



Genetic Transcriptions

Newsletter of the AAP Section on Genetics and Birth Defects

October 2003

From the Chair

I cannot believe that four years have passed from the time I wrote my first column as Chair of the Section on Genetics and Birth Defects. In all, I have been a member of the SOGBD executive committee for ten years. I have learned a great deal during this time and, hopefully, contributed to the functioning of the SOGBD.

One thing I have come to appreciate to a much greater degree is what a phenomenal organization the AAP is. By starting with the desire to improve the healthcare of children, the AAP recognizes that this can only be accomplished by casting a wide net. In order to meet its charge, the AAP functions at multiple different levels, nationally, regionally, and locally within communities. The Academy recognizes that if the needs of pediatricians and pediatric specialists are not properly met, their ability to care for children will be impaired. Therefore, there are significant efforts made to improve billing and coding, enhance education at a primary care and tertiary care level, and to influence the training of future pediatricians. There are also significant efforts to enhance and expand both clinical and laboratory research related to improving healthcare for children.

Most pediatric subspecialists have found a comfortable home in the AAP. Most pediatric subspecialties are certified through the American Board of Pediatrics and, therefore, have a natural home in the AAP. There are exceptions, however. Allergists have their own academy, as do cardiologists. Medical genetics is in a unique position. We have our own specialty board and our own College (the American College of Medical Genetics). In addition, membership fees for both the ACMG and the AAP can approach \$1,000 yearly.

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Cincinnati, Ohio

Ronald P. Bachman, MD
Executive Committee Member
Oakland, California

Mira Irons, MD
Executive Committee Member
Boston, Massachusetts

Hope Northrup, MD
Executive Committee Member; Membership Chair
Houston, Texas

Robert A. Saul, MD
Executive Committee Member; Program Chair
Greenwood, South Carolina

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Nominations Committee Chair
Burlington, Vermont

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Subcommittee on Research Grants Chair
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David B. Flannery, MD
Subcommittee on Outcomes Research Chair
Augusta, Georgia

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2003 SOGBD Young Investigator Research Grant Award

After a one-year hiatus, the AAP Section on Genetics and Birth Defects Young Investigator Research Grant Award has been reinstated. The competitive grant award provides \$20,000 to a promising young investigator performing basic or clinical research in the area of genetics and birth defects. The award is now generously supported by an educational grant from Genzyme Corporation.

For this year's award, the SOGBD Grant Subcommittee received nine excellent applications from young investigators across the country. Given the outstanding quality of the applications, the Grant Subcommittee had a difficult assignment in selecting the 2003 award winner.

The 2003 Young Investigator Research Grant awardee is Brett Harrison Graham, MD, PhD from the Department of Molecular and Human Genetics, Baylor College of Medicine, Houston, Texas. Dr. Graham will use the award to study "Drosophila VDACS: A potential genetic model for insights into mitochondrial function and disease." Thanks to all who submitted applications and congratulations to Dr. Graham! We look forward to hearing about his progress next year.

Katrina M. Dipple, MD, PhD
Chair, Subcommittee on Research Grants

SOGBD Educational Programs at AAP NCE

The SOGBD Executive Committee worked hard to develop 21 educational program proposals for the 2004 AAP National Conference and Exhibition. The 2004 NCE is scheduled from October 9 - 13, 2004 in San Francisco, California.

The NCE Planning Group, which is charged with reviewing and selecting all educational content for the annual NCE, accepted 55% of all submitted proposals this year. Eight specific topics submitted by the SOGBD were accepted and are as follows:

1. Behavioral Dysmorphology: How Should Syndromes Behave?
2. Genetic Unknowns
3. Genetic Arrays: Why this Profiling is Good
4. Is My Child at Risk for Cancer? Who, How, and When to Screen
5. Fetal Brain Development: Environment, Genes, and Everything You Always Wanted to Know But Were Afraid to Ask
6. Evaluation and Management of the Child with Mental Retardation
7. Familial Thrombophilia
8. Connective Issues: Diagnosis and Treatment of Marfan Syndrome and Other Connective Tissue Disorders

In addition, Fragile X and other genetics related topics will be presented. We are indebted to Gene Hoyme, MD (our liaison to the NCE Planning Group) for his hard work to ensure that we would have this number of topics and a strong representation of genetics issues on the NCE program. Many of the unaccepted proposals were deemed important but could not be accommodated in this cycle.

Proposals for the 2005 National Conference and Exhibition will be due in the spring of 2004. We welcome suggestions from the SOGBD members as we prepare for those submissions. Please send your suggestions to Dr. Robert Saul at rsaul@ggc.org.

Robert A. Saul, MD
Program Chair

From the Chair continued from page 1

This leaves many to question whether it is necessary to belong to both organizations. I strongly feel that membership in both organizations is important, worthwhile, and beneficial. Speaking for the AAP, membership brings with it a great deal of influence with the individuals in the trenches, the primary care pediatrician. We design the educational programs of the National Convention and Exposition. The SOGBD has designated a speaker's bureau so we can teach pediatricians in our local regions and communities. Through our liaison with the Committee on Genetics (COG) we determine and write the policy statements which educate and outline health management of several genetic disorders and birth defects. The policy statements have become the standard for the health maintenance of thousands of children with genetic disorders and pediatricians have depended upon them for much needed standard of care information. The future will necessitate updating old statements and creating new ones. In order to accomplish this, it is clear that the COG, in collaboration with the SOGBD, will have to look to you, the section members, for your expertise in order to meet this demand. By utilizing section members, we are assured that any information forwarded is appropriate and up to date.

The ACMG does have important functions, but the focus is much more targeted to genetic issues of training and research. There is no specific focus on the health of children or education of primary care physicians. By strengthening both the ACMG and the SOGBD, we have an amplified voice and the opportunity to reach more individuals. Working with the AAP, we have a much stronger voice in Washington, DC and our state capitols. Combining the strengths of the AAP and the ACMG we have a direct impact on geneticists and pediatricians. There is no similar committee in internal medicine, surgical specialties, or pathology. Obstetrics and gynecology has identified this need, but still does not have the structure seen in the AAP and SOGBD.

The SOGBD is an active and vital Section of the AAP. Its existence benefits all of us. We need to continue growing in order to preserve our presence in the AAP. I encourage you to maintain your membership and to persuade your eligible colleagues, fellows, and residents to join the AAP and the SOGBD. We also have an associate membership for genetic counselors who are interested in pediatric genetics issues. Although associate members cannot vote in Section elections, they are eligible to become members of sub-committees and participate in other SOGBD functions.

The past four years have been a wonderful experience for me. The new Chair of the SOGBD is Bob Saul and I am confident that he will offer strong leadership and new ideas to help us continue growing and evolving. I have only been able to succeed for the past four years because of your support. Bob will also need your support. Together we all can make a difference for the future of human genetics and the healthcare of children with genetic conditions and birth defects.

Howard M. Saal, MD
Chair, Section on Genetics and Birth Defects



SOGBD Strategic Planning Process

In order to ensure that the Section on Genetics and Birth Defects is meeting the needs of its members, the Executive Committee is currently reviewing and refining the Section's Strategic Plan. Phase one of this process, which involved surveying section members in the Spring of 2003 and compiling the results, has been completed. Thanks to all Section members who completed and returned their surveys! The survey identified the following five issues as priority concerns for the members of the section:

1. Advocate for adequate reimbursement for pediatric subspecialties.
2. Provide cost efficiency and decreased morbidity and mortality when care is provided by a pediatric geneticist.
3. Encourage integration of genetics training into all aspects of pediatric training, including the identification of genetic disorders and incorporation of genetics into rounds, clinics, etc.
4. Increase the amount of genetics curriculum included in regional and local AAP-sponsored CME courses.
5. Advocate for adequate reimbursement for case management of children with special health care needs.

During the coming months, the Executive Committee will be considering these issues to determine how and where Section resources can be most effectively utilized to meet the needs of the section.

Please note that in recent years, the Section Executive Committee has been actively addressing item number 1 ("advocate for adequate reimbursement") through Subcommittee on Health Systems Chair Dr. Marc Williams and in collaboration with the American College of Medical Genetics. With regard to item number 4 ("increase genetics curriculum included in local AAP CME courses"), the section has been very successful in increasing the genetics programming offered during the AAP National Conference and Exhibition, and the Executive Committee is already considering opportunities to carry this success over to regional and local AAP-sponsored CME offerings. Given the results of the Strategic Planning Survey, however, the Executive Committee will consider ways to enhance and strengthen their efforts on these issues, as well as the other priorities identified in the survey.

If you have any questions or comments on this process, or if you feel strongly about an issue that was not included among the top 5 identified priorities, please feel free to contact SOGBD Executive Committee member Dr. Mira Irons via email at mira.irons@tch.harvard.edu.

Mira Irons, MD
Strategic Planning Coordinator

Plan to attend...

The AAP Section on Genetics and Birth Defects Annual Business Meeting & Breakfast

from 7:00 am - 8:00 am on
Thursday, November 6, 2003!

The SOGBD Annual Business Meeting and Breakfast is scheduled in conjunction with the 2003 American Society of Human Genetics (ASHG) Annual Meeting in Los Angeles, California. The event will be held in the Tiffany room of the Millennium Biltmore Hotel.

This year's SOGBD Business Meeting and Breakfast will feature brief presentations by recent Young Investigator Research Award winners. Also, representatives from the SOGBD Executive Committee and the AAP Committee on Genetics will speak about section activities and other genetics-related initiatives at the Academy.

The fee to attend the Breakfast and Business Meeting is \$10. Please provide the information requested below and return this form, along with a check for \$10 made payable to "American Academy of Pediatrics," to: Barb Drelicharz, American Academy of Pediatrics, 141 Northwest Point Blvd., Elk Grove Village, IL, 60007-1098. **Registration is requested by Monday, October 27, 2003.**

If you have any questions, please do not hesitate to contact Paul Spire via email at pspire@aap.org or by phone at 847-434-7148.

Name: _____

Address: _____

City: _____

State: _____

Zip: _____

Phone: _____

Email: _____

SOGBD Executive Committee Election Results

The 2003 AAP Section Elections were recently completed. Thanks to all section members who voted! Please note the following results, which take affect in November 2003:

Robert A. Saul, MD has been elected Chairperson of the SOGBD Executive Committee. Dr. Saul is with Greenwood Genetics Center in Greenwood, South Carolina.

Emily Chen MD, PhD with Kaiser Permanente in San Jose, California and **Shawn E. McCandless** with University of North Carolina in Chapel Hill, North Carolina have been newly elected to serve as members of the Executive Committee.

Mira Irons, MD with Children's Hospital, Boston, Massachusetts has been re-elected to her position on the Executive Committee.

Executive Committee members Ronald Bachman, MD and Hope Northrup, MD have continuing terms and complete the six-member Executive Committee.

Relatedly, please also note that **Howard M. Saal, MD**, outgoing SOGBD Executive Committee Chairperson, was recently appointed to the AAP Committee on Genetics to serve as SOGBD Liaison to the committee.

Congratulations to all!

Leah W. Burke, MD
Chair, Nominations Subcommittee

Upcoming Events

2003 AAP National Conference and Exhibition

November 1 - 5, 2003
New Orleans, Louisiana

53rd Annual Meeting of The American Society of Human Genetics

November 4 - 8, 2003
Los Angeles, California

2003 SOGBD Annual Business Meeting & Breakfast

7:00 am - November 6, 2003
Tiffany Room, Millennium Biltmore Hotel
Los Angeles, California

2004 Annual Clinical Genetics Meeting

American College of Medical Genetics (in conjunction with the March of Dimes Symposium on Craniofacial Anomalies and the SIMD Symposium on New Diagnostic and Treatment Modalities for Inborn Errors of Metabolism)
March 4 - 7, 2004
Kissimmee, Florida

2004 AAP National Conference and Exhibition

October 9 - 13, 2004
San Francisco, California

AMERICAN ACADEMY OF PEDIATRICS



*National Conference & Exhibition
November 1-5*

Report from the AAP Committee on Genetics

The AAP Committee on Genetics (COG) held its fall meeting in Chicago on October 4-5, 2003. This was my first opportunity to act in the capacity of chairperson. This was also the first meeting attended by newly-appointed committee members Dr. Marilyn Bull and Dr. Nancy Mendelsohn. Both bring a wealth of clinical experience and scholarly activity to the committee. Additionally, Dr. Howard Saal attended for the first time in his new role as liaison from the Section on Genetics and Birth Defects (SOGBD). His past years' work as the Chair of the SOGBD provided a perfect backdrop for the committee's work plans this year.

An important part of the meeting's business was defining a strategy for expanding the scope of the work of the committee. It is clear that many of the work products (Policy Statements, Technical Reports, and Clinical Reports) are valued and well-utilized by pediatric healthcare providers. However, practical limitations exist as to man-power and available work hours from within the committee. It became obvious that an under-utilized resource is the membership of the section. Out of this meeting came a renewed sense of collaboration between the COG and SOGBD. We hope to expand the activities of the committee by soliciting consultation and partnership from within the section's membership. Look for upcoming requests for input and contributions from the section through Dr. Saal and his liaison activities.

Another highlight of the meeting was the opportunity for the COG to meet with Dr. Rodney Howell from the NIH. Dr. Howell attended with Dr. Jim Hanson to report on the new initiatives of the Center for Developmental Biology and Perinatal Medicine, National Institute of Child Health and Human Development (NICHD). Updates on new initiatives and strategies for improving and expanding Newborn Screening Services were emphasized. A major effort of the committee will be to keep the AAP membership aware of the rapid changes in practice and policy in this area.

Overall, the meeting was highly successful and productive. We look forward to a successful and productive year of projects with the section.

G. Bradley Schaefer, MD
Chair, AAP Committee on Genetics

Be Informed !! Get Involved !!

Join the Section on Genetics and Birth Defects ListServ Today!

The SOGBD ListServ allows your Section Executive Committee and AAP Staff to communicate with you in a timely and efficient manner. Currently, approximately 70% of the section membership belongs to the SOGBD ListServ. If you have not yet joined, or if your email address has recently changed, please provide the information requested below and fax this form back to the attention of Barbara Drelicharz at 847-434-8000. Alternatively, you may send an email to bdrelicharz@aap.org requesting to be added to the ListServ.

Thank You!

Name: _____

City: _____ State: _____

Email Address: _____

Previous Email Address to be Removed from ListServ (if applicable):

**Sign Up NOW!
for SOGBD
ListServ!**

FAX BACK THIS FORM TO!

Barbara Drelicharz - 847-434-8000

