
KaleidoScope

Volume 1 No. 1 Spring 2000

Publication of AAKSIS--the American Association of Klinefelter Syndrome Information and Support

In This Issue

- An Overview of Klinefelter Syndrome
- AAKSIS Couple First
- Membership Info
- Chicago Hope--the conference
- Early Identification of Speech Disorders
- Meet the organizers
- Regional Support
- Adult LD Resources

Into the new Millenium

Welcome to AAKSIS

The *American Association for Klinefelter Syndrome Information and Support*. AAKSIS (pronounced "access") is a new national, non-profit, volunteer organization dedicated to meeting the needs and challenges of the XXY/Klinefelter Syndrome community. We strive to become the clearinghouse for information, provide support and to respond to the needs of this community.

AAKSIS is a membership organization. As part of this association, you will receive the newsletter and discounted rates to our conferences. In addition, you can actively assist and participate in determining our direction and advocacy role.

Our first major undertaking will be the annual conference, "*Chicago Hope---Planning in the New Millenium,*" to be held in Chicago, Illinois from July 21st to 23rd. This national meeting will include workshops to inform as well as sessions devoted to member participation in planning for the growth and development of AAKSIS. For full details concerning the conference, please see page

If you are unable to join us in Chicago, please consider joining our organization at this time. (See page for membership form and information. Active involvement is important to AAKSIS. We welcome ideas, suggestions, and assistance. There is a place in this organization for all who want to fulfill the needs and interests of XXY and its variants. YOU are needed to make a difference.

AAKSIS
Toll-free HOT LINE
1-888-466-KSIS
(5747)

Klinefelter Syndrome

What to expect, how to respond?

by Wolfram Nolten, M.D.

Klinefelter Syndrome (KS) affects 1 in 500 male conceptions and is therefore the most common sex chromosome abnormality. It results in small testes, testosterone deficiency, infertility, and often (in) swelling of glandular breast tissue (gynecomastia). The first report in 9 patients, by H.F. Klinefelter Jr, E.C. Reifenstein, Jr., and F. Albright, in 1942, was followed by the discovery of an extra X chromosome as the cause of KS by P.A. Jacobs and J.A. Strong in 1959.

More than two thirds of KS patients have the 47, XXY karyotype. Few present with more than 2 X chromosomes or with imosaicism, which is having a normal XY and another abnormal XXY cell line. Testicular biopsy shows hyalinized and fibrotic seminiferous tubules, with few if any areas of sperm production. Seminiferous tubules normally comprise 85% of the testicular volume, and degenerative changes, such as fibrosis and hyalinization of the seminiferous tubules are the cause of testicular atrophy, which is invariably present in KS.

The chromosomal abnormality in KS is caused by an error in the division process in the production of gametes, where sperm or ovum contain an extra X chromosome, in addition to the normal X or Y chromosome.

The American Association for Klinefelter Syndrome Information and Support acknowledges the pioneering efforts of KS&A in spearheading a national forum for the Klinefelter Syndrome community. Its work brought together those now responsible for the creation of AAKSIS. AAKSIS expresses appreciation for what has come before and seeks to add its voice of service and support to the XXY cause. We dedicate our efforts to improving the quality of life for all affected by Klinefelter Syndrome.

Whereas a smear of the buccal mucosal membranes can show sex chromatin (Barr bodies), representing the extra X chromosome in KS men, a chromosome analysis, performed in peripheral blood cells will usually confirm the diagnosis of KS. In addition, the analysis will also define the number of X chromosomes, and provide information about the presence of mosaicism.

In the untreated adult KS patient, testosterone levels are usually low or below normal, with elevated LH. The diagnosis of KS is frequently made by prenatal genetic testing. In infancy the KS boy may present with a small penis, hypospadias, undescended testes. At school-age the child may show behavioral problems or learning disabilities. The adolescent may have tall stature, excessive growth of the lower extremities, delayed puberty, eunuchoidal features, and small testes, usually remaining at a volume of 2-3 ml. Testosterone production reaches a plateau after age 14 years and may never reach the midnormal adult range, in spite of high levels of LH stimulation. Adult KS patients have decreased bone mineral density, which could be caused by testosterone deficiency. Many adult KS men are first diagnosed during an infertility evaluation, when testicular atrophy and absence of sperm in the ejaculate are noticed.

KS patients have a high incidence of learning disabilities with decreased verbal IQ, but usually normal intellectual performance. Language skills can be impaired, patients may be aggressive, may not be inclined to participate in social activities, and may have a tendency to depression.

Possible Health Complications

KS can be complicated by

- mild adult-onset diabetes mellitus with insulin resistance
- hypothyroidism
- varicose veins of the lower extremities which can cause venous ulcers
- thinning of the surface of the teeth (taurodontism), which can cause early decay,
- breast cancer that may develop in middle age leukemia and Hodgkin and non-Hodgkin lymphoma *and in the young by*
- gonadal or extragonadal germinal cell tumors, frequently
- located in the mediastinum

Importance of Early Diagnosis

It is desirable to establish the diagnosis of KS as early as possible. This will direct the observation of parents to learning disabilities and behavioral abnormalities, which require remedial action. Attention should be paid to the possibility of complications, and screening tests should be ordered.

In adolescence, as soon as gonadotropin levels increase above normal, testosterone replacement should be initiated. Testosterone doses must be advanced over years with yearly increments, imitating the increase in testosterone production that occurs during normal puberty. Testosterone not only will advance virilization and support development of adequate bone mineralization, it also can improve fatigue, muscle strength, academic performance and further social adjustment.

In earlier years KS was thought to be invariably associated with antisocial behavior and mental retardation. This impression was prompted by institutionalized patients, whose findings served for earlier reports. Clinical experience with a wider segment of the population and decades of follow-up of KS men provide much more positive information. Early diagnosis, observation, and remedial action for emotional and behavioral abnormalities and timely treatment with testosterone can modulate the clinical course of KS

Editor's Note: Dr. Nolten has given us a broad overview of Klinefelter Syndrome. In the preceding article, he referred to medical, educational, and psychological problems that can be related to this disorder. In future issues of *Kalaedoscope*, we will examine those topics in depth. Dr. Nolten is a founding member of AAKSIS and is also on its Board of Directors



New Course Charted by AAKSIS Couple

Daniel and Wendy Becker wanted to start a family. But in 1994, after a year of failing to conceive a child, the couple was told that Daniel was sterile. Daniel discovered he had a genetic anomaly called Klinefelter Syndrome. What their doctors didn't count on was that this couple was not ready to accept this idea and so, they set out to disprove theory.

Together, Daniel and Wendy combed the Internet for information and support. Their unbridled courage helped convince a urologist to perform a testicular biopsy, seeking a

definitive diagnosis of sterility. Those results led to an inclusion in New York Hospital's IVF program. But even more significant, those results would forever change the notion that Klinefelter Syndrome equals absolute sterility. The real testimony of their determination came on February 12, 1997 with the birth of their child, a healthy XY boy. The Becker's son is the first child in the world to be born to an XXY male using Intra Cytoplasmic Sperm Injection (ICSI). AAKSIS is proud to have them as members of its Board.



Reading Tool Assists the Dyslexic

Recording for the Blind and Dyslexic (RFB&D) is an educational library for people with print disabilities such as dyslexia. Now in its 50th year, RFB&D provides recorded textbooks (in 4-track cassette or computer disk) and other classroom materials, from kindergarten through post-graduate levels. (This format differs from the typical books on tape found in bookstores or libraries.) Subjects included in this library are literature, history, math, science, reference and professional material. RFB&D has 80,000 titles which are available on loan. To participate in this service, an application process and a certification process must be completed to verify the learning disability. If you or someone you know could benefit from this service, contact RFB&D at 1-800-221-4792, or visit their website at www.rfbd.org for further information and fees.

Need Some “Inspiration”?

Organizing thoughts for written reports and essays can be challenging. The software, *Inspiration* assists the writing process by helping you arrange your ideas with visual organizers such as webs, concept maps, diagrams, and other prewriting aids. It easily converts the visual diagram into outline form to help create clear, concise writings. *Inspiration* also helps students comprehend concepts and information. The award-winning *Inspiration* is used in many schools. A free trial is available for downloading at www.inspiration.com.

Early Identification of Speech-Language Disorders

“Why is speech-language treatment needed? Everyone in our family was a late talker. My child will talk when the time is right.”

“Why worry now? Speech and language services will be available when school starts.”

“Treatment? Looks like they are just playing games to us.”

You may have had similar thoughts or comments. Usually, there is concern about a child's speech and language skills if there is no speech by the age of 1 year, if speech is not clear, or if speech or language is different from that of other children of the same age.

WHAT IS EARLY IDENTIFICATION?

It is estimated that 2% of all children born each year will have a disabling condition. Many of these children will have speech and/or language delays and disorders that may have a significant effect on personal, social, academic, or vocational life. Although some children will develop normal speech and language skills without treatment by the time they enter school, it is important to identify those who will not. Many people falsely believe that speech-language treatment cannot and should not begin until a child begins to talk. Yet, research has shown that children know a great deal about their language even before the first word is said. For example, children can distinguish between their native language and a foreign language, use different nonverbal utterances to express different needs, and imitatedifferent patterns of speech through babbling. Early identification includes the evaluation and treatment provided to families and their children under three years old who have, or are at risk for having, a disability or delay in speech, language, or hearing.

WHO SHOULD BE EVALUATED?

Children identified as at-risk or high-risk, such as those from neonatal intensive care units, should be tested early and at regular intervals. Other risk factors include diagnosed medical conditions, such as chronic ear infections; biological factors, such as fetal alcohol syndrome; genetic defects, such as Down Syndrome; neurological defects, such as cerebral palsy; or developmental disorders, such as delayed language. Children with no obvious high risk factors should be evaluated if their speech and language is not similar to that of other children of the same age. A child can quickly fall behind if speech and language learning is delayed. Early identification increases the chances for improving communication skills.

HOW IS THE EVALUATION DONE?

Evaluation may be formal or informal and include any combination of standardized tests; direct observation of play and interaction with caregivers; reports analysis of spontaneous speech samples. Several sessions as well as ongoing evaluation may be required to obtain enough information to make an accurate diagnosis. The early intervention team may consist of the speech-language pathologist, audiologist, psychologist, neurologist, electrophysiologist, otolaryngologist, pediatrician, nurse, and social worker. Because speech-language delays and disorders may be due to a variety of causes, each professional makes valuable contributions to the evaluation.

WHAT IS SPEECH LANGUAGE TREATMENT?

From the results of the evaluation, certain services may be recommended. Prevention includes those children who have been identified as at-risk for a communication delay or disorder because of low birthweight.

Services may be provided before a specific diagnosis has

been made. Remediation increases function in identified areas and may serve to prevent other related problems. For example, remediation of a language disorder can help offset learning disabilities.

Language is taught in a natural setting. It is presented at the child's developmental level. Responses are consistently stimulated. Output is rewarded. Play may be used to teach communication, language models or rules of conversation, such as taking turns.

If you are concerned about a possible speech-language delay or disability, consult a speech-language pathologist. A list of certified speech-language pathologists in your area may be obtained by calling the

American Speech-Language-Hearing Association's toll-free HELPLINE at 1-800-638-8255.

Compliments of American Speech-Language-Hearing Association and the Learning Disabilities Association

Websites for Adults with Learning Disabilities

- ✓ *Association of Higher Education and Disability* (AHEAD) --an international, multicultural organization of professionals committed to full participation in higher education for persons with disabilities. www.ahead.org (617-287-3880)
 - ✓ *Heath Resource Center*, the National Clearing House on Postsecondary Education for Individuals with Disabilities www.acenet.edu/About/programs/Access&Equity/HEATH (1-800-544-3284)
 - ✓ *Job Accommodation Network (JAN)* -- free consulting service. Information on: equipment, methods and modifications to improve work environment. Specific to the disability, including learning disabilities. www.janweb.icdi.wvu.edu (800-526-7324)
 - ✓ *National Adult Literacy and Learning Disabilities Center* -- promotes awareness about the relationship between adult literacy and learning disabilities <http://novel.nifl.gov/nalldtop> 800/953-ALLD (953-2553)
 - ✓ *President's Committee on Employment of People with Disabilities* www.pcepd.gov (202-376-6200)
 - ✓ *President's Task Force On Employment Of Adults With Disabilities* www.ptfead.gov
 - ✓ *National Council on Disability (NCD)*
www.ncd.gov (202-272-2004)
 - ✓ *National Rehabilitation Information Center (NARIC)* www.cais.net/naric
 - ✓ *National Association of Protection And Advocacy Programs/Client Assistance Programs (NAPAS/CAP)*
www.protectionandadvocacy.com
 - ✓ *Recording for the Blind and Dyslexic (RFB&D)* www.rfbd.org
 - ✓ *Rehabilitation Engineering And Assistive Technical Society of North America (RESNA)*
www.resna.org
 - ✓ **VOCATIONAL REHABILITATION AGENCIES OSERS**
www.ed.gov/offices/OSERS/RSA
-

Regional Support Contacts

SAN DIEGO AREA KLINEFELTER SYNDROME SUPPORT GROUP

Contact: Fred S. Diener
bear4u2c@aol.com
Klinefelter Syndrome San Diego
Area Support Group
Post Office Box 600112
San Diego, California 92160-0112

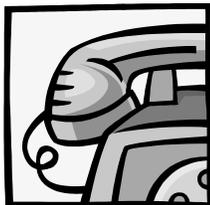
SOUTHEAST REGIONAL/ATLANTA AREA KLINEFELTER SYNDROME SUPPORT GROUP

Contact: Susan Johnson
(770) 983-7456
Smagee@worldnet.att.net
or Stefan Schwarz
sschwarz13@mediaone.net

PHILADELPHIA, PENNSYLVANIA

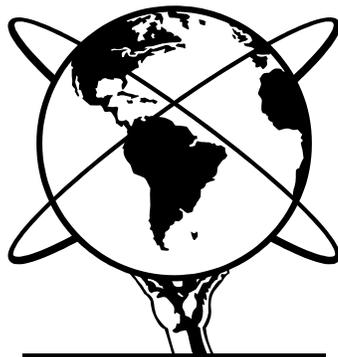
Contact: Christine Azzara
Krissie63@aol.com
(215) 334-6089

To list an event of interest to this organization,
contact the editor of KaleidoScope



Need information or
assistance?
Call the AAKSIS *HOT LINE*

1-877-4-AAKSIS
or
1-877-422-5747



www.aaksis.org

**First Annual Conference presented by
AAKSIS
Chicago Hope--Planning for our Future**

To be held at The Holiday Inn in Skokie, Illinois (Chicago Suburb)
5300 W. Touhy Avenue.

From 7:00 P.M. Friday, July 21, 2000
to 12:00 noon, on Sunday, July 23, 2000

WHO SHOULD ATTEND?

- individuals with XXY or any variation
 - families members
 - endocrinologists
 - genetic professionals
- psychologists, pediatricians
 - obstetricians
- primary care providers
 - nurses
- public health professionals
- and ANYONE interested in learning more about this condition.

WHAT TO EXPECT

Support sessions those interested in issues pertaining to

- children 0-10
- 11 through 18 y.o.
- young adults (19 y.o.) through adulthood
 - variation of 47,XXY
- sibling issues of XXY individuals
- wives and significant others 8. For "fathers only"
- couples dealing with reproductive and (in)fertility issues

Informative Sessions on

- Klinefelter Syndrome and
 - The importance of testosterone replacement therapy
 - medical complications related to Klinefelter Syndrome
- "Ask the Doctor" (exclusively for XXY and variants thereof individuals)
 - "Cutting Edge" methods to achieve reproduction, i.e., ICSI
 - Alternative types of reproductive techniques
 - Educational Issues
- "How To" session for successful lobbying of your legislature

And much more



Guest Speaker

J. Giedd, M.D.

NIH Researcher investigating the effects of XXY on the brain.

HOTEL INFORMATION

AAKSIS has reserved a block of rooms at a special rate of

\$89.00 + 9 % room tax = \$97.01 per night.

As many as 4 adults may stay in one room at this special rate.

If needed, rollaway beds are \$10.00 each per night + tax.

To receive this rate, you must

- call the Holiday Inn directly at **1 - 8 4 7 - 6 7 9 - 8 9 0 0**
- ask for the Sales Department
- identify yourself as an attendee to the AAKSIS National Conference.
- The above special rate is in effect until July 1, 2000. The reservation **MUST** be guaranteed with a credit card. After July 1, the special room rate will no longer be available to AAKSIS conference attendees.
(The usual room charge of \$159.00 + 9% tax, based on availability.)
- Check-in time is 3:00 P.M. Check-out is 12:00 noon.

CANCELLATION POLICY IS 72 HOURS PRIOR TO THE INDIVIDUAL'S ARRIVAL.

This Holiday Inn offers a Holidome Activity Center which includes an indoor swimming pool, indoor whirlpool, exercise facilities, sauna, E-Space including electronic entertainment/video games, badminton court, shuffle board court, pool table, and ping pong table.

There is also a Pippindale's Restaurant and Lounge in the Hotel.

Area Airport Information

O'Hare International (ORD)

Distance: 10 MI NE

Est. Taxi Fee: \$28.00

Alt. Transportation: Airport Express

Midway Airport (MDW)

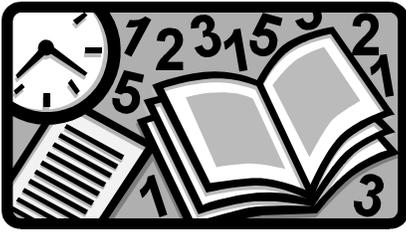
Distance: 25 MI N

Est. Taxi Fee: \$45.00

Alt. Transportation: none



MEET THE ORGANIZERS OF AAKSIS



Roberta Rappaport has a B.A. degree in elementary education from Roosevelt University, Chicago, IL. and her husband organized the first *Chicago/Midwest States Support Group* and Information Meetings in 1995. These meetings are now held twice a year. Roberta is the mother of Barbara, 29 and Michael, 32 who was diagnosed 47, XXY at 12. Presently, she works "full-time" as a volunteer, offering support and information to individuals and families with this condition.

Daniel Becker is an attorney in New York City. He is active in Reproductive & Genetic Rights, Donor Egg & Sperm Issues and Insurance Law. He is the former Interactive Service Director and Male Factor Board Moderator for *INCIID*. He is a former Board Member of *Resolve NYC* and a current Board Member of the *American Infertility Association*.

Wolfram E. Nolten, M.D., FACP, was born and raised in Germany and graduated from the University of Munich Medical School. He completed his medical residency at Ohio State University and his Fellowship in Endocrinology, Diabetes and Metabolism at the University of Cincinnati Medical School. Currently, Dr. Nolten is Associate Professor of Medicine, Division of Endocrinology, Diabetes and Metabolism at the University of Wisconsin, in Madison.

His special interests are: Andrology, including congenital sex chromosome abnormalities, hypogonadism, male infertility, erectile dysfunction. Dr. Nolten directs the Andrology Clinic at

the University of Wisconsin Hospital. His professional memberships include: the American Society for Reproductive Medicine, Andrology Society and the American College of Physicians. Dr. Nolten has presented repeatedly at Midwest Regional (XXY/Klinefelter Syndrome) Support Group Meetings and National Meetings of KS&A.

Penny Schwarz is a former teacher of music and Spanish. She graduated from Chatham College, 1962 with a B.A. and received her M.A. in Romance Languages from The Johns Hopkins University. She has been the Director of Miss Baltimore's Best Pageant, "Baltimore's Best Program," and is the Founder and Director of Penny Promotions Public Relations. Active in the cultural arts community of Baltimore, Penny is the mother of Stefan, diagnosed 46xy/47xxy in 1995.

Susan Johnson, currently a principal of an evening high school for nontraditional students, credits being a mother of an XXY son with opening up new perspectives and dimensions which enrich her life everyday. She and her son hosted the first support group meeting in the southeast and have worked together to educate professionals who serve the XXY community.

Richard Rappaport is an attorney practicing law in Chicago, IL. He received his B.S. degree and his J.D. degree from Loyola University, Chicago. Richard, husband of Roberta, and the father of Michael, and Barbara believes that early diagnosis of the XXY condition (or one of its variants) is essential. Richard and his wife, Roberta, are the organizers of the *Chicago/Midwest States XXY/Klinefelter Syndrome Support Group* and Information Meetings.

Michael J. Schwarz has been a practicing attorney in Baltimore, Maryland for 40 years. He received a B.A. from Johns Hopkins University in 1957 and is a graduate of the University of Maryland School of Law. In addition,

he is also a C.P.A. Michael is the father of Stefan who was diagnosed 46xy/47xxy at age 25.

Margaret Garvin, M.Ed. is a graduate of Kent State University and has done graduate work at Ohio State and Xavier University. A former teacher and library media specialist, she was more recently on staff at Ohio's special education Parent and Training Information Center. She has also served on the Central Ohio Board of the *Learning Disabilities Association*. In addition to being the parents of a 16 year old son diagnosed 47XXY at age one, she and her husband also have two older daughters. Margaret has been involved with the XXY community since 1995.

Wendy Becker is a graduate of the State University of New York at Albany with a B.A. in Psychology. Upon graduation, she moved through the ranks in publishing and in 1992 opened her own literary agency. Wendy continues to be active in issues respecting infertility and is a member of the XXYWives List.

Preston Garvin is a practicing attorney in Columbus, Ohio. He is a graduate of the University of Delaware and the University of Akron School of Law. His teenage son was diagnosed with 47XXY at age one.

Join us on the World Wide Web at <http://www.AAKSIS.org>

Our web master is

Vaughn Hambley who designed and created our website. Vaughn is 47XXY and lives in St. Catharines, Ontario, Canada.

Vaughn graduated from Brock University, St. Catharines, with a First Class Honors in English Lit, has an in-progress MA at Carleton, University, Ottawa, and a post-grad certificate in Technical Writing from Seneca College, Toronto.