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STICKLER INVOLVED PEOPLE
December 2013 NEWSLETTER

Coordinator's Comment

We are all so blessed. No matter how many struggles we are given, we are blessed with our abilities and the ability to seek help. We have so much and all of us can share. So, if you have a roof over you, be Glad. I am sitting out in minus one wind chill, watching my dogs run. It is so cold that my I pad's AutoCorrect will not work. But, I am blessed with dogs, with a yard and my I pad. Plus, I have the option to go in the house and get warm. I chose to do that now, because my Internet mittens are too cold to work on my I pad screen. No matter how small our blessings may be, there are others worse off than us. So, in this holiday season, find time to realize your blessings and to share at least one blessing with others. Happy Hanukkah and Merry Christmas! Pat **(P.S. Shop IGIVE.Com)**

ANNUAL CONFERENCE NEWS

We are **STILL** 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.

If the University of Colorado Medical Center receives a grant they applied for earlier, we will need MANY, MANY Stickler bodies at the next conference, which could be in the Denver area. We will know more in **March 2014** and plan the conference from there. If this Colorado location does not work out, we do have a second and third locations scouted. Please mark your calendars to attend the next SIP conference scheduled for July 11-13, 2014.



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.

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** 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.

IGIVE.COM is an easy way to help SIP

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** and make some money for us. There are some places that have special offers for **IGIVE.COM** subscribers. We get \$5 for each new person who places an order through **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 do your Holiday shopping! **SPECIAL NOTE:** a recent example of IGIVE shopping; three Stickler giving shoppers netted \$15.10 to SIP. Every dollar counts!!!! Please remember to shop through IGIVE.com this Holiday Season.



URGENT ANNOUNCEMENT!

FDA Tells Google-Backed 23andMe to Halt DNA Test Service!

23andMe Inc., the Google Inc. backed DNA analysis company co-founded by Anne Wojcicki, was told by U.S. regulators to halt sales of its main product because it's being sold without marketing clearance or approval. The Saliva Collection Kit and Personal Genome Service, or PGS, tells users whether they carry a disease, are at risk of a disease and would respond to a drug. Most of the uses fall into the category of a medical device and require Food and Drug Administration approval, the agency told the Mountain View, California-based company in a Nov. 22 letter made public. Read more of the article at: <http://www.bloomberg.com/news/2013-11-25/fda-tells-google-backed-23andme-to-halt-dna-test-service.html>

Genes in Life Highlights Whole Genome Sequencing

Have questions about whole genome sequencing? Genetic Alliance invites you to visit GenesInLife.org throughout November and December to catch our spotlight on this exciting topic and have your questions answered by our whole genome sequencing experts!

For the next two months Genes in Life will feature professionals with experience in all different aspects of whole genome sequencing from medicine to biotechnology to ethics. Readers will be able to pose their questions to our featured experts using our [Ask the Experts feature](#), and answers will be promptly provided via email and posted to our [FAQ page](#). The [Genes In Life blog](#) will delve into the technology and science behind whole genome sequencing and provide a space for interactive discussion on how whole genome sequencing might play a role in the future of health and healthcare.

Visit Genes in Life throughout the next two months to participate in our spotlight on whole genome sequencing. We're excited to hear your thoughts and answer your questions on such a captivating topic in genetics and health.



Please help us spread the word about Genes in Life and our current feature on whole genome sequencing by sharing this email with anyone who may be interested or have questions. We also encourage you to link to us on your website and join us in this effort to connect individuals with resources on genetics, genetics services, and health. If you would like to partner with us on Genes in Life, or just want to find out more, please contact Rachel Koren at 202.966.5557 x213 or rkoren@geneticalliance.org.

Sincerely,
The Genes In Life Team, Genetic Alliance

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.....

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.



PLEASE SIGN THIS PETITION TO KEEP THE NIH/NIA CONNECTIVE TISSUE STUDY ACTIVE

We received an email from Kari Ulrich and she wanted to reach out to the sticklers community about an important study that has been recently been closed. This affects stickler patients. Please open and review this petition to reinstate the connective tissue studies and sign it for all our benefit.

The petition is close to having 5,000 signatures. It is Kari's goal to get 10,000 signatures to present to NIH/NIA. With this, it is her expectation that the study will be reinstated and we will be assured that our data and specimens will not be lost. She remains confident that with each signature we are spreading awareness.

To sign this Petition, go to: <http://chn.ge/1bsPjZo>

Click here to read more about the Study's closure: [NIA Intramural Research Program - Volunteers for Clinical Study](#)

A Second 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 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/>.

UPDATE - UPDATE - UPDATE

"We wanted to share this with you. The local news station that aired our story in March saw the YouTube video on Sticklers and aired a story on it on the evening news in Rochester, NY tonight. The link is below. The channel posted a link to the SIP website and Graham's video on their website and referenced it during the story on the news.



Thanks so much for all of your support. We hope to continue to advocate for Graham and spread information about SS to everybody we can possibly reach. We look forward to the conference in July!”

Best,
Amanda, Tim, Julian and Graham Elder

See the newsreel here:

<http://www.rochesterhomepage.net/story/baby-grahams-story/d/story/14Grzf23CEOgf2VltQ>

Genetic Alliance News

Have you checked out the ALL NEW Advocacy ATLAS?

Genetic Alliance, in partnership with Family Voices and Parent to Parent USA, announced last week the launch of a new online resource for individuals with special healthcare needs and their families. The Advocacy ATLAS: Accessible Tools for Leadership and Advocacy Success, features over 250 tools and resources to help new and experienced advocates communicate their needs around services, support, and access.

These resources aim to help individuals with special healthcare needs and their families navigate a world often full of barriers and inaccessibility. The Advocacy ATLAS features resources and perspectives on youth leadership, transition to adulthood, insurance and financial assistance, communicating about your health, access to healthcare, and more. Genetic Alliance, Family Voices, and Parent to Parent USA will continue to reach out to partners for new resources to expand the utility of the Advocacy ATLAS with new and experienced advocates alike.



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

A member of the stickler list serve has compiled 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

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

You can now request specialists, in your area, recommended by someone in the group by contacting Pat Houchin at sip@sticklers.org. She will respond to you with listed specialist(s) in your area. Not all major Cities are represented at this time.

Stickler DVDs

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

Additional Resources

(click on blue heading for more information)

[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.



[Sibling Support Project](#) The Sibling Support Project is a national effort dedicated to the life-long concerns of brothers and sisters of people who have special health, developmental, or mental health concerns.

[Tips for Protecting your Knees](#) studies show that exercise can help reduce knee pain and stiffness, strengthen the surrounding muscles, and improve your flexibility and range of motion.

[NEW tablet-based Pediatric Family Health History Tool](#) this is an electronic pediatric family history tool. It is designed to support the provider in family history risk assessment and genetic evaluation. Also to Identify additional evaluation, preventative services, or personalized management for children who are at increased risk for health conditions based on family health history.

WE ARE HEARD AROUND THE WORLD

Here is a note from a Student in **Saudi Arabia**. Our web site reaches many beyond the United States!

Hello there,

I'm Shahd Benafif, a student in Dar Al-Hekma College in Jeddah, Saudi Arabia. My program of studying is Speech, Language and Hearing Sciences and I actually have this presentation about Stickler Syndrome in my Audiology class where I need some pictures to support the body of knowledge I gathered. So can I use some pictures from your website, please? Thank you for your time.

Additionally, in an unrelated communication, SIP got a request from **Japan**, asking for contact with a person in one of the Stickler DVDs.