

SWAN Repository Dataset Documentation

Study: App#038, "Ovarian Aging in Fragile X Carriers Compared with Non-Carriers", Pastore

Dataset: SWANRep038_FRAXdata

Cohort: SWAN, all sites with DNA (excludes site 16)

Assay Description

DNA was amplified by PCR using primers that flank the Fragile X (FRAX-A) repeated trinucleotide sequence (CGG)_n on the X chromosome at Xq27.3. The PCR product was electrophoresed through a capillary and detected by the 6FAM fluorescent label on one of the PCR primers. By comparing to a standard curve, fragments were sized in basepairs and this result was converted to number of CGG repeats for each allele by subtracting the size of the primers, dividing by 3, and adding a correction factor for the instrument.

Code Book

Sample #: Sequential number of the sample

ARCHID: Encrypted SWAN Subject ID

Allele 1 Size (bp): Size of allele 1 in basepairs

Allele 2 Size (bp): Size of allele 2 in basepairs

Allele 1 Calculated # of CGG repeats: Number of trinucleotide repeats calculated from bp size

Allele 2 Calculated # of CGG repeats: Number of trinucleotide repeats calculated from bp size

Notes:

In some cases a 3rd allele was noted and the information was recorded on the spreadsheet.

In some cases no DNA was present or amplifiable and the information was recorded on the spreadsheet.