school seems to feel he is not longer affected, and therefore, no longer in need of services. Here in Maryland, that is legally not the case. Why can't they just give him what every child deserves; a chance.

Discrimination is unfair, wrong, and illegal. We as parents need to advocate for our children; even our unaffected children. No child should "fall through the cracks". We must educate ourselves, about Noonan Syndrome and about the laws where we live. Then we must communicate with and educate others. With this, we can

NEWS FROM SOUTH AFRICA

Mrs L. Cinnamond
P .0. Box 64216
Highlands North 2037

14th March 2003

Dearest Wanda

Hi -I printed the renewal form out and work and I am enclosing same for record purposes. I will always be a life long member and support the organisation in any way I can. I have worked on so many things and I am enclosing a few bits and pieces what I can get my hands on which is lying around my office.

As you know I sit on the committee of SAIDA (South African Inherited Disorders Association) and from there I am able to promote Noonan Syndrome. The SAIDA news goes out to over 800 members mainly in the medical fraternity.

I attended the Birth Defects Conference and the policy guidelines for Genetic Disorders and Birth Defects was published. So any reference to SAIDA jn the booklet wanting info on Noonan Syndrome will then get forwarded to me. I am doing my little bit from South Africa and hope to contribute and make a difference.

With some money I raised I had 3000 pamphlets printed and have distributed some at the birth defects conference, and various other conferences held in SA where the Profs from SAIDA attended and took pamphlets with. Taryn is in the front of the pamphlet, Shauna Palmer on the inside and Leanne Kirkman's little boy Michael-Jon.

Just a brief update of what I have been up to -I will copy the videos and send them with an item to raffle.

Best wishes to you and the family. Most importantly thank you for what you do for so many families around the world. Your efforts are outstanding and without you we would all be so lost.

With much love,

Lucy Cinnamond

MEDICAL NEWS
21 NEW ARTICLES SO FAR IN 2003

Information found on the world wide web at Medscape.com.

1. Horiguchi T, Takeshita K
Neuropsychological developmental change in a case with Noonan syndrome: longitudinal assessment [In Process Citation] Brain Dev (Netherlands), Jun 2003, 25(4) p291-3

2. Deleon SY, Dorotan J, Abdallah H, et al.
Annular and Leaflet Augmentation in Noonan's Syndrome with Dysplastic Pulmonary Valve [epub ahead of print] [Record Supplied By Publisher] Pediatr Cardiol (United States), Apr 30 2003, p

3. Mazzanti L, Cacciari E, Cicognani A, et al.
Noonan-like syndrome with loose anagen hair: A new syndrome? [In Process Citation] Am J Med Genet (United States), Apr 30 2003, 118A(3) p279-86

4. Jung A, Bechthold S, Pfluger T, et al.
Orbital rhabdomyosarcoma in Noonan syndrome. J Pediatr Hematol Oncol (United States), Apr 2003, 25(4) p330-2

5. Ho WL, Wang JK, Li YW
Radiological features of late-onset lymphoedema in Noonan's syndrome [In Process Citation] Pediatr Radiol (Germany), Mar 2003, 33(3) p200-2

6. Bertola DR, Carneiro JD, D'Amico A LA, et al.
Hematological findings in Noonan syndrome [In Process Citation] Rev Hosp Clin Fac Med Sao Paulo (Brazil), Jan-Feb 2003, 58(1) p5-8

7. Delhaas T, Muhler EG
Proximal left coronary artery occlusion in a 15-year-old boy with noonan syndrome and hypertrophic cardiomyopathy.
Pediatr Cardiol (United States), Jan-Feb 2003, 24(1) p67-9

8. Tartaglia M, Cotter PD, Zampino G, et al.
Exclusion of PTPN11 mutations in Costello syndrome: further evidence for distinct genetic etiologies for Noonan, cardio-facio-cutaneous and Costello syndromes [In Process Citation] Clin Genet (Denmark), May 2003, 63(5) p423-6

9. Noordam C, Thoonen G, van der Burgt CJ
[The Noonan syndrome from a pediatric perspective] Ned Tijdschr Geneeskd (Netherlands), Apr 5 2003, 147(14) p644-8

10. Ishikawa Y, Sekiguchi K, Akasaka Y, et al.
Fibromuscular dysplasia of coronary arteries resulting in myocardial infarction associated with hypertrophic

(Continued on page 14)Articles

Thank you to the following people, companies and foundations for their continued support.
They are making a difference.
Because of them we will continue to tell others about Noonan syndrome! January 2003 –May 2003

- Eugene Snell
- John MaCarthur and Renee Khatami
- Saint Matthias' Episcopal Church, East Aurora, NY
- David Maag
- Michelle Ellis
- United Way of Merrimack Valley, Keith Sandler
- United Way of Northeastern NY,
- CSX PacMatch, Jim Ebanks
- United Way of Kitsap County
- Drs. Kurt and Stephanie Bowers
- Martin and Cynthia Chappell
- Inna Snitokovskaya
- Mrs. Philomena McCaughey,
  In Honor of Ryan James Shaw
- Sherrill Smith
- Carlos Collado
- Tara Lillis,
  In Honor of Patrick Rosen
- Carolyn Hudlow,
  In Honor of Matthew Chapman
- Sharon Siegel
- William Hudlow
- Denise Michaelis
- United Ways of New England
- Karen Durbin
- James and Carolyn Farley
- Regina Soos
- Brenda and Melvin Schrager
- Traci and Scott Schutt
- Eleanor Bondurant,
  In Memory of Toms' Birthday
- SBC Employee Giving United Way Campaign
- Grace Johnson,
  In Honor of Heather Johnson
- Laurie Dolly
- Linda Manglass,
  In Honor of Joe Shapiro's 14th Birthday
- Kathy Max
- Elizabeth F. Swanson-Hyland
- IGive
- Lenore Boak
- Rebecca Bockrath
- Steve and Lorie Harding
- Thomas and Cheri Chapman,
  In Honor of Braeden Chapman
- Robert and Sistine Emons
- Carol Roque
- Gary and Joan Thompson,
  In Memory of Danny Thompson
- Mark and Terri Gilallon
- Donna Ryczek O'Toole
- Scott and Leanne Davis,
  In Honor of Brayden Davis
- Robert and Mary Patz,
  In Honor of Juliette Rose Walker
- Lawrence and Charlotte Franke Sandra Nolen
- Shirley and George Trevains
- Pat and Stanley Gralski
- Cynthia Weaver
- Doris Reba,
  In Honor of Kandece Carmen
- Roland and Lavina Jackson
- Peter and Danielle Backes,
  In Honor of Christopher
- Anne Sluys
- Anne King
- Lorraine Long,
  In Honor of Lauren Long
- Pascuala Espinoza
- Samantha Pollard,
  In Honor of Autumn Pollard
- Josephine and Louis Marrelli SR.
- Jan Irvine
- Daile Hawley
- Louis and Lillian Levy
- Kristi Nance-Hmurovich
- Kathryn Anderberg
- Brenda Dickinson
- Louis and Josephine Marrelli
- Gloria Isaacson
- Scott and Lisa Carman
- Nancy Tafrow,
  In Honor of Kandece Carmen
- Joseph and Rose Csorgo,
  In Honor of Kandece Carmen
- Paula Saylor
- Joy Minns
- Clifford and Cindee Hersey
- Loretta and Thomas Gliniany,
  In Memory of Lyle Larkin
- Laurie and Mark Pappenfus,
  In Memory of Lyle Larkin
- William and Elizabeth Engelking,
  In Memory of Carol and Lyle Larkin
- Rick Moore,
  In Memory of Corey Ellis
- Arthur and Claire Powers,
  In Memory of Marguerite Koken

CONFERENCE QUESTIONNAIRE

We are starting to plan our next conference, which will be held in Maryland in the summer of 2004. In order fulfill everyone's needs and encourage as many people as possible attend we are requesting your input. **Thank you for filling out this form-please return it to: TNSSG, P O Box 145, Upperco, MD 21155**

The meeting starts in the evening with registration and a social, allowing people to mingle, kids to play and for some people an opportunity to talk to our speakers. We also encouraged people to make story boards that introduce their family. Snacks are provided along with a cash bar.

1. How important is the social?
   Important ____ Not Important _____
2. Is it important to make story boards (posters)?
   Important _____ Not Important _____
3. How important is it have snacks?
   Important ____ Not Important ____
4. How important is a cash bar?
   Important ____ Not Important ____

The registration fees for the last conference included the social on Thursday evening, continental breakfast and lunch on Friday and Saturday, a buffet breakfast was included on Sunday. Dinner on Saturday Evening was optional and required an additional registration fee.

1. How important is it to provide a continental breakfast ?
   Important ____ Not Important _____
2. How important is it to provide lunch?
   Important ____ Not Important _____
3. How important is the dinner on Saturday Evening?
   Important ____ Not Important _____
4. I would rather be responsible for all my meals?
   Yes_______ No_________
5. I would rather be responsible for my breakfasts?
   Yes_______ No______
6. I would rather be responsible for my lunch?
   Yes ______ No _______

In the past Daycare has been provided at every meeting, for all children, at no charge to the parents. The group has incurred all expenses related to Daycare. In order to continue to provide a safe, fun and secure environment for the children needing Daycare this will have to change.

1. If your child requires Daycare, how much would you be willing to pay per day? $ _______
2. Would you still bring your children to the conference if you had to pay for Daycare?
   Yes ______ No _______
3. Would you attend the conference if Daycare was not provided?
   Yes ______ No ______

Below are some of the topics people have expressed an interest in hearing about. Please check areas of interest and presentation format that you would like, if you were to attend the meeting.

KEY: **SGS**= Speaker/General Sessions, Professionals knowledgeable about Noonan syndrome: **WS**= Work Shop, small groups lead by facilitator. : **R**= Research , small groups providing information to Doctors/researchers about Noonan syndrome : **PE**: Personal Experiences shared by Adults with Noonan Syndrome, panel of adults with Noonan syndrome answering questions.

TOPIC	SGS	WS	R	PE
Noonan syndrome -History				
Associated Disorders				
Genetics				
Prevalence				
Similar Disorders				
Diagnosis - Pregnancy-Early Development				
Early Intervention				
Eating/Feeding issues				
Genetic testing/counseling				
Pregnancy and birth				
Sleep disorders				
Cognitive/Social/Behavioral -Issues				
Behavioral Aspects				
Cognitive aspects				
Psychiatric Aspects				
Social aspects				
Children's Activities				
Adolescences with Noonan Syndrome - (Advocacy-Positive self)				
Sibling Work Shop				

TOPIC	SGS	WS	R	PE
Medical Physical Characteristics- and Intervention				
Anesthesia				
Auto immune issues				
Bleeding disorders				
Cardiovascular Disorders				
Dental Issues				
Eating/Feeding Disorders				
Growth Disorders				
Hearing				
Lymphatic issues				
Sexual Development				
Vision				
Other				
Fund Raising—what you need to do				
Insurance and Wills-Protecting your child's future .				
Parents Corner—parents share				

Dr. Noonan Requests Your Help

Dear Noonan Syndrome Adult,

I am conducting some research on Noonan syndrome, with a particular emphasis on growth. This questionnaire will be helpful and the response kept confidential. Your identity will not be used in any possible publication.
If you are willing to participate, please complete this questionnaire and return it to me.

Thanks,
Dr. J. Noonan
Dept. of Ped Cardiology
Room MN 470
University of Kentucky Medical Center
800 Rose ST
Lexington, KY 40536-0298

Patient Questionnaire for Adult with Noonan syndrome.

Hi, This is a little questionnaire from the department of pediatric Cardiology of University of Kentucky Children's Hospital. It's a questionnaire about Noonan syndrome, it contains some questions about your health and well being. It will only take about 10 minutes. (If you feel uncomfortable with any question or don't know the answer, fell free to leave it open.) We would be very grateful if you would want to fill out this questionnaire on behalf of a research we are doing on Noonan syndrome (You could ask a family member to help you if you want} .
Thank you!

1. What is your sex?

2. What is your birth?

3. What is your height? (Measured without your shoes)

4. What is your weight?

5. What was your birth weight?

6. What was your birth height?

7. What was the duration of your mother's pregnancy (gestation)?

8. How far did you go in school? (what grade, high school, college, university?)

9. Did you ever have learning difficulties or special education?

10. Are you employed? (If not please explain reason)
What kind of job do you have?

11. Have you had any heart problem in the past? If yes did this problem require surgery?

12. How is your health at the present time?

13. When was the last time you saw a physician? (e.g.- primary health doctor, cardiologist, pediatrician)

14. Are you using any medication? (If yes please write down name and dose)

15. Have you ever used growth hormone? (If yes please tell us how long and at what age)

16. Are you married?

17. Do you have children? (If yes please write down sex and age of your children and if they have Noonan syndrome)

18. Does one of you family members have Noonan syndrome? Who?

19. For females: at what age did you start having your periods?
For males: At what age did your voice lower?

At what age did you start shaving?

20. Did you ever have any psychological problems?
(If yes please describe.)

At what age?

FEBRUARY IN MINNESOTA– COLD WEATHER STIMULATES WARMTH!

On February 13, 2003, a dedicated group of individuals in Minnesota raised thousands of dollars for The Noonan Syndrome Support Group, Inc. More than 130 people attended a black tie dinner dance with silent and live auction. The tables were set with white linen table clothes and red napkins, with a chocolate heart on each plate and chocolate kisses for everyone. As in the past, an awesome array of fresh flowers added to the ambiance and helped to set the mood for the evening.

Guests arrived, greeted by the melodic sounds of talented musicians playing the piano and flute. The silent auction gave people an opportunity to bid on an astonishing number of items, shoes, artwork, vacation packages and much more. During a short program after dinner Peggy Edwards read the poem ‘Welcome to Holland’, Dr. Pierpont and Dr. Rebhuhn also spoke, as did Darcie Robinson. The live auction included round trip airline tickets donated by North West Airlines, a weekend at a beautiful lake front ‘cabin’ hosted by gracious and entertaining families, sports memorabilia and vacations at a resort in Mexico. The evening ended as elegantly as it began with a jazz trio providing the entertainment.

The Noonan Syndrome Support Group, Inc. would like to thank all of the families involved in the Minnesota Chapter for making this event such a success. *Many people contributed to the success of this event, especially, Peggy and Paul Edwards, Clair and Peter Peitsch, Patrick and Connie Larkin, and Tracy and Alden Skar. In addition, we would like to recognize Marion and Cameron Meed, Ian and Megan Bissett, Christian Peitsch, Sarah Storvick and Surdyk’s for the 48 bottles of wine donated for the dinner.

TNSSG would also like to acknowledge the following people who contributed to the success of this event:

John Kopas / Abbey Carpet, Larry & Kathy Allen, Jim & Mary Anderson, LeRoy & Carla Anderson, Ray & Carmel Ayotte, Carol Baldwin, Brenda Bartz, Kay Bartz, Jim & Phyllis Bastian, Craig & Ann Baumann, Thomas Baumgardner, Ph.D., Ken & Betty Behnken, Jay & Ann Behnken, Greg & Teri Beinlich, Bob & Carmen Bell, Brooks & Linda Berg, James Betz, Linda Biehn, Ian & Megan Bisset, Steve Boehlke, Jeff & Toni Burns, Craig & Mary Burton, Bonnie Butenhoff, Jim & Doreen Carter, Tom & Nancy Conlin, Jeff & Ruth Ann DeFrang, Bill Blinco & Vicki Dellwo, Jim & Lynne Dornfeld, Deven & Melissa Dubuque, Ronald & Rhoda Ebersole, Jim & Sue Edberg, Jonathan & Renee Edwards, John & Phyllis Edwards, Paul & Peggy Edwards, Mark & Cheryl Elsbernd, Chris & Brenda Erikstrup, Donna Erstad, Owen & Marjorie Erstad, Tom Evenrud, Martin Evenrud, Hamid & Michelle Fallahi, John & ViAnn Fort, Marion & Connie Freed, Marie Garbe, Lee & Kim Glenna, Joseph & Loretta Gliniany, Eric Griffin, Cindy Grochowski, Chad & Ann Grochowski, Ryan & Jill Haddorff, Hilree Hamilton, Dan & Karla Hansen, David Hegle, Lynne Heitner, Marty & Joan Heuer, Paul & Tina Heuer, Mark & Kathy Heuer, Jay & Betsy Hickey, Jensen, Bell, Converse & Erickson, P.A., Dave & JoAnne Johnson, Greg & Debbie Johnson, Brian & Diane Jordahl, Bob & Reno Kapaun, Dave & Karen Karnuth, Paul & Cindy Karos, Jamie & Dawn Keelin, Brian & Barbara Kimm, Scott & Kathy Kinkade, Judith Koch, Brad & Marcia Kolstad, Maggie Kottman, Jim & Mary Kowalski, Kathleen Kulus, Eric & Ann Kunz, Travis & Shannon Kunz, Scott & Lisa Lacek, Lyle & Carol Larkin, Patrick & Connie Larkin, Mark & Julie Lauze, Tom & Carol Lindborg, Scott Machut, Dean & Jacquie Mahlum, Mike & Barb Mahoney, Anne Kelly & Tom McCarthy, Craig & Shirley McCourt, Mike & Becky McGraw, Mike & Sharon McNulty, Paul & Joyce Melchert, Jeff & Brenda Michniewicz, Mark & Carey Moe, Paul & Sean Moriarty, Mary Morley, Dan & Stacy Mott, Stephan & Lori Nalefski, Marty & Jodi Nelson, Richard & Nancy Nelson, Jeff & Ann Nordeen, Mike & Pat Nordeen, Dave & Katie Ossell, Edward & Lois Elaine Peitsch, Peter & Clare Peitsch, Kip & Anne Pendleton, Kenneth & Nancy Petchenik, Brady & Lisa Pierce, Dr. Mary Ella Pierpont, Artie & Jennifer Pingolt, Michael Pintz, Tim & Patti Pitera, Dennis & Nancy Raas, Scott Ree, Frank Rimell, Jim & Mary Beth Roane, Paul & Rhonda Robinson, Dave & Peggy Rollins, Chuck & Wendy Ross, Rev. Kenneth & Lorraine Roufs, Jennifer Rudolph, Jolene Satre, Stephen & Patricia Schmid, Dale & Kim Schreiner, Matt & Jean Schriner, Irene Schultz-Albert, Ken & Rachel Schwalbe, Joseph Seidel, Roger & Emmy Simmons, Maurice & Doris Skar, Kathy Jones & Sherwin Skar, Alden & Tracy Skar, Jeff & Kathleen Smith, David & Marie Sortland, David & Cindy Splettstoeszer, RBC Dain Rauscher Sponsor table, Jenean Starwalt, Rob & Heidi Stevenson, Mark & Gretchen Stevenson, Martha Stoehr, Steve & Penny Storo, Jon & Sarah Storvick, Matt & Kim Stratton, Jim & Lynne Surdyk, Dr. Robert & Nancy Swanson, Chip & Ginger Symington, Steve & Jeanne Szczeck, Roger & Judy Tews, , Pastor Gary & Maxine Van Hunnik, Ted & Sara Vanderpan, Mary Wagoner, Edward & Esther Walenga, David & Kelly Warfield, Gerald & Cheryl Waste, Jennifer Weichert, Rae Lynn Westbury, Karen Wiegert, Sandy Wiemer, Howard & Betsy Wiener, Steve & Gwenna Wilson, Chuck & Beth Wright, George & Susan Yoshida, Russ & Susan Pierre Zilles,

Hundreds of people made this event possible, all of them contributed to making a difference in the lives of people affected by Noonan syndrome.

* We apologize in advance if we inadvertently did not include anyone who contributed to this event.

3,000 EGGS HIDDEN IN MN!

A special word of thanks to Alex and Isaiah Edwards who on April 19, 2003 at Shawnee Park, on Battle Creek Lake in Woodbury, MN hosted the 6th annual Easter egg hunt, in honor of the children and families affected by Noonan Syndrome.

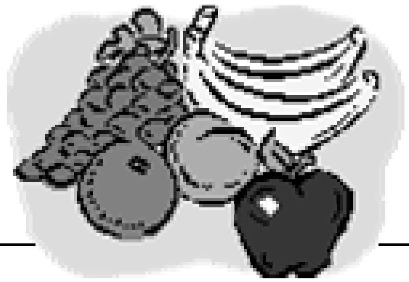
This year's event included more than 3,000 eggs, a special hunt area for the tikes, along with prizes, raffles and tons of fun. Hot dogs, chips and cotton candy were also part of this extravaganza.

Low Fat Diet ...Improves Quality of Life?

Last Oct, my son John,13, NS, came down with chylothorax **(lymph fluid** leaking into the pleural(lung) cavity. Our thinking at the time was that there were no signs. However, after he had a chest tube put in in ped. ICU to drain the fluid, we noticed that a lot of the things we thought were just normal for him, had now disappeared. He lost his night sweats, he lost his reflux, he had more stamina, more muscle tone, more ability to run faster and longer, less irritable, he ate a lot more, and all these things mentioned he had for all his life.

Now, a few weeks ago, I notices that he started having mild night sweats, he started his throwing up again, his muscle tone decreased somewhat, as did his stamina, he started saying he wasn't hungry even though he should have been, and became more irritable. I thought the lymph may be back. Within a few days, I noticed that his scrotum seemed much larger, and his groin seemed more puffy. But, he had no pain. We took him to the pediatrician and were told that yes, in fact, he had a new lymph obstruction in the area, and now has lymph edema in the area. We continue to keep him on a very low fat diet (20 grams a day). He has been on this diet since October.

It's possible, as I look back, that in the last month, at times he may have had an extra 5 - 10 grams of fat a day at times. Maybe this kicked it in, maybe it had nothing to do with it. In the meantime, the doctor said that he expects that the fluid will be reabsorbed into his system, and to keep him to his 20 gram a day diet. This is really tough to do and has significantly changed our family's lifestyle and more so, John's. But, as with all our little Noonan Syndrome kids, we do all we can to keep them healthy and alive.



It's challenging, and upsetting at times, but if we succeed, it's so rewarding. If anyone else has had experience with this, I'd love to hear about it and how you handled it and if it ever resolved totally. What about the adults? Can you add to this. Does it ever go away after puberty, or with Growth Hormone, or some other trick?

Mary Timmins
from Long Island.

PS I received a phone call from a young woman about a month ago. Her 3 1/2 year old NS daughter was in the hospital with lymph leaking into the pleural cavity. She was so scared I felt so bad for her. The hospital was only about 15 minutes from my home. It's so great that we have this group so we can all find each other for support, esp. at the times that we are feeling so scared and alone. Thank you Wanda and everyone in the group who works so hard to keep us connected.

A mother shares her journey as she learns about a Fatty Acid Oxidation Disorder

Kris asks 'is there a connection between NS and the FAODs.'

Hello Wanda Robinson. I haven't been in contact with the group since we moved almost two years ago, but have come upon some information that I would love to share with other parents of children with Noonan Syndrome.

My daughter, 6, always had feeding problems and failure to thrive. For most of her life, she became extremely sick to her stomach every six weeks -- every six weeks like clockwork -- with a fever and severe vomiting. Over the course of several days she would become completely listless, unable to keep down even water down. She threw up a lot of bile. The episodes became worse as she got older.

Finally, about a year ago, she was diagnosed with a fatty acid oxidation disorder. It's a horribly complex metabolic phenomenon that basically means she does not break down fat normally. The fatty acid oxidation disorders are thought to be the cause of somewhere between 5 and 15% of sudden infant death syndrome, according to very recent medical research (it's only been in the last several years that it was possible to diagnose fatty acid oxidation disorders.)

As I've learned more, I've become fascinated by what appear to be some similarities between Noonan Syndrome and the fatty acid oxidation disorders, like short stature, ptosis, low muscle tone, failure to thrive, enamel defects. And I've been reminded of how many list-serv comments there used to be about eating problems and failure to thrive.

I certainly have no scientific basis for suggesting that there's a connection between NS and the FAODs. (There's no medical literature.) But if there's anybody out there dealing with severe episodic gastrointestinal crises, I'd love for them to be able to bring up the topic of fatty acid oxidation disorders with an endocrinologist.

We never would have been diagnosed at all had we not coincidentally picked an endocrinologist for growth hormone therapy who just happens to have an interest in fatty acid oxidation disorders. There is a medication, called Carnitore, that drastically improves the quality of life for kids who don't metabolize fats. Without it, sudden death is a distinct possibility. My daughter's "episodes" were becoming so severe, I know she would have died during one of them eventually. Aside from that, she felt rotten so much of the time, I so wish I could have had a diagnosis sooner.

Kris Newcomer
Fort Worth, TX.
knewco@aol.com

KNOW YOUR (FERPA)!

The General Education Provisions Act at 20 U.S.C. 1232g, also known as The Family Educational Rights and Privacy Act of 1974 (FERPA)

If you live the United States and you have children who impacted by IDEA it is important to understand the Family Educational Rights and Privacy Act of 1974 (FERPA).
"(FERPA applies to all educational agencies and institutions (schools) to which funds have been made available under any program administered by the Secretary of Education.)" in other words the federal government.

- Did you know that you have a right to inspect and review any education records relating to your children that are collected, maintained, or used by the school? Ask to review those records.
- Did you know that the school will comply with a request without unnecessary delay and before any meeting regarding an IEP, and in no case more than 45 days after the request has been made.
- Did you know you have the right to a response from the participating school to reasonable requests for explanations and interpretations of the records
- Did you know you have the right to request that the school provide copies of the records that are "directly related to a student and are maintained by an educational agency or institution or by a party acting for the agency or institution? Given the definition, it follows that tests taken by a child are included in the education records." You may be charged a reasonable fee for copies. Ask for copies.
- Did you know you have the right to have your representative inspect and review the records? Take someone with you, spouse, neighbors, friends.

You already know how important it is to collect your children's health records. As the laws continue to change regarding the education of our children it is as important to collect your child's educational records.

Here is what IDEA actually says, this is taken from 'discover IDEA CD 2002'
Subpart E-Procedural Safeguards
Confidentiality of Information
§300.562 Access rights.

(a) Each participating agency shall permit parents to inspect and review any education records relating to their children that are collected, maintained, or used by the agency under this part. The agency shall comply with a request without unnecessary delay and before any meeting regarding an IEP, or any hearing pursuant to §§300.507 and 300.521-300.528, and in no case more than 45 days after the request has been made.

(b) The right to inspect and review education records under this section includes-

(1) The right to a response from the participating agency to reasonable requests for explanations and interpretations of the records;

(2) The right to request that the agency provide copies of the records containing the information if failure to provide those copies would effectively prevent the parent from exercising the right to inspect and review the records; and

(3) The right to have a representative of the parent inspect and review the records.

Discussion: Part B incorporates and cross-references the Family Educational Rights and Privacy Act (FERPA). Under Part B, the term "education records" means the type of records covered by FERPA as implemented by regulations in 34 CFR Part 99. Under §99.3 (of the FERPA regulations), the term "education records" is broadly defined to mean those records that are related to a student and are maintained by an educational agency or institution. (FERPA applies to all educational agencies and institutions to which funds have been made available under any program administered by the Secretary of Education.)
The term "all education records" is from the statutory reference to "all records relating to such child" at section 615(b)(1) of the Act. The Department has always interpreted the term to mean all of the child's education records to be consistent with the purpose of IDEA and the applicable confidentiality provisions of the General Education Provisions Act at 20 U.S.C. 1232g, also known as the Family Educational Rights and Privacy Act of 1974 (FERPA) as directed by section 617 (c) of the Act.
Education records are defined at §300.560 by reference to the definition of education records in 34 CFR part 99 (the regulations implementing FERPA). The term means those records that are directly related to a student and are maintained by an educational agency or institution or by a party acting for the agency or institution. Given the definition, it follows that tests taken by a child are included in the education records available for review by a parent. The discussion following §300.562 in the attachment further discusses what is considered an education record of a child and the timelines for parental inspection and review of education records.

Knowledge and understanding lead to positive results for people of all abilities. Find out how you can help insure positive results. Be a part of the answer. See page 13

Will my child with Noonan syndrome be cognitively impaired (mentally retarded)?

A common question parents have when told that their child has Noonan syndrome

♥ **Will my child with Noonan syndrome be cognitively impaired (mentally retarded)?**

In information published about Noonan syndrome it was typically said that people with Noonan syndrome were mentally retarded.

One of the first documents published by our group in 1996 recognized and stated that our children "most often have normal intelligence, and approximately 1/3 of individuals with Noonan syndrome will have learning disabilities and some will have mental retardation." [1]

In 1999 it was revealed the more severe cardiac defect and more evident facial and skeletal anomalies, was associated with a specific pattern of deficits and capacities in cognitive functioning.[2]

A paper published in 2002 uses these words "varying degrees of mental retardation" as a characterization of people with Noonan syndrome.[3]

As a group, we are in a unique position of having an opportunity to validate and improve the educational outcomes and treatment of people with Noonan syndrome. **<u>However, we need your help.</u>**

Here is what **you can do**, <u>mail TNSSG a copy of all non-medical results from evaluations of affected individuals in your family.</u> These would include but are not limited to tests in these areas:

- Intellectual/Cognitive Functioning
- Academic Performance
- Communication
- Motor Skills
- Sensory Status
- Health/Physical Status
- Functional Skills
- Emotional/Social/Behavior Development

All individual results are confidential. Dr. Thomas Baumgartner* has agreed to review our collected records with the express purpose of presenting the initial results at our 2004 conference. So, act today, collect and copy your reports and put them in the mail! (see address below)

When mailing reports please make sure that your name and contact information is included, in case we have any questions.
Reports should be mailed to
TNSSG, Inc. (Reports)
P O Box 145
Upperco, MD 21155 USA

Knowledge and understanding leads to positive results for people of all abilities. Find out how you can help insure positive results. Be a part of the answer.

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[1] Maura Kenton, MS Eric Wulfsberg, MD, TNSSG, Inc. Understanding Noonan syndrome A Parents Guide 1996

[2] Van der burgt I, Toonen G, Rossenboom N, Assman-Hulsmans C, Gabreels F,Otten B, Brunner HG, Patterns of Cognitive functioning in school-aged children with Noonan syndrome associated with variability in phenotypic expression. J Pediatr 1999; 135: 707-713

[3] M. Menahe, R. Arbel, D. Raveh, R. Achiron and S. Yagel , Poor prenatal detection rate of cardiac anomalies in Noonan syndrome Ultrasound Obstet Gynecol 2002; 19: 51-55

*Dr. Baumgardner is a clinical neuropsychologist in private practice in Lutherville, Maryland where he specializes in the assessment and treatment of children and adolescents with behavioral, learning, emotional, and cognitive disabilities. He received his Doctorate from the California School of Professional Psychology, including Internship and Post-doctoral training at the Kennedy Krieger Institute. From there he joined the research team of the Learning Disabilities Research Center at KKI, where he studied children and families with Turner's Syndrome, Fragile X Syndrome, Tourettes Syndrome, Neurofibromatosis Type-I, and Reading Disability. While remaining involved in clinical research and publication, Dr. Baumgardner's clinical work includes providing neuropsychological evaluations for primarily children but also adults. He is part of a multidisciplinary group of psychologists, psychiatrists, social workers, and behavioral pediatricians located at Johns Hopkins at Greenspring Station.

IN THE NEWS NOONAN SYNDROME AND A RARE FORM OF LEUKEMIA

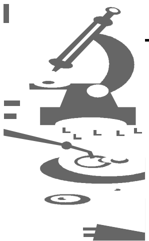
The research program in the Gelb laboratory concerning Noonan syndrome continues to be very active. This program recently was approved for support from the National Institutes of Health for a five-year award as well as a two-year award from the March of Dimes. The most important recent discoveries relate to blood cell production problems that are associated with Noonan syndrome. Study of five children with Noonan syndrome and a rare form of leukemia as well as two other kids with short stature, pulmonary stenosis and that same blood disease revealed that all had inherited abnormalities in the Noonan syndrome gene (called *PTPN11*). It also appeared that there was a particular mutation that predisposed to the leukemia since five of seven had that DNA change while less than 2% of all children with Noonan syndrome had it. This finding has also led to new research concerning leukemia. It now appears that mutations in the *PTPN11* gene acquired in the bone marrow after birth are an important cause of some blood disorders in childhood. This work is described in a forthcoming publication in *Nature Genetics*.

Current efforts in the Gelb lab are directed towards understanding how *PTPN11* mutations result in the specific problems observed in patients with Noonan syndrome, hopefully leading to new and better ways of treating certain aspects of the disorder. Efforts are also in progress to discover the genetic cause for the 50% of Noonan syndrome not attributable to *PTPN11* abnormalities.

For more information, please contact Dr. Bruce Gelb at the Mount Sinai School of Medicine.

Tel: 212-659-6705. E-mail: bruce.gelb@mssm.edu

Research Opportunities



We need research volunteers! We are looking for adults and teenagers with Noonan syndrome who would be willing to undergo a skin biopsy (removal of a small piece of skin from the arm). We can also enroll children who are undergoing heart surgery. The purpose of the study is to understand the effects of the gene changes in Noonan syndrome on the way in which cells work. For more information, please contact Dr. Bruce Gelb at the Mount Sinai School of Medicine.

Tel: 212-659-6705. E-mail: bruce.gelb@mssm.edu

Articles (Continued from page 6)

cardiomyopathy in Noonan's syndrome.
Hum Pathol (United States), Mar 2003, 34 (3) p282-4

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Spectrum of mutations in PTPN11 and genotype-phenotype correlation in 96 patients with Noonan syndrome and five patients with cardio-facio-cutaneous syndrome [In Process Citation] Eur J Hum Genet (England), Feb 2003, 11(2) p201-6

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Different mutations in the NF1 gene are associated with Neurofibromatosis-Noonan syndrome (NFNS) [In Process Citation] Am J Med Genet (United States), May 15 2003, 119A(1) p1-8

13. Kondoh T, Ishii E, Aoki Y, et al.
Noonan syndrome with leukaemoid reaction and overproduction of catecholamines: a case report [epub ahead of print] [Record Supplied By Publisher]
Eur J Pediatr (Germany), May 9 2003, p

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Noonan syndrome [In Process Citation]
Indian Pediatr (India), May 2003, 40(5) p431-2

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Etiopathogenic analysis of the caries on three patients with Noonan Syndrome [In Process Citation] Med Oral (Spain), Mar-Apr 2003, 8(2) p136-42

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Noonan's syndrome and autoimmune diseases [In Process Citation]
J Pediatr Endocrinol Metab (England), Feb 2003, 16(2) p217-8

17. Agras PI, Baskin E, Sakallioglu AE, et al.
Neurofibromatosis--Noonan's syndrome with associated rhabdomyosarcoma of the urinary bladder in an infant: case report [In Process Citation] J Child Neurol (United States), Jan 2003, 18(1) p68-72

18. Schollen E, Matthijs G, Gewillig M, et al.
PTPN11 mutation in a large family with Noonan syndrome and dizygous twinning [In Process Citation]
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The unusual association of three autoimmune diseases in a patient with Noonan syndrome.
J Adolesc Health (United States), Jan 2003, 32(1) p94-7

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Somatic mutations in PTPN11 in juvenile myelomonocytic leukemia, myelodysplastic syndromes and acute myeloid leukemia [In Process Citation]
Nat Genet (United States), Jun 2003, 34(2) p148-50

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Hematological findings in Noonan syndrome [In Process Citation]
Rev Hosp Clin Fac Med Sao Paulo (Brazil), Jan-Feb 2003, 58(1) p5-8

HAVE FUN, RAISING FUNDS

Spring is here! We can help you help us. Clean out that bedroom closet, garage or basement for things to sell on eBay- all for TNSSG Inc.. By selling just one item per month, now through the end of September, you can help raise thousands of dollars, or more to help our continued efforts.

Just think of the benefits, you will have more space, and rise funds for TNSSG.

It is so easy to sell your goods on eBay–Here are some ideas to get you started:

- **8-TRACKS**
- **ART**
- **BARBIE DOLLS**
- **BASEBALL CARDS**
- **BEANIE BABIES**
- **CABBAGE PATCH KIDS**
- **COINS**
- **COLLECTIBLES**
- **COMPUTER EQUIPMENT**
- **CONCERT MEMORABILIA**
- **COSTUME JEWELRY**
- **DVD MOVIES**
- **FISHING TACKLE**
- **FURNITURE**
- **HATS**
- **HISTORIC PHOTOS**
- **JEWELRY**
- **MUSICAL INSTRUMENTS**
- **OLD BOARD GAMES**
- **OLD BOOKS**
- **OLD FURS**
- **OLD TOYS**
- **POGS**
- **POSTCARDS**
- **RAILROAD PARAPHERNALIA**
- **RECORD PLAYER**
- **ROTARY PHONES**
- **SKIS**
- **SPORTS EQUIPMENT**
- **STAMPS**
- **VIDEO GAMES**
- **VINTAGE CLOTHING**
- **VINTAGE POSTERS**
- **VINYL RECORDS**

These are just a few ideas the possibilities are endless.

1 Register on eBay: http://www.eBay.com it's quick and it's free. Create your personal eBay seller account. You will need a credit or debit card and your checking account details. Rest assured your information is kept private on eBay's secure servers.

2. List item to sell. Describe your item and add photo [optional but helpful]. Search for similar items on eBay for descriptions and value. From as little as 30 cents to $3.30 you can list your item.

3. For the fastest and easiest payment option, choose PayPal to collect payments from your buyer, be sure to include shipping costs. Our PayPal account for the buyer is:
donation@noonansyndrome.org
You may also choose to accept personal checks or Money Orders

4. Your item is now ready, let the bidding begin!

5. When your auction ends, both you and your buyer will receive an email notice. Depending on your payment instructions, the information you receive in this email will include your buyer's shipping address and payment method. Once you receive a confirmation from Pay Pal that buyer has paid item + shipping costs directly to the TNSSG PayPal account:
donation@noonansyndrome.org
ship item to buyer. You may ship your item!

For Personal Check or Money Order Buyer mails payment to you. Cash payment. Once payment clears, ship item to buyer. Then, make donation to TNSSG, Inc. send your personal check to

TNSSG, Inc.
P O Box 145
Upperco, MD 21155

Congratulations, you contributed to TNSSG, Inc.!

Questions: eBay has a very easy eBay HELP button with more details on selling and invoicing: http://www.eBay.com

If your closet is clean you can send a monetary donations to:

TNSSG. Inc.
P O Box 145
Upperco, MD, 21155

Get involved. Tell your family and friends about selling items on eBay for TNSSG.

Please note, eBay does charge a Final Value Fee. When your listing ends, you will be charged a Final Value Fee based on the final sale price (or "final value") of your item: The Final Value Fee is calculated per item sold, based on the final sale price of the item.

The final value is the closing bid. Remember, there is no Final Value Fee charged if the reserve is not met.

DESCRIPTION (FILL IN AND SEND WITH ORDER FORM)	#	$$ DUE
Growth Hormone in Noonan syndrome by Kees Noordam The studies presented in this thesis were funded by Pharmacia B.V., Woerden. Publication of this thesis was financially supported by Pharmacia B. V. , Woerden and FBW Kindergeneeskunde Nijmegen. - **$25.00 (Book)**		
Noonan Syndrome A Parents Guide- Excellent booklet written in easy to understand language, produced by The Noonan Syndrome Support Group, Inc.(1 Free with each parent package) - Additional books **$4.00 (Book)**		
Cómo Comprender El Síndrome De Noonan Guía Para Los Padres- Preparada Para El Grupo De Ayuda Del Síndrome De Noonan (Noonan Sydrome Support Group, Inc.) (1 Free with each parent package) - Additional books **$4.00 (Book)**		
The Many Faces of Noonan Syndrome Excellent booklet with photographs of people of various ages affected by Noonan syndrome. Produced by The Noonan Syndrome Support Group, Inc. -**$4.00 (Book)**		
Another Noonan Connection- A cookbook of favorite recipes (over 250 pages) from the families and friends of TNSSG, Inc. Produced by The Noonan Syndrome Support Group, Inc. **$10.00 (only**		
The Noonan Support Group Pin - This pin is shaped like a heart and within the heart is a box with the group's logo. The pin is set on a gold background. **$2.00**		
Noonan-syndrome Characteristics Development, and Intervention - by Dr. Stephen von Tetzchner, Center For Rare Disorders, The National Hospital, Norway: This book is a comprehensive guide that covers the many aspects of Noonan syndrome. **$20.00** "IF YOU ARE AFFECTED BY NOONAN SYNDROME YOU SHOULD OWN THIS BOOK" "THE MOST COMPREHENSIVE SOURCE , OF INFORMATION ABOUT NS		

(Compact Disks) Recording of individual speakers who presented at the 2002 Conference. Each disk is approximately 60 minutes long. $5.00 each	**# @ $5.00**	**$$ DUE**
CD: Dr. Steve Band: Director, Division of Pediatric Psychology and Neuropsychology, Mount Washington Pediatric Hospital One of the most common questions he has been asked by parents is how they can promote healthy self-esteem in their children with special needs. It is an important question that remains relevant today.		
CD: Dr. Thomas Baumgardner: Clinical neuropsychologist. He specializes in the assessment and treatment of children and adolescents with behavioral, learning, emotional, and cognitive disabilities. He is part of a multidisciplinary group of psychologists, psychiatrists, social workers, and behavioral pediatricians located at Johns Hopkins at Greenspring Station.		
CD: Dr. Bruce Gelb: The Arthur J. and Nelly Z. Cohen Professor of Pediatrics Professor of Human Genetics. Director Cardiovascular Genetics Program, Mt. Sinai School of Medicine, New York, NY.		
CD: Dr. Bryan Hall: Professor Emeritus Department of Pediatrics Division of Genetics and Dysmorphology, University of Kentucky, Lexington, KY. Special diagnostic interests: multiple congenital anomaly syndrome identification and teaching thereof.		
CD: Dr. Dolores Njoku: Pediatric Anesthesiologists on Staff at Johns Hopkins, Baltimore, MD. Research interests include the Immune effects of anesthetics		
CD: Dr. Jacqueline Noonan: University of Kentucky Medical Center, Lexington, KY. Professor of Pediatrics in the Division of Pediatric Cardiology.		
CD: **Dr. Alicia Romano:** Director of Pediatrics Endocrinology Our Lady of Mercy Medical Center Bronx, NY.		
CD: Dr. Frank E. Schafer :Associate Professor of Pediatrics, MCP Hahnemann University School of Medicine; Director General Hematology Clinic and Bleeding Disorders Program, St. Christopher's Hospital for Children Philadelphia, PA.		
CD: Dr Judith Allanson: Clinical geneticist with a longstanding interest in Noonan Syndrome. Her research focuses on the face: how it changes with age; how we evaluate facial appearance; how important knowledge of facial change is in the diagnosis of Noonan Syndrome.		

TNSSG ORDER FORM: PLEASE PRINT

DATE________________

NAME:
____

MAILING ADDRESS: ____

CITY: ________________________________, STATE: _______ ZIP: ___________

AREA CODE : ______TELEPHONE NUMBER :____________________________

____ Check Enclosed (MAKE PAYABLE TO: TNSSG, Inc.)

OR

For your convenience we now accept credit cards:
Check card type: Discover ___ Visa ___ Master Card _____

____

PRINT NAME AS IT APPEARS ON CREDIT CARD

_____ / ____
4 DIGIT EXPIRATION DATE : **MONTH / YEAR**

__ __ __ __ - __ __ __ __ - __ __ __ __ - __ __ __ __
ACCOUNT NUMBER

____
AUTHORIZED CARDHOLDER SIGNATURE

IN US DOLLARS PLEASE INDICATE :
TOTAL FOR ITEMS PURCHASED $ _________
TOTAL SHIPPING COST # ITEMS X $ 3.00 = $ ________
TAX DEDUCTIBLE CONTRIBUTION + $ _________
TOTAL AMOUNT = $_________

Mail completed order form to:
TNSSG, Inc.
P O Box 145
Upperco, MD 21155

QUESTIONS?
Telephone:
888-686-2224
410-374-5245
Email:
wandar@bellatlantic.net
ORDER ON LINE:
HTTP://WWW.NOONANSYNDROME.ORG

2002-2003 TNSSG, Inc. MEMBER/CONTACT RENEWAL FORM

After you contact us for information, we add you on our mailing list to receive the Noonan Connection and other mailings. We have no recurring dues or fees for membership. Therefore, depend on your generous contributions to support our efforts to educate people about Noonan syndrome. . The only way of knowing if you are still interested in receiving this information is to periodically ask you to confirm your desire to receive information from The Noonan Syndrome Support Group, Inc. Our first Newsletter mailed in the fall of 1996 went to fewer than 50 people.

The Summer 2002 Noonan Connection was mailed to more than 1,600 people in 25 countries, again, made possible because of your generous contributions. We need to establish a way of assuring that we don't mail newsletters to people who are no longer interested in receiving information about Noonan syndrome and our group. Therefore, if you wish to remain on our mailing list, **you must** fill out and return this **MEMBER/ CONTACT RENEWAL FORM** . Additionally, the only way you can be added to or remain on our **People You Can Call List** (see attached) is to fill out a **MEMBER/CONTACT RENEWAL FORM** .

The people listed on the People You Can Call List have given TNSSG, Inc. permission to publish their contact information. Please be courteous when calling and remember to check time zones. The only way we can publish this information is with written permission.

***If you don't return this* 2002-2003 TNSSG, Inc. MEMBER/CONTACT RENEWAL FORM, *you will be removed from our files and our* People You Can Call List.**

PLEASE PRINT Only those people who did not return there renewal form in the last 3 months, need to fill out this form.

2002- 2003-MEMBER/CONTACT RENEWAL FORM

____ I want to continue to receive information from The Noonan Syndrome Support Group, Inc.
____ I want TNSSG, Inc. to send an information package to the following person.
____ I want to continue to be included on the People You Can Call List*.
____ I want to be added to the People You Can Call List*.

♥LAST NAME: ____________________________________ FIRST NAME: ___________________________
♥MAILING ADDRESS:_______________________ ____
♥CITY: ________________________ STATE:_____ZIP:_________ COUNTRY: ______________
♥PHONE NUMBER:______ - _____________________ E-MAIL ADDRESS:______________________________
♥ UPDATE/CHANGE: ___________

HOW ARE YOU AFFECTED? (please indicate)

♥_____Directly Affected (an adult with NS):_____Indirectly Affected (a parent or guardian of a child with NS.)
♥_____G.R.A.N.S(Grandparents Relatives Advocating for NS.): ____N.A.P.(Noonan Angel Parents/ People). (These people have had someone they love die from complications associated with NS.):_____:Professionals (doctors, teachers etc. taking an interest in those affected by NS)

CHOOSE ONE IF YOU ARE PART OF THE PEOPLE YOU CAN CALL LIST

♥*ADD MY NAME TO THE CONTACT LIST AND WEB SITE CONTACT PAGE : ______
♥*ADD MY NAME TO THE CONTACT LIST ONLY (published in The Noonan Connection): ____
♥NAME OF PERSON AFFECTED WITH NS:_____________________DATE OF BIRTH: ___/___/___
___ Inherited: _____Sporadic Occurrence/ Mutation
♥NAME OF PERSON AFFECTED WITH NS:_____________________DATE OF BIRTH: ___/___/___
♥___ Inherited: _____Sporadic Occurrence/ Mutation
♥NAME OF PERSON AFFECTED WITH NS:_____________________DATE OF BIRTH: ___/___/___
♥___ Inherited: _____Sporadic Occurrence/ Mutation

I HEREBY AUTHORIZE RELEASE OF MY NAME, MY CHILD'S NAME AND THE INFORMATION ON THIS FORM TO THE NOONAN SYNDROME SUPPORT GROUP AND THE FAMILIES INVOLVED THEREIN.
♥SIGNATURE: ____ DATE: ___________

Return completed form to:
TNSSG, Inc.
P O Box 145,
Upperco, MD 21155

QUESTIONS?
Telephone: 888-686-2224 OR 410-374-5245
Email: wandar@bellatlantic.net
HTTP://WWW.NOONANSYNDROME.ORG

Our Web site has been visited by over 100,000 people! We have given people another source for information! If not for the Web site, and all the attached material, people would still be looking for information about Noonan syndrome. A special thank you to Eugene K. As our former Web manager, he has created a place I am glad to call ours.

We're on the web! http://www.noonansyndrome.org

WANT TO 'TALK' TO MORE THAN 200 PEOPLE AFFECTED BY NOONAN SYNDROME?

- ♥ ADULTS WITH NOONAN SYNDROME
- ♥ FAMILY MEMBERS
- ♥ YOU CAN JOIN FOR FREE!
- ♥ SIMPLY JOIN OUR GROWING 'LIST' OF PEOPLE WHO HAVE ALREADY SUBSCRIBED TO OUR INTERNET 'LIST' (DISCUSSION FORUM).
- ♥ DETAILS ON HOW TO JOIN AND AVAILABLE OPTIONS FOLLOW...

We operate a list service (discussion forum). If you would like to subscribe, and be a part of our family, send an e-mail to

♥ **listserv@home.ease.lsoft.com**

with only the following command (words) in the message body:

♥ **subscribe noonan-syndrome**

That is it, nothing in the subject line, and only the words subscribe noonan-syndrome in the message body. (AOL subscribers also need to type a period "." in the subject line)

- ♥ To post a new message to the mailing list, address it to: NOONAN-SYNDROME@HOME.EASE.LSOFT.COM
- ♥ To receive a digested version, send an e-mail to: LISTSERV@HOME.EASE.LSOFT.COM in the message body write (without quotes) "SET NOONAN-SYNDROME DIGEST"
- ♥ To get a short acknowledgement that your message has been posted send a message to: LISTSERV@HOME.EASE.LSOFT.COM in the message body write (without quotes) "SET NOONAN-SYNDROME ACK NOREPRO"
- ♥ To turn off acknowledgements send a message to: LISTSERV@HOME.EASE.LSOFT.COM in the message body write (without quotes) "SET NOONAN-SYNDROME NOACK NOREPRO"
- ♥ To Un-subscribe or Sign off, send a message to: LISTSERV@HOME.EASE.LSOFT.COM in the message write (without quotes) "SIGNOFF NOONAN-SYNDROME"

Chit chat list

This list is intended for people affected by Noonan syndrome who want a place to communicate or chit/chat. Birthdays or poems, or anything that is important to your family.

Subscribe*:tnssgchitchat-subscribe@yahoogroups.com
Post message: nssgchitchat@yahoogroups.com
Unsubscribe:tnssgchitchat-unsubscribe@yahoogroups.com
List owner:tnssgchitchat-owner@yahoogroups.com

We also have a list for

- ♥ **Adults with Noonan syndrome.**
- ♥ **Kids with NS (moderated by an adult with NS)**
- ♥ **TNSSG information list**

For information on how to get on any of these lists, please contact wandar@bellatlantic.net

HERE ARE SOME OF THE WAYS YOU CAN SUPPORT TNSSG, INC.

- **MAKE A TAX DEDUCTIBLE CONTRIBUTION**
- **DESIGNATE TNSSG, INC. ON YOUR PLEDGE FORM WHEN GIVING TO THE UNITED WAY OR COMBINED FEDERAL CAMPAIGN**
- **ARRANGE FOR A MATCHING FUNDS DONATION THROUGH YOUR EMPLOYER**
- **HONOR A LOVED ONE OR CLOSE FRIEND WITH A BEQUEST IN THEIR NAME**
- **ORGANIZE AND PLAN A FUND RAISER**
- **PARTICIPATE IN A FUND RAISER**
- **MAIL INFORMATION TO OTHERS ABOUT NOONAN SYNDROME**
- **TALK TO OTHERS ABOUT NOONAN SYNDROME**
- **BECOME A CONTACT, FILL OUT THE ATTACHED CONTACT RELEASE INFORMATION FORM.**

DO SOMETHING -WE NEED YOUR CONTINUED SUPPORT- BE A PART OF THE SOLUTION
"The only disability is a bad attitude."

THE NOONAN SYNDROME SUPPORT GROUP.
PO BOX 145
UPPERCO, MD 2115

NON-PROFIT
U.S. POSTAGE PAID
UPPERCO, MD
PERMIT # 13

IS THIS YOUR LAST COPY OF THE NOONAN CONNECTION?

INFORMATION YOU CAN USE FROM TNSSG. Inc.

Including ...

Growing up with Noonan syndrome...

Disability and Discrimination in Noonan syndrome...

3000 Eggs hidden in MN...

Chylothorax (lymph fluid leaking into the pleural(lung) cavity...

CONFERENCE QUESTIONNAIRE Plus much more.