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**STICKLER INVOLVED PEOPLE  
SEPTEMBER 2013 NEWSLETTER**

## Coordinator's Comment

Some of you know that my family has been through a rough patch for 15 months. We are seeing the light at the end of the tunnel- hope - and thank all of you for good thoughts. Again, I am reminded that we are all in phases; that we have to support and enjoy those dear to us through the good and the bad (like wedding vows?).

I spent many conversations with a lady from Poland in the last month. She really feels the aloneness and the "not knowing" part of the syndrome. So, if you have passed that phase, take a moment to be grateful! I sure had a good "walk down memory lane", trying to help her. I really was reminded how far we have come, how comfortable we have become with Stickler syndrome and its symptoms. I hope this is a long phase, with no new symptoms. Best wishes to you

## ANNUAL CONFERENCE NEWS

We had a great 2013 conference in Las Vegas, with super speakers this year. Maybe, I say that every year, but these folks were so dedicated to their patients that it was amazing to me. We are in the process of determining the location of our **2014 Annual American Stickler Involved People's conference** which is held the second weekend in July.

Please have a good thought for some researchers that are currently in touch with SIP. If the University of Colorado Medical Center receives a grant they applied for, we will need MANY, MANY Stickler bodies at the next conference, which could be in the Denver area. We will know more in January 2014 and plan the conference from there.



There are some special circumstances to be worked out for a special activity for the support group if this Colorado location works out. We do have a second and third location scouted out if our Colorado plans do not come together as expected. Please mark your calendars to attend the next SIP conference scheduled for July 11-13, 2014.

**Just FYI:** one thought that the SIP Board discussed was to combine our conference with another connective tissue disorder conference. We looked at this idea for several years. There could have been several advantages like cheaper hotel, less planning for our committee, some speakers would overlap. The final decision was that SIP would lose the *family feel* of our conferences and that would be too great a loss. We put it on hold for now.

## **SIP SCHOLARSHIP**

Dr. and Mrs. Stickler donated the start-up money for a fund to provide high school seniors, who have a diagnosis of Stickler syndrome, with a four year scholarship. Applications are available on line and due June 15 each year.

On behalf of the Scholarship Committee and the entire Stickler Involved People organization, we are pleased to report that **David Baylies** is the 2013 recipient of the Dr. Gunnar B. Stickler Scholarship. Although there were several highly qualified candidates, the Scholarship Committee was very impressed with his many accomplishments and obvious drive to excel. We wish David continuing success as He begins his studies at Boston University.

**David's response:** "THANK YOU! I truly appreciate this, and I will not forget it. Living with Stickler Syndrome is tough, but it's just become a part of me. Judging from how great this summer has already been, I am hopeful that my college experience will be just as rewarding as, if not more than, high school, and mostly uninhibited by my affliction".

***We thank the Scholarship committee; Jim Brown, Lori Vickery and April Murphy for their review of applications and selection. Well done!***



Please consider making a donation to the **Gunnar B. Stickler Scholarship** fund. The new address for the scholarship fund is:

Stickler Involved People  
Gunnar B. Stickler Scholarship Award  
PO Box 775  
Cologne, NJ 08213

**Note:** [Cappex.com](http://Cappex.com) is a resource where students can apply to thousands of additional scholarships including the Stickler B. Gunnar scholarship. Check it out for your student. Then click on “Get Scholarship Matches” and register.

### **An easy way to help**

[IGIVE.com](http://IGIVE.com) has sent Stickler Involved People over \$900 since it started. When you buy from the web, please remember to start with [IGIVE.COM](http://IGIVE.COM) and make some money for us. There are some places that have special offers for [IGIVE.COM](http://IGIVE.COM) subscribers. We get \$5 for each new person who places an order through [IGIVE.COM](http://IGIVE.COM). So, now is the time to look it over, subscribe, give Stickler Involved People as your cause, and place an order. You are helping SIP and may save yourself some money and time. REMEMBER IGIVE.COM as you begin your Christmas shopping!

### **Genetic Alliance Announcement**

In a time when searching for quality health information can be like searching for a needle in a haystack, Genetic Alliance is thrilled to announce the launch of its newest public resource, [Genesinlife.org](http://Genesinlife.org). Genes in Life harmonizes many existing resources and tools in a one-stop source for general information related to health and genetics services.

On [GenesInLife.org](http://GenesInLife.org), individuals and families can learn how and why to collect a family health history and share it with a healthcare provider; understand the differences between various types of genetic testing and services, and figure out which genetics professionals to consult. The site will also host interactive features including blog campaigns encouraging comments and discussion as well as an “ask the experts” page, allowing users to request more information about the topics they care most about. Check us out at [www.genesinlife.org](http://www.genesinlife.org).



## **Stickler Syndrome Clinic Announcement**

We are pleased to announce that Joseph V. Thakuria, MD, MMSc, Medical Genetics and Metabolism Attending Physician, Medical Director, Personal Genome Project at MGH Center for Human Genetics Research with the Massachusetts General Hospital has agreed to take over the clinic. He spoke at the Las Vegas SIP conference and shared his new ideas about the clinic.

The clinic day may change so check with Sandy. We wish Ruth Liberfarb the best and give her a GREAT THANKS for all she has done for Stickler Involved People and for all persons with Stickler syndrome.

## **TO VISIT THE STICKLER CLINIC**

Patients will need to book the appointment with Sandy Massalski at: 617-726-1561. The patient has to pre-register with Mass. General Hospital to give billing information, and to get a hospital number.

Regularly scheduled clinic appointments are usually on Thursday afternoon from 1-5pm. The clinic can see 3- 4 people. The patient needs to submit medical records in advance. The Genetics Unit has a training program for physicians doing fellowships in Med. Genetics. Some of these "fellows" might want to participate in the clinic.

The clinic is not free, but it is a "dream come true" for persons with Stickler syndrome. Recently, I was able to answer someone that this is THE one place to visit with all the experts you need. Be sure to make your needs known when you call for an appointment.



## **FOR PATIENTS THAT REQUIRE GENETIC TESTING FOR OCULAR STICKLER SYNDROME TYPE I/II.**

CONTACT **THE MATRIX DNA DIAGNOSTICS LAB** AT TULANE UNIVERSITY TO ORGANIZE YOUR INDIVIDUALIZED TESTING NEEDS OR FOR ADDITIONAL INFORMATION. Charlene M. Wille, MBA, BSMT (ASCP), Laboratory Supervisor, Matrix DNA Diag. Lab, Tulane University Health Science Center (HSC), Phone: (504) 988-7706 Fax: (504) 988-7704  
<http://tulane.edu/som/regenmed/services/dnadiagnostics.cfm>

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- Flexible billing option including: insurance, volume, advance payment, research study and self-pay discounts.
- NO up-front payments required.  
We accept MOST insurance policies and plans.
- Over a decade of expertise.
- Complete sequencing of both COL2A1 and COL11A1 available for prenatal, infant, juvenile and adult samples.

**Please visit our website above to download our requisitions.**

## **SOCIAL SECURITY DISABILITY INCOME HELP**

If you need any assistance with your Social Security Disability claim, you can contact the following individuals for information and suggestions.

Jon Rodis            [WSALMGCDJM@aol.com](mailto:WSALMGCDJM@aol.com)  
Kathy Cashell      [SANDCREEKSTUIO@bresnan.net](mailto:SANDCREEKSTUIO@bresnan.net)  
Rick Bishop        [FIFIBISH@aol.com](mailto:FIFIBISH@aol.com)

## **A New SIP Public Service Announcement Video**

***SIP has finished a second public service announcement entitled "Graham Elder's Story" for YouTube. His parents, Amanda and Tim Elder, shared their plight to discover what caused Graham's hearing and vision disorders. Tim Elder came across our web site [www.sticklers.org](http://www.sticklers.org) which helped him and Amanda to get a proper Stickler syndrome diagnosis for Graham. Preview this touching story at <http://www.sticklers.org/sip2/content/view/28/39/>.***



## MEET A SIP TEEN

Some of you had the opportunity to help Eline Van Oostrum, an 18 year old with Stickler syndrome, complete her senior research project about Stickler syndrome. She received 100 responses and the data has been analyzed and edited for review. The link on our web site is:

[http://www.sticklers.org/sip2/images/stories/Surveys/  
ElinevanOostrumSurvey%201b.pdf](http://www.sticklers.org/sip2/images/stories/Surveys/ElinevanOostrumSurvey%201b.pdf)

## \*\*NEW\*\* Doctor's Referral List Project \*\*NEW\*\*

A member of the stickler list serve is compiling a list of specialists, to share with those interested in finding a physician in their area. If you have specialists whom you consider knowledgeable about Stickler syndrome (even if you trained them), send an email to:

[Doctorlist949@gmail.com](mailto:Doctorlist949@gmail.com)

Please include your email address (which will not be shared), the doctor name, address and phone, AND the type of specialist they are.

By the first of the year, we think that you will be able to request specialists, in your area, recommended by someone in the group.

We do have some listings now, because this was submitted to the list serve and to the Facebook groups a couple of months ago. Please be patient, as this is a one person volunteer project.

## Stickler DVDs

The 1st film is now available on <http://www.sticklervideo.org> , for free. The 2nd film, "Finding Hope" is now on the same website.

## GENETIC ALLIANCE TIP

Physical activity is an important part of staying healthy. What kinds of activities should kids—and adults—be doing?

The [Physical Activity Pyramid for Young Children](#) from the Twin Falls, Idaho South Central Public Health District provides examples of ways children can be physically active. [Click here](#) to view the pyramid in Spanish.



This pyramid can be adapted to fit the needs of anyone, regardless of their physical abilities or limitations. Visit [My Child Without Limits](#) for more ideas on physical activities and adapted exercises to help encourage all children to get moving!

### Quick Tips on How to Get Kids Moving

- Switch it up! Variety can help encourage participation.
- Involve families and parents.
- Help them learn while they play.
- Go outside.
- Learn what they like and help them do it.
- Emphasize participation in activity and enjoyment over competition.
- Model Behavior: Set a good example.
- Join in! Participate with the children in physical activity.
- Introduce new skills and movements.
- Provide time for unstructured play each day.
- Don't use physical activity as punishment.
- Plan activities for the whole family or group
- Provide encouragement.

## Additional Resources

### [National Center on Health, Physical Activity and Disability \(NCHPAD\)](#)

This organization provides a wealth of resources on physical activity for people with different medical conditions and disabilities. Search the site for activities specific for children or adults or by condition on NCHPAD's homepage.

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### [2013 Stickler Involved People Conference, Las Vegas, NV Attendees](#)

