EGL Genetics | Eurofins Clinical Diagnostics

Lora J. H. Bean, PhD, FACMG, Director
2460 Mountain Industrial Boulevard
Tucker, GA 30084
Toll Free: (855) 831-7447
International: (470) 378-2200
FAX: (470) 378-2250
CLIA#: 11D0683478
CAP#: 7181693

Cytogenetics Laboratory

PATIENT INFORMATION — FINAL REPORT

PATIENT NAME: **Egl Genetics**
DATE OF BIRTH: **2/13/2017**
PATIENT SEX: **Female**
CROSS REFERENCE #: **123-456**
PATIENT ID: **T0302434**

LABORATORY #: **18VA05834**
TYPE OF SPECIMEN: **Whole Blood (EDTA & Sodium heparin)**
DATE COLLECTED: **6/19/2018**
DATE RECEIVED: **6/20/2018**
DATE INITIATED: **6/20/2018**
FINAL REPORT: **6/27/2018**

REFERRING DIAGNOSIS: **F84.0 - Autistic disorder**
REFERRING CLINICIAN/INSTITUTION: Irma Doctor 555-555-5555 (Fax)

Chromosomal Microarray: EmArray Cyto

CLINICAL INTERPRETATION AND SIGNIFICANCE

MICROARRAY RESULTS **NORMAL**

RECOMMENDATIONS

- These results should be interpreted in the context of this individual's clinical features.
- Genetic evaluation and counseling are warranted to discuss the implications of this result.
- For more information, please visit eglgenetics.com or call 855-831-7447 (toll free) to contact a laboratory genetic counselor or consult with a laboratory director.

TECHNICAL RESULTS AND METHODS

NOMENCLATURE arr[hg19] (1-22,X)x2

METHODS The EmArray Cyto is a custom oligonucleotide microarray designed to detect genomic gains and losses by combining targeted and genome-wide coverage. The array includes high-density coverage for clinically relevant deletion/duplication syndromes, subtelomeric, and pericentromeric regions. Genome-wide coverage has an average probe spacing of approximately 75 kb to detect gains and losses at a minimum of 400 kb. The array was designed using a commercial platform (Oxford Gene Technology, Inc.) that has approximately 60,000 oligonucleotide probes that represent coding and noncoding sequences in the human genome (content sourced from the UCSC hg19 human genome: GRCh build 37, February 2009). Patient DNA is cohybridized with same-sex control DNA pooled from five DNA samples. Analysis and interpretation are performed using data from the UCSC Genome Browser (http://genome.ucsc.edu), the Database of Genomic Variants (http://projects.tcag.ca/variation/), and an internal copy number database.

Disclaimer: This test will only detect gains or losses of DNA, not balanced rearrangements such as translocations or inversions. Also, this test is not intended to detect low-level mosaicism. Abnormalities in regions not represented on the array or DNA sequence changes (such as point mutations, small deletions, or insertions) will not be detected by this assay. FISH or G-banding may confirm abnormal CGH findings, either at the time of initial testing or upon receipt of parental samples, depending on the abnormality.

Benign copy number changes (bCNCs) present in >1% of the population are not reported, but are available upon request.

Pursuant to the requirements of CLIA '88, this test was developed and its performance validated by EGL Genetic Diagnostics LLC. It has not been cleared or approved by the U.S. Food and Drug Administration (FDA). This test is used for clinical purposes.

Electronically signed by Mariana Kekis, Ph.D., DABMG on 6/27/2018 at 12:57 PM

Michael Gambello, M.D., Ph.D., FACMG
Medical Consultant