Chromosomal mosaicism in the placenta. Presence and consequences
Blom, G.H.

Citation for published version (APA):

General rights
It is not permitted to download or to forward/distribute the text or part of it without the consent of the author(s) and/or copyright holder(s), other than for strictly personal, individual use, unless the work is under an open content license (like Creative Commons).

Disclaimer/Complaints regulations
If you believe that digital publication of certain material infringes any of your rights or (privacy) interests, please let the Library know, stating your reasons. In case of a legitimate complaint, the Library will make the material inaccessible and/or remove it from the website. Please Ask the Library: http://uba.uva.nl/en/contact, or a letter to: Library of the University of Amsterdam, Secretariat, Singel 425, 1012 WP Amsterdam, The Netherlands. You will be contacted as soon as possible.
CONTENTS

Chapter 1  General introduction  1
1.1  Conventional cytogenetics  3
1.2  Fluorescence in situ hybridization  3
1.3  Prenatal diagnosis  5
1.3.1  Amniocentesis  5
1.3.2  Chorionic villus sampling  7
1.4  Development of the placenta during early pregnancy  8
1.4.1  Normal development  8
1.4.2  Abnormal morphology of the placenta in relation to an abnormal karyotype  11
1.5  Chorionic villus sampling and placental mosaicism  11
1.5.1  Confined placental mosaicism  11
1.5.2  Combining short-term culture and long-term culture  14
1.6  Uniparental disomy and genomic imprinting  15
1.7  Confined placental mosaicism, uniparental disomy and fetal growth  18
1.8  Non-invasive fetal cell diagnosis  18
1.9  Aim of the thesis  20
1.10  Outline of the thesis  20
1.11  References  21

Chapter 2  FISH studies of placentas suspected of chromosomal mosaicism  31
2.1  Molecular cytogenetic analysis of term placentae suspected of mosaicism using fluorescence in situ hybridization  33
2.2  Placenta studies after chromosome mosaicism at CVS: an informative (t)issue  43

Chapter 3  Generalized mosaicism: two case reports  49
3.1  Maternal uniparental disomy for chromosome 22 in a child with generalized mosaicism for trisomy 22  51
3.2  Trisomy 8 in chorionic villi – unpredictable results in follow-up  58