Cytoscan can be used to detect uniparental isodisomy

Cytoscanoptima suite is a device that includes arrays, reagents and easy to use dataanalysis software. It is used for a cost effective and streamlined analysis of theparental and product of conception (POC) samples. The traditional cytogenetictechnique such as karyotyping and FISH has certain limitations due to cellculture failure and usually lack the appropriate sensitivity. Cytoscan optimaon the other hand, offers whole genome coverage for accurate detection ofchromosomal abnormalities with increased probe coverage targeting 396 relevantregions for parental and miscarriage applications.

This high resolution method providesgenotyping information for the detection of copy neutral loss/absence ofheterozygosity (LOH/AOH), which can be used to detect uniparental isodisomy(UPD). Since 50% of miscarriages are caused due to chromosomalabnormalities. Thesoftware used here to analyze the results of the following test, is calledChromosome analysis suite (ChAS). Thesoftware is tailored according to Affymertix to fit the cytogenetic researchanalysis. Due to its high resolution, it becomes easier to distinguish between aberrationsand artifacts.

The observed advantages of the software are Quick streamlinedresults, Summarized version of chromosomal aberrations across the whole genome, Focused analysis at different levels of resolutions on specific regions ofsignificance, Can directly access the external databases such as NCBI, OMIM, Ensemble etc., Thefemale infertility samples are processed using Cytoscan Optima and analyzedusing ChAS software. Theother types of Cytoscan are as follows: Cytoscan 750k and Cytoscan HD. Cytoscan750K array method enables one to detect high resolution copy number

acrossgenome and provide allelic imbalance from SNPs. This technique makes use of750, 000 markers for copy number and about 200, 000 genotype-able SNP for thepurpose of providing high resolution copy number and LOH detection. LOH rightlystands for Loss of Heterozygosity.

It can be defined as the phenomenon where, at the chromosomal crossing event, it results in the loss of an entire gene. CytoscanHD is a highly sensitive and reliable detection method for about 25-50kb copynumber changes across the genome. The technique covers 2. 6 million copy numbermarkers. This helps detect SNPs at about 99% genotype accuracy.

ChAS is thesoftware used for analysis purpose.