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Supplemental Data

Consensus Statement: Chromosomal Microarray Is a First-Tier Clinical Diagnostic Test for Individuals with Developmental Disabilities or Congenital Anomalies

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General Recommendations of the ISCA Consortium

1. These recommendations are intended as a guide for clinical application of microarrays, not a laboratory guideline.
2. These recommendations apply only to genetic testing of individuals with unexplained developmental delay/intellectual disability (DD/ID), autism spectrum disorders (ASD), or multiple congenital anomalies (MCA). They are not intended as a guide for testing of prenatal or cancer samples.
3. Terminology of genomic copy number imbalance.
    a. Use the terms “copy number variant” (CNV) or “copy number change” (CNC) interchangeably to refer to any change in copy number, regardless of clinical implications.
    b. Use the term variant of uncertain clinical significance (VOUS) to denote copy number variants for which the clinical significance is not well established or is ambiguous.

4. As a clinical genetics community, we should develop and implement clinical laboratory standards for CMA.
    a. Recommendations should be neutral with regard to technology, platform and manufacturer.
    b. Minimum genomic effective resolution and genomic coverage should be comparable and available among all clinical laboratories.
5. To promote more uniform clinical care, we should enhance sharing of knowledge among clinical laboratories to improve the quality and consistency of interpretive information provided to clinicians and patients.
    a. Establish standards for classification of copy number variants as pathogenic (related to disease), uncertain clinical significance, or benign (not clinically significant),
    b. Develop databases for sharing information about clinical interpretation of copy number variants among clinical laboratories.
6. To improve the accuracy, reliability, and reproducibility of CMA, we should develop metrics for proficiency and link clinical service to evidence-based recommendations.
7. To ensure that clinical geneticists (physicians, genetic counselors, cytogeneticists, molecular geneticists, and trainees) have an adequate understanding of CMA, educational tools should be developed and utilized in American Board of Medical Genetics programs for trainees in clinical and laboratory genetics, and for maintenance of certification and recertification.