Appendix: Patient Organizations in Connection with Small Supernumerary Marker Chromosomes

As underlined by the multiple personal case reports, sSMC patients and parents of sSMC carriers are shocked after receiving the aberrant cytogenetic result. Thus, in this situation they are in need of any kind of reliable specialist support they can get. Besides clinicians being well informed about the sSMC problem, patient support groups are a good source of help. In the following, rare chromosome disorder support groups, in general, and chromosome-specific support groups are listed. We apologize in advance for those not listed, as we did not miss them out them intentionally. Facebook (www.facebook.com) provides a useful source of informal contact and support for families. There is a rapidly growing number of pages on Facebook specific to particular chromosomes or disorders.

A1 Rare Chromosome Disorder Support Groups

A1.1 Unique: Rare Chromosome Disorder Support Group

Website: http://www.rarechromo.org
E-mail: info@rarechromo.org
Helpline: +44-1883-330766
Address: Unique, PO Box 2189, Caterham, Surrey CR3 5GN, UK

Unique offers support and information primarily in English but some information guides on individual rare chromosome disorders are available in languages such as German, French, Spanish, Italian, Danish, Greek, Romanian, Arabic and Dutch. Unique publishes a family-friendly guide to sSMC in English and German.

Having a child with a rare chromosome disorder can be a huge shock and stir up a range of emotions and a desire to learn more about the child's disorder. Almost everyone who works for Unique has been through these experiences. Some parents want to find another, older child with the same disorder as their child. Although this may be possible for some, it does not mean that the two children will develop in the

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same way. However, just talking to other parents with a child with a rare chromosome disorder can be a great relief and can help to dispel feelings of isolation and "Why me?"

Unique runs a helpline for families and professionals to find out more about specific rare chromosome disorders. It has developed an extensive computerized database detailing the effects of specific rare chromosome disorders among its members. The database can be used to link families on the basis of a specific rare chromosome disorder. Often of more practical benefit, however, is to link families on the basis of problems as they arise, whether these are medical, developmental, behavioral, social, or educational.

Unique also maintains close links with similar groups around the world, thus increasing the range of possible family contacts. Information about a specific rare chromosome disorder can be prepared from the Unique database without revealing the identity of the families concerned. This service is widely used by geneticists and genetic counselors worldwide.

Many local groups and contacts have been formed throughout the UK and in some other countries. Families affected by any rare chromosome disorder can get together locally for support and friendship and to pass on information about local services available. Unique publishes a regular magazine in which families can write about their experiences and exchange information. The group also holds study meetings and conferences in the UK where families and professionals can meet and discuss the latest developments. Unique can also act as a go-between to enable families to participate in any research projects relevant to their child's condition. Whatever a family's specific needs, Unique tries to provide them with tailor-made information and help relevant to their child's disorder.

A1.2 Contact a Family: for Families with Disabled Children

Website: http://www.cafamily.org.uk/rda-uk.html
E-mail: helpline@cafamily.org.uk
Telephone: 0808-8083555 (UK only)
Address: Contact a Family, 209–211 City Road, London EC1V 1JN, UK

Contact a Family provides support, advice, and information for families with disabled children; it is primarily active in the UK.

A1.3 LEONA: Verein für Eltern chromosomal geschädigter Kinder e.V.

Website: http://www.leona-ev.de
E-mail: info@leona-ev.de
Telephone: +49-4421-748669
Address: LEONA e.V. Kreihnbrink 31, 30900 Wedemark, Germany

LEONA is a group of rare chromosome disorder families with 400 members, more than 800 contact families, and representing over 300 syndromes. LEONA is active chiefly in German-speaking countries. Information and help are free of charge.

A1.4 Valentin APAC

Website: http://www.valentin-apac.org
E-mail: contact@valentin-apac.org
Helpline: +33-1-30379097
Address: Valentin APAC, 52, la Butte Eglantine, 95610 Eragny, France

Valentin APAC is the rare chromosome disorder support group for France and the French-speaking countries of Europe. It has more than 3,500 contact families.

A1.5 Unique Danmark

Website: http://www.uniquedanmark.dk
E-mail: formand@uniquedanmark.dk
Telephone: +45-3250-4155
Address: Unique Danmark, c/o Dorte V. Moeller, Blykobbervej 9 st.tv., 2770 Kastrup, Denmark

Unique Danmark is a rare chromosome disorder support group for families in Denmark.

A1.6 Chromosome Disorder Outreach

Website: http://www.chromodisorder.org
E-mail: info@chromodisorder.org
Telephone: +1-561-3954252
Address: Chromosome Disorder Outreach, Inc., PO Box 724, Boca Raton, FL 33429–0724, USA

Chromosome Disorder Outreach is a nonprofit organization providing support and information for families caring for a child, teen, or adult diagnosed with a rare chromosome disorder, including chromosome deletions, duplications, rings, inversions, and translocations. Chromosome Disorder Outreach operates primarily in English but has limited material available in Spanish, French, and Italian.

A1.7 Asociaţia Prader Willi din România

Website: http://www.apwromania.ro
E-mail: doricad@yahoo.com
Telephone: +40-260-616585
Address: Asociaţia Prader Willi din România, Str. Simion Bărnuţiu Nr. 97, Bl. SB88, ap. 14, Zalău, Sălaj, Romania

Asociaţia Prader Willi din România is a rare-disease-orientated organization specifically created for Romania and provides support in Romanian and English. Initially it was created by parents of a child with Prader Willi syndrome.

A1.8 Living with Trisomy

Website: http://www.livingwithtrisomy.org/
E-mail: fawna33@mindspring.com
Telephone: not applicable
Address: not applicable

This website in English provides links to family pages offering information and support to other families living with trisomy. Most of the linked family webpages will have trisomy support organization links of their own, but this site is specifically for families to connect with other families through their own webpages.

A1.9 National Organization for Rare Disorders

Website: http://www.rarediseases.org
E-mail: orphan@rarediseases.org
Telephone: 55 Kenosia Avenue, PO Box 1968, Danbury, CT 06813-1968, USA
Address: +1-203-744-0100

The National Organization for Rare Disorders [NORD] is a federation of voluntary health organizations dedicated to helping people with rare “orphan” conditions.

A2 Chromosome-Specific Support Groups

In the following, mainly different chromosome-specific support groups are listed.

A2.1 Chromosome 7

A2.1.1 Child Growth Foundation

Website: www.childgrowthfoundation.org
E-mail: info@childgrowthfoundation.org
Telephone: +44-20-8995-0257
Address: Child Growth Foundation, 2 Mayfield Avenue, Chiswick, London W4 1PW, UK

The Child Growth Foundation aims to support people with growth disorders, including Russell-Silver syndrome, to promote and fund relevant research, to educate both the public and professionals, and to encourage regular monitoring of growth and development criteria.

A2.1.2 Restricted Growth Association

Website: http://restrictedgrowth.co.uk
E-mail: office@restrictedgrowth.co.uk
Telephone: +44 (0)300-111-1970
Address: *see the website*

The Restricted Growth Association supports individuals and families affected by genetic restricted growth conditions, including Russell-Silver syndrome.

A2.1.3 Human Growth Foundation

Website: http://hgfound.org
E-mail: hgf1@hgfound.org
Telephone: +1-800-451-6434
Address: Human Growth Foundation, 997 Glen Cove Avenue, Suite 5, Glen Head, NY 11545, USA

The Human Growth Foundation aims to help people with growth and growth hormone disorders, including Russell-Silver syndrome, through research, education, support, and advocacy.

A2.1.4 The MAGIC Foundation

Website: http://www.magicfoundation.org
E-mail: dianne@magicfoundation.org
Telephone: +1-708-383-0808
Address: The MAGIC Foundation, 6645 W. North Avenue, Oak Park, IL 60302, USA

The MAGIC Foundation exists to provide support services for the families of children with a variety of growth conditions and disorders, including Russell-Silver syndrome.

A2.2 Chromosome 9

A2.2.1 9TIPS Trisomy 9 International Parent Support

Website: http://www.trisomy9.org/9tips.htm or http://health.groups.yahoo.com/group/9tips/
Email: bl737@bellsouth.net or 9tips@yahoogroups.com
Telephone: +1-909-8624470
Address: not applicable

9TIPS is an international support group for families dealing with all variations of duplications (trisomy) of chromosome 9.

A2.2.2 Trisomy 9

Website: http://www.trisomy9.org
E-mail: mandycain@bigpond.com
Telephone: not applicable
Address: not applicable

A2.3 Chromosome 11

A2.3.1 European Chromosome 11 Network

Website: http://www.chromosome11.eu
E-mail: see the website
Telephone: see the website
Address: see the website

The European Chromosome 11 Network is a support group for patients with chromosome 11 disorders, their families, and relatives.

A2.3.2 Beckwith-Wiedemann Syndrome Support Group

Website: http://www.mdjunction.com/beckwith-wiedemann-syndrome
E-mail: contact@mdjunction.com
Telephone: not applicable

Address: not applicable

An online community of patients, family members, and friends dedicated to dealing with BWS together.

A2.3.3 Beckwith-Wiedemann Support Group

Website: http://bws-support.org.uk
E-mail: r.baker881@btinternet.com; rbaker5165@aol.com
Helpline: +44-1258-817573 (evenings); +44-7889-211000 (cell phone)
Address: Beckwith-Wiedemann Support Group, The Drum and Monkey, Hazelbury Bryan, Sturminster Newton, Dorset DT10 2EE, UK

The UK Beckwith-Wiedemann Support Group aims to promote awareness of BWS, to support and encourage research, and to support affected families.

A2.3.4 L'Associazione Italiana Sindrome di Beckwith-Wiedemann

Website: http://www.aibws.org
E-mail: aibws@libero.it
Telephone: +39-345-3121850 (cell phone)
Address: L'Associazione Italiana Sindrome di Beckwith-Wiedemann, Piazza Turati, 3, 21029 Vergiate (VA), Italy

L'Associazione Italiana Sindrome di Beckwith-Wiedemann aims to increase knowledge about BWS and to contribute usefully to the care of patients and their families.

A2.4 Chromosome 12

A2.4.1 PKS Kids

Website: http://www.pkskids.com and http://www.pkskids.ning.com
E-mail: info@pkskids.net
Telephone: not applicable
Address: PO Box 94, Florissant, MO 60302-0094, USA

PKS Kids is a nonprofit organization and is where families and professionals can find the latest about what is happening in the world of PKS. For families and professionals the website is http://www.pkskids.com.

The PKS Kids social and support forum is at http://www.pkskids.ning.com. Families are invited to join the forum, offer help and friendship, and ask others for advice and answers. Sharing photos and videos of children makes it more fun!

A2.4.2 PKS Support Online

Website: http://groups.yahoo.com/group/pks_support
E-mail: not applicable
Telephone: not applicable
Address: not applicable

PKS Support Online is an international support group for sharing information, real-life experiences, and many other aspects of living with or caring for people diagnosed with PKS.

A2.4.2 PK Syndrome Online

Website: http://www.pk-syndrome.org
E-mail: andrea@pk-syndrome.org
Telephone: not applicable
Address: not applicable

This is a PKS lay support group, offering an interface with PKS-related information in English and Italian. It acts as an entry point to reach useful organizations, saving time and reducing frustration.

A2.5 Chromosome 14

A2.5.1 International Association Ring 14

Website: http://www.ring14.org
E-mail: info@ring14.it
Telephone: +39-0522-421037 (office) or +39-340-8681962 (cell phone)
Address: International Association Ring 14, Via Victor Hugo 34, 42123 Reggio Emilia, Italy

A2.6 Chromosome 15

A2.6.1 Dup15q Alliance

Website: http://www.dup15q.org
E-mail: info@dup15q.org
Telephone: +1-855-3871572
Address: Dup15q Alliance, PO Box 674, Fayetteville, NY 13066, USA

Dup15q Alliance, provides family support and promotes awareness, research, and targeted treatments for chromosome 15q duplication syndrome

A2.6.2 Nonsolo15

Website: http://www.idic15.it
E-mail: info@idic15.it
Telephone: +39-0575-583950
Address: Associazione Nonsolo15, Il Castello 15, Papiano, 52017 Stia (AR), Italy

Nonsolo15 operates in Italian and English.

Nonsolo15 is a nonprofit association providing support for families and friends of people affected by isodicentric chromosome 15 syndrome and supporting relevant research. The organization gives advice, information, and support to families with children with isodicentric chromosome 15 syndrome and to their friends; it helps them to meet each other to share experiences and receive support; it maintains an updated website, translating international information about research and therapies and publishing information material; it promotes research and maintains contact with researchers and physicians; it publicizes the experiences of affected families to the Italian community; and it grows every day with families' own children in pain and joy, and helps them to understand and grow too.

A2.6.3 idic15-EU

Website: http://health.groups.yahoo.com/group/idic15eu
E-mail: not applicable
Telephone: not applicable
Address: not applicable

This is a group for European families with a family member with isodicentric chromosome 15 syndrome, using any European language.

A2.6.4 Prader–Willi Syndrome

There are country-specific support groups and associations for PWS in many countries. In the first instance, contact the International Prader-Willi Syndrome Organisation (see below) or look at the links on the New Zealand Association website.

A2.6.5 International Prader-Willi Syndrome Organisation

Website: http://www.ipwso.org
E-mail: ceo@ipwso.org or g.fornaz@alice.it
Telephone: +39-0444-555557

Address: C/-B.I.R.D. Europe Foundation Onlus, via Bartolomeo Bizio, 36023 Costozza (VI), Italy

A2.6.6 Prader Willi Syndrom Vereinigung Deutschland e.V.

Website: http://www.prader-willi.de
E-mail: info@prader-willi.de
Telephone: +49-5141-3747327
Address: Prader Willi Syndrom Vereinigung Deutschland e.V., Am Bruückhorst 2a, 192 29227 Celle, Germany

Prader Willi Syndrom Vereinigung Deutschland e.V. was started by parents of children with PWS and is active chiefly in German-speaking countries.

A2.6.7 Prader-Willi Syndrome Association (UK)

Website: http://pwsa.co.uk
E-mail: admin@pwsa.co.uk
Telephone: +44-1332-365676
Address: Prader-Willi Syndrome Association (UK), 125a London Road, Derby DE1 2QQ, UK

A2.6.8 Prader-Willi Association (USA)

Website: http://www.pwsausa.org
E-mail: national@pwsausa.org
Telephone: 800-9264797 (USA only) or +1-941-3120400
Address: Prader-Willi Association (USA), 8588 Potter Park Drive, Suite 500, Sarasota, FL 34238, USA

A2.6.9 Prader-Willi Syndrome Association (NZ)

Website: http://www.pws.org.nz
E-mail: ceo@pws.org.nz
Telephone: 0800-479743 (toll-free in New Zealand)
Address: Prader-Willi Syndrome Association (NZ), PO Box 258, Silverdale, Auckland 0944, New Zealand

A2.6.10 Angelman Syndrome

There are many country-specific support groups and associations for AS. In the first instance, contact the International Angelman Syndrome Organisation (see below) or look at the New Zealand Association website.

A2.6.11 The International Angelman Syndrome Organisation

Website: http://www.international.angelmansyndrome.org
E-mail: president@angelmansyndrome.org
Telephone: not applicable
Address: not applicable

A2.6.12 ASSERT Angelman Syndrome Support Education and Research Trust

Website: http://www.angelmanuk.org
E-mail: angelmanuk@live.co.uk
Telephone: +44-300-9990102
Address: ASSERT, PO Box 4962, Nuneaton CV11 9FD, UK

A2.6.13 Angelman Syndrome Foundation

Website: http://www.angelman.org
E-mail: info@angelman.org
Telephone: +1-630-9784245
Address: Angelman Syndrome Foundation, 4255 Westbrook Drive, Suite 219, Aurora, IL 60504, USA

A2.6.14 Angelman Syndrome Association of Australia (Inc)

Website: http://www.angelmansyndrome.org
E-mail: president@angelmansyndrome.org
Telephone: not applicable
Address: Angelman Syndrome Association, PO Box 554, Sutherland, NSW 1499, Australia

A2.6.15 Angelman New Zealand

Website: http://www.angelman.co.nz
E-mail: angelman.info@nzord.org.nz
Telephone: +64-7824-7376
Address: Angelman New Zealand, PO Box 128, Ngaruawahia, New Zealand

A2.7 Chromosome 16

A2.7.1 Disorders of Chromosome 16 Foundation

Website: http://www.trisomy16.org
E-mail: doc16foundation@yahoo.com
Telephone: +1-760-6448867
Address: DOC16 Foundation, PO Box 230448, Encinitas, CA 92023-0448, USA

Disorders of Chromosome 16 Foundation is dedicated both to promoting research and providing information on chromosome 16 abnormalities.

The foundation provides information, education, and support for families of children living with a chromosome 16 disorder and to expectant parents confronting a similar diagnosis. The foundation also serves as a resource aiding family, friends, caregivers, and medical professionals in their supportive roles.

A2.8 Chromosome 17

A2.8.1 familyofchromosome17disorders

Website: http://health.groups.yahoo.com/group/familyofchromosome17disorders
E-mail: familyofchromosome17disorders@yahoogroups.com
Telephone: not applicable
Address: not applicable

This is a group started by the parent of a child with a microdeletion syndrome but which is for parents of a child with any type of rare disorder affecting chromosome 17

A2.8.2 Dup-17p11-2, 17p11.2 Duplication

Website: http://health.groups.yahoo.com/group/Dup-17p11-2
E-mail: dup-17p11-2@yahoogroups.com
Telephone: not applicable
Address: not applicable

The 17p11.2 Duplication Yahoo Group is designed to promote communication and the exchange of information about this chromosome disorder among parents, professionals, and researchers.

A2.8.3 Chromo 17 Europe

Website: http://www.chromo17europe.webs.com
E-mail: via the webpage
Telephone: not applicable
Address: not applicable

This is a support group and information site and forum for parents, carers, grandparents, and friends of children with conditions within the 17th chromosome.

A2.9 Chromosome 18

A2.9.1 Chromosome 18 Registry and Research Society

Website: http://www.chromosome18.org
E-mail: office@chromosome18.org
Telephone: +1-210-6574968
Address: Chromosome 18 Registry and Research Society, 7155 Oakridge Drive, San Antonio, TX 78229, USA

The Chromosome 18 Registry and Research Society is a lay advocacy organization composed primarily of the parents of individuals with one of the chromosome 18 abnormalities. Its mission is to help individuals with chromosome 18 abnormalities to overcome the obstacles they face so they may lead happy, healthy, and productive lives. The society provides specific information on tetrasomy 18p – see http://www.chromosome18.org/TheConditions/Tetrasomy18p/tabid/129/Default.aspx.

A2.9.2 Chromosome 18 Registry and Research Society (Europe)

Website: http://www.chromosome18eur.org
E-mail: secretary@chromosome18eur.org
Telephone: +44-1236-823455
Address: Bonnie McKerracher, 14 Main Street, Twechar, East Dunbartonshire G65 9TA, United Kingdom

Chromosome 18 Registry and Research Society (Europe) is a charity set up to support families whose children are affected by all chromosome 18 disorders. It provides information and support through its website, and parents who join the registry have access to a daily listserv, where they can ask questions and receive advice and encouragement from other families facing the same challenges every day. The charity aims to hold biannual family conferences. It is closely affiliated to the Chromosome 18 Registry and Research Society in the USA, the leading research body into the disorders.

A2.9.3 Tetrasomy18p.ca

Website: http://www.tetrasomy18p.ca
E-mail: tetrasomy18p@yahoo.ca
Telephone: not applicable
Address: not applicable

This is an information-sharing website, set up by a Canadian family.

A2.9.4 Tetrasomy 18p

Website: http://health.groups.yahoo.com/group/tetrasomy18p/
E-mail: tetrasomy18p@yahoogroups.com
Telephone: not applicable
Address: not applicable

This is a private community for those who have loved ones with this rare genetic syndrome. Tetrasomy 18p is a syndrome based on chromosome 18.

A2.10 Chromosome 20

A2.10.1 Ring Chromosome 20 Foundation

Website: http://www.ring20.org
E-mail: info@ring20.org

Telephone: +44-1708-403620 (UK) or +1-212-8602552 (USA)
Address: Ring Chromosome 20 Foundation, 62 Ravel Gardens, Aveley, Essex RM15 4NH, UK or Ring Chromosome 20 Foundation, 1045 Park Avenue, New York, NY 10028, USA

A2.11 Chromosome 21

A2.11.1 International Mosaic Down Syndrome Association

Website: http://www.imdsa.org/
E-mail: brandy@imdsa.org
Telephone: +1-513-9886817 (Skype calls from USA only)
Address: International Mosaic Down Syndrome Association, PO Box 354, Trenton, OH 45067, USA

International Mosaic Down Syndrome Association is designed to assist any family or individual whose life has been affected by mosaic Down syndrome, assist in research, and provide support without regard to race, sex, or religion.

A2.12 Chromosome 22

A2.12.1 Emanuelsyndrome.org or Chromosome 22 Central

Website: http://www.emanuelsyndrome.org or http://www.c22c.org
E-mail: info@emanuelsyndrome.org or steph.stpierre@gmail.com or murney.rinholm@c22c.org
Telephone: +1-705-2683099 or +1-919-5678167
Address: Chromosome 22 Central Inc, c/o Stephanie St-Pierre, 338 Spruce Street North, Timmins, ON P4N 6N5, Canada or c/o Murney Rinholm, 7108 Partinwood Drive, Fuquay-Varina, NC 27526, USA

Emanuelsyndrome.org is part of the larger parent group Chromosome 22 Central (http://www.c22c.org). Chromosome 22 Central provides families with information and offers opportunities for them to network with others through the Internet and at family gatherings. It provides information in English and has a contact for inquiries in Spanish.

A2.12.2 Ring 22

Website: http://health.groups.yahoo.com/group/ring22/
E-mail: ring22@yahoogroups.com
Telephone: not applicable
Address: not applicable

This is a resource for families and friends of people with ring chromosome 22.

A2.13 X and Y Chromosomes

There are many country-specific support groups and associations for TS. In the first instance, log on to the Turner Syndrome Society of the United States website (see below) and follow the links for International Contacts.

A2.13.1 Turner Syndrome Society of the United States

Website: http://www.turnersyndrome.org
E-mail: tssus@turnersyndrome.org
Telephone: 800-3659944 (toll-free in the USA)
Address: Turner Syndrome Society of the United States, 11250 West Road #G, Houston, TX 77065, USA

A2.13.2 Turner Syndrome Support Society (UK)

Website: http://www.tss.org.uk/
E-mail: turner.syndrome@tss.org.uk
Helpline: + 44-845-2307520
Address: Turner Syndrome Support Society (UK), 13 Simpson Court, 11 South Ave, Clydebank Business Park, Clydebank G81 2NR, UK

The society provides information on TS and the many aspects of living with the condition on a daily basis.

Glossary

This book is about a very complicated topic that can sometimes be hard for the layman to understand. Here an attempt is made to explain some of the most important technical and medical terms related to genes and chromosomes.

Chromosomes and Nomenclature

The human body is made up of billions of cells. Most of the cells contain a complete set of approx. 21,000 genes which act like a set of instructions, controlling growth and development and how the body works. Genes are carried on microscopically small, threadlike structures called chromosomes. There are usually 46 chromosomes, 23 inherited from the mother and 23 inherited from the father. Apart from two sex chromosomes (two X chromosomes for a girl and an X and a Y chromosome for a boy), there are 22 pairs of chromosomes that are numbered from 1 to 22, generally from largest to smallest.

The cytogenetic description of the chromosome set a person carries is expressed by a karyotype. This shorthand code usually states the total number of chromosomes, e.g., 46, followed by the sex chromosomes, e.g., XX or XY, followed by the numbers of cells studied in square brackets, e.g., [15]. A karyotype for a healthy female should normally read as 46,XX[15], and that of a healthy male should read as 46,XY[15]. A girl with Down syndrome describes in most cases the presence of an additional chromosome 21, so the karyotype is written as 47,XX,+21 [15]. If an sSMC is present, the karyotype of a boy reads as 47,XY,+mar[15]. The better an sSMC is characterized, the more complex the karyotype formula becomes. If the sSMC has a ring shape, is derived from chromosome 1, including bands 1p12 to 1q12, and is present in mosaic in a girl, the karyotype would be, e.g., 47,XX,+r(1)(::p12->q12::)[15]/46,XX[35]. The formula looks complicated and can be much more complicated, spanning several lines in a report, if all molecular cytogenetic probes applied are listed. In general, the length of the formula cannot be aligned with the severity of the clinical phenotype to be expected! The advantage of using

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karyotypes is that they are understood in every country by cytogenetic specialists, irrespective of the language a report is written in. The principles of the nomenclature are summarized in the so-called International System for Human Cytogenetic Nomenclature – ISCN (2009).

Alphabetic List of Terms

- ***Acrocentric*(*s*)** are chromosomes without a short p-arm, i.e., chromosomes 13, 14, 15, 21, and 22; as they have similar shapes with the centromere being near one chromosomal end, they are distinguished from all other, so-called nonacrocentric chromosomes.
- ***Alleles*** are one of two or more forms of a particular gene; alleles differ by their DNA sequences.
- ***Alpha fetoprotein*** is a major plasma protein produced by the developing baby. Some of this protein passes across the placenta and can be detected in the mother's blood during pregnancy. Testing alpha fetoprotein (AFP) values is a routine test for pregnancy surveillance.
- ***Alphoid or satellite DNA*** is a repetitive sequence stretch primarily located in the centromeric region of chromosomes. In humans there are specific sequences at almost each centromere.
- ***Amniocentesis*** is used in prenatal diagnosis of chromosomal abnormalities and fetal infections. Around 10 ml of amniotic fluid containing fetal cells is acquired by needle aspiration under ultrasound control.
- ***Anaphase*** is one of the five stages of mitosis.
- ***Aneusomy*** means any numerical deviation from a normal diploid karyotype; it may be a gain or a loss of a chromosome.
- ***Array-based comparative genomic hybridization*** (aCGH) is a DNA-directed array technique. Array-based methods are gradually replacing microscopy as the preferred approach to chromosome analysis (karyotyping) for children with developmental disorders as they have a higher diagnostic yield.
- ***Array techniques*** are recently invented methods which allow a high-resolution analysis of human DNA, RNA, and proteins.
- ***Ataxia*** is a clinical sign implying dysfunction of the parts of the nervous system that coordinate movement, such as the cerebellum. It consists of gross lack of coordination of movement.
- ***Atresia*** means that a passage in the body is congenitally closed or absent e.g., "gut atresia" means that there is a developmental blockage in the gut.
- ***Autosomes*** are all human chromosomes from chromosome 1 to chromosome 22 (i.e., all the chromosomes except the X and Y chromosomes, which are known as the sex chromosomes).
- ***Azoospermia*** describes the fact that a male has no measurable level of sperm in his ejaculate.

- ***Centromere*** is the narrow part of the chromosome between the short and the long arms. The centromere is the attachment spot for the spindle apparatus during mitosis.
- ***Chorion*** is a membrane present only during pregnancy between the developing fetus and the mother. Chorionic villi may be biopsied as an alternative to amniocentesis to study the chromosomal makeup of the developing pregnancy in pregnancies undergoing prenatal diagnosis. Chorionic villus sampling is usually undertaken at approximately 11–12 weeks of pregnancy.
- ***Chromatids*** (two of them) form one chromosome; they are joined at the centromere.
- ***Clinodactyly*** describes a bend or curvature of a finger.
- ***Coarctation*** is an abnormal narrowing in a blood vessel.
- ***Coloboma*** is a gap in one of the structures of the eye, e.g., the iris.
- ***Cryptorchidism*** is the failure of descent of one testis or both testes into the scrotum.
- ***Cyanosis*** indictes a lack of oxygen, which may happen during birth or postnatally.
- ***Cytoband*** is a G bands by trypsin using Giemsa stain (GTG)-light or GTG-dark chromosomal subband.
- ***Cytogenetics*** is a synonym for chromosome analysis.
- ***Cytomegalovirus*** belongs to the herpesvirus group.
- ***Epicanthus*** (adjective: epicanthic) describes a skin fold of the upper eyelid in the inner corner of the eye.
- ***Epigenetics*** refers to any heritable influence (in the progeny of cells or of individuals) on chromosome or gene function that is not accompanied by a change in DNA sequence, e.g., X-chromosome inactivation, imprinting, centromere inactivation, and position effect variegation.
- ***Euchromatin*** is genetic material containing actively transcribed/translated genes.
- ***Gamete*** is a human cell that fuses with another gamete during fertilization, i.e., sperm and oocyte.
- ***Gametogenesis*** is the process by which gametes form, i.e., spermatogenesis and oogenesis.
- ***Genotype*** is constituted by the genetic information present in a cell or a person.
- ***Gonadoblastoma*** is a (benign) tumor derived from different germ cells.
- ***Gonosome*** is a nonautosomal chromosome, i.e., a sex chromosome; in humans this is X and Y chromosomes.
- ***Hemizygosity*** means that a chromosomal region in a diploid organism is only present in one copy.
- ***Hemorrhage*** is the medical term for bleeding.
- ***Heterochromatin*** is used in this book as genetic material without (active) genes.
- ***Heterodisomy*** describes a special type of uniparental disomy. In heterodisomy there are two different homologous chromosomes derived from one parent <−> isodisomy.
- ***Homologous*** chromosomes designate a pair of two identical chromosomes.

- ***Howell–Jolly*** bodies are small DNA-containing inclusions of erythrocytes (red blood cells) and are often present after splenectomy.
- ***Hypertelorism*** is an abnormally increased distance between two organs – the term is most commonly used to describe wide spacing of the eyes.
- ***Hypospadias*** is a birth defect in which the opening of the urethra is unusually positioned, e.g., on the shaft of the penis rather than at the tip.
- ***Hypothyroidism*** is a deficiency of thyroid hormone.
- ***Icterus*** means jaundice.
- ***Intrauterine*** means within the uterus.
- ***Isochromosome*** is a derivative chromosome consisting of two identical short arms or two identical long arms.
- ***Isodisomy*** describes a special type of uniparental disomy. In isodisomy there are two identical homologous chromosomes derived from one parent <−> heterodisomy.
- ***Karyograms*** are depictions of chromosomes which are sorted by size, centromere position, and banding pattern in a standard format.
- ***Karyotypes*** describe the number of chromosomes, and what they look like under a light microscope. There is special nomenclature for that description, which ends up in a karyotype formula understandable worldwide if correctly applied.
- ***Kinetochore*** is the protein structure on chromosomes where the spindle attaches during cell division.
- ***Meiosis*** (adjective: meiotic) is a special type of cell division necessary for sexual reproduction leading to the formation of gametes.
- ***Mesoderm*** is one of the three primary germ cell layers in the very early embryo.
- ***Microphthalmia*** is a developmental disorder of the eye and just means "small eye."
- ***Mitosis*** is the process by which a eukaryotic cell separates the chromosomes in its nucleus into two identical sets in two nuclei.
- ***Monosomy*** means that instead of two copies of a chromosomal region only one copy is present.
- ***Mosaic/mosaicism*** describes the presence of two cell populations with different genotypes in one individual who has developed from a single zygote.
- ***Neoplasia*** is another word for tumor.
- ***Nondisjunction*** is the failure of chromosome pairs to separate properly during cell division.
- ***Occiput*** is the anatomical term for the back portion of the head.
- ***Omphalocele*** is an abdominal wall defect leading to a smaller or larger bulge containing the intestines at the site of the umbilicus ("tummy button").
- ***Oocyte*** is the female gamete.
- ***Phenotype*** is any observable characteristic of an individual.
- ***Postzygotic*** is the time after the first cell of an individual formed, i.e., the zygote.
- ***Scoliosis*** means that a person's spine is curved from side to side.
- ***Sirenomelia*** is a very rare malformation in which the legs are fused together, giving the appearance of a mermaid's tail.
- ***Spermatocyte*** is a male gamete.

- ***Spermatogenesis*** is the process during which a male gamete differentiates to a sperm.
- ***Stenosis*** is an abnormal narrowing in a blood vessel or another tubular structure of the body.
- ***Strabismus*** is a condition in which the eyes are not properly aligned with each other.
- ***Tetrasomy*** means that instead of two copies of a chromosomal region four copies are present.
- ***Tracheostomy*** describes the procedure of making an incision in the trachea to enable a patient to breathe.
- ***Trimester*** is a time period during pregnancy; the time between fertilization and delivery is divided into three equal parts: the first, second, and third trimesters.
- ***Trisomy*** means that instead of two copies of a chromosomal region three copies are present.
- ***Trophectoderm*** is, like the chorion, a membrane present only during pregnancy between the developing fetus and the mother.
- ***Zygote*** is formed by fertilization of an oocyte by a sperm. It is the single cell from which a multicellular organism is formed.

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