Small Supernumerary Marker Chromosomes (sSMC)

Thomas Liehr

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A Guide for Human Geneticists and Clinicians

With contributions by Unique
(The Rare Chromosome Disorder Support Group)

Springer

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Foreword

When Unique started up in 1984 as the Trisomy 9p Support Group, there was virtually no information or support for families about any rare chromosome disorder. Today it is different for people diagnosed with a known syndrome, but for the majority, including most people with a small supernumerary marker chromosome, little has changed. With its ever-increasing membership – currently standing at more than 10,000 individuals in 80 different countries – Unique fills that gap.

Small Supernumerary Marker Chromosomes is a welcome collaboration between a leading scientist and a family support group to create an up-to-date picture of one type of rare chromosome disorder. Scientific and clinical reports are brought to life by families' descriptions of the consequences of having a child with a small extra chromosome. Eighteen Unique families tell you in words and photographs what having this rare chromosome disorder means. Most of the children's names have been changed in accordance with their parents' wishes.

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Preface

Since 1992 I have been working in the field of clinical cytogenetics. My diploma, i.e., a master's thesis, was about a special subgroup of patients with small supernumerary marker chromosomes (sSMC), the cat eye syndrome (Liehr et al. 1992). Since that time much progress has been achieved in the field of sSMC. Especially the sSMC homepage (Liehr 2011) with presently more than 4,000 single sSMC case reports together with the advance of technical possibilities for a comprehensive characterization of this special group of rearranged chromosomes enables today much better genotype–phenotype correlations than when I started to study sSMC.

Nonetheless, I recently met a family with the following story, providing evidence that lots of knowledge on sSMC that is nowadays available did not reach the public health system as it should. An sSMC was detected after amniocentesis in the fetus of a pregnant woman who was referred for cytogenetic analysis because of advanced maternal age; sonographic findings were normal. The gynecologist told the couple that the cytogenetic finding was connected with an adverse prognosis and that the developing child would be "100% disabled and mentally retarded." The parents thus terminated the pregnancy. Later, it turned out that the sSMC was not only parentally derived but also that the first healthy child of the couple also had the same sSMC. This book is intended to help avoid similar situations and to be informative to clinicians, cytogeneticists, and families.

Besides the present knowledge on sSMC, including the biological background, also clinically relevant information is included together with personal reports of families having a child affected with an sSMC. The latter was realized in close collaboration with Unique, the Rare Chromosome Disorder Support Group (http://www.rarechromo.org/), and by contributions provided by families in contact with the author.

Jena, October 2011 Thomas Liehr

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Contents

Abbreviations

AC	Accessory chromosome
aCGH	Array-based comparative genomic hybridization
ACH	Accessory chromosome
AS	Angelman syndrome
BWS	Beckwith–Wiedemann syndrome
caNC	Cancer-associated neochromosome
CBG	C bands by barium oxide using Giemsa stain
CES	Cat eye syndrome
CGH	Comparative genomic hybridization
CVS	Chorionic villus sampling
der	Derivative chromosome
ES	Emanuel syndrome
ESAC	Extra structurally abnormal chromosome
FISH	Fluorescence in situ hybridization
GTG	G bands by trypsin using Giemsa stain
hUPD	Heterodisomy
i18pS	Isochromosome 18p syndrome
inv dup	Inverted-duplication-shaped small supernumerary marker chromosome
ISCN	International System for Human Cytogenetic Nomenclature
iUPD	Isodisomy
LCR	Low copy repeat
Mb	Megabase
min	Centric minute-shaped small supernumerary marker chromosome
NMC	Neocentric marker chromosome
NOR	Nucleolus organizing region
OMIM	Online Mendelian Inheritance in Man
p	Short chromosome arm
PCR	Polymerase chain reaction
PKS	Pallister–Killian syndrome
PWS	Prader–Willi syndrome

q	Long chromosome arm
r	Ring chromosome
SAC	Small accessory chromosome
SBAC	Small bisatellited additional chromosome
SMRC	Supernumerary minute ring chromosome
SMC	Supernumerary marker chromosome(s)
SRC	Supernumerary ring chromosome
SRS	Silver–Russell syndrome
sSMC	Small supernumerary marker chromosome(s)
TND	Transient neonatal diabetes
TS	Turner syndrome
UBCA	Unbalanced chromosomal abnormality
UPD	Uniparental disomy