**UW Medicine - Pathology**

400-11-01-02

Array Data Analysis Procedure

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| Adopted Date: 10/12/09Review Date: 07/2010Revision Date: 07/2010 |

PURPOSE

After scanned microarray data is processed and loaded in Genoglyphix, data analysis using Genoglyphix software allows for evaluating the information of regions of copy number gain or loss in the genome and determining the significance of the changes.

PROCEDURE

### Equipment

* + - 1. Genoglyphix software from Signature Genomic Laboratories

### Procedure

1. Choose a patient folder from the "to be analyzed" rack in the main office.
2. Log onto Genoglyphix ([www.genoglyphix.com](http://www.genoglyphix.com)) using the login and password provided by Signature Genomic Laboratories.
3. Click on **Subject List** to view a list of cases created. Alternatively, click on **Search for Cases** using subject ID (case number) or physician.
4. Click on the subject ID (case number) that you are going to analyze. Subject Summary will open. Check all the patient identifiers and slide lot number (Slide info in yellow) matching with the fax notice from Signature Genomic Laboratories.
5. Click on **Analysis Status** on the left side menu, the whole genome plot and each individual chromosome plot will show up from the oligo array uploaded previously. Check the standard deviation (SD), the value should be <0.2. Then check through the whole genome plot.
6. On the whole genome plot, take note of the end of the plot where the sex chromosomes are located. For male patients, there should be no significant symmetrical gain (pink dotes) and loss (blue dots). For female patient there should be a minor symmetrical gain and loss in the Y region. If sex does not seem to match or there is another pattern present, either a sample handling error or a sex chromosome abnormality should be considered.
7. Scroll down through the rest of the chromosome plots and take note of any telomeres that appear "chatter". If several chromosomes display telomere chatter, check if the oligo array needs to be repeated or if the DNA needs to be extracted again.
8. Check each individual chromosome plot to see if any region that should be flagged is missed.
9. Click on **Karyogram Review** from the tool bar to see if there is any detectable mosaicism apparent. Print the Karyogram from the browser.
10. Click on **Create Plots**, download "Include All Chart". A PDF file of the whole genome plots will be open, save it in the patient case folder located in the "Cytogenetics" folder under "Shared Files - CGH\_cases". Print the PDF file on color printer and staple printout and put patient label on it.
11. Click on the **Notation Review**. This page will show each flagged region of the gains and losses of 5 or more adjacent oligonucleotide probes of significant value determined by NimbleScan, DNA Analytics and Genoglyphix.
12. Go through each flagged region and determine the significance of the copy number changes by investigation these features and make notes for:
	1. Size of the gain and loss
	2. Location of segmental duplications and gaps in coverage
	3. Genes involved, number of OMIM genes, and number of other genes in the region
	4. Known benign copy number variants (CNVs)
	5. Previous UWMC cases (MyGCAD) and Signature Genomics (GCAD) case of similar size and location
	6. Known syndromes in the region (SGL GPS)
	7. Database of genomic variants listings (DGVs)
	8. FISH probes available in the region
13. When the analysis is complete, click on **Analysis Status**, fill in your initials, then click on the "case reviewed" box.
14. Go through the Array CGH Quality Assurance Checklist in the patient folder, initial and date all check marks under Primary Tech and make sure it's complete and accurate.
15. After related studies are done, click on the **Final Review & Report**, enter ISCN diagnosis for array result and FISH result, make sure all annotation for the gains and losses are complete and accurate, and click on save report.

REFERENCES

1. Shaffer LG, Beaudet AL, Brothman AR, Hirsch B, Levy B, Martin CL, Mascarello JT, Rao KW; Working Group of the Laboratory Quality Assurance Committee of the American College of Medical Genetics. 2007. Microarray analysis for constitutional cytogenetic abnormalities. Genet Med. 9(9):654-62.
2. <https://www.genoglyphix.com>
3. <http://www.signaturegenomics.com>

Written By: Director Approval:

(Signature and Date) (Signature and Date)

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**UW Medicine - Pathology**

 **Cytogenetics - UWMC**

**SIGNATURE PAGE FOR POLICIES AND PROCEDURES**

Procedure / Policy Title: Array Data Analysis Procedure

Procedure / Policy Number: 400-11-01-02

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