Resource Summary Report

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ISCA Consortium

RRID:SCR_006168
Type: Tool

Proper Citation

ISCA Consortium (RRID:SCR_006168)

Resource Information

URL: https://www.iscaconsortium.org/

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Description: THIS RESOURCE IS NO LONGER IN SERVICE. Documented on June 22, 2022. A rapidly growing group of clinical cytogenetics and molecular genetics laboratories committed to improving quality of patient care related to clinical genetic testing using new molecular cytogenetic technologies including array comparative genomic hybridization (aCGH) and quantitative SNP analysis by microarrays or bead chip technology. They improve clinical care by providing a large publicly available database and forum where clinicians and researchers can share knowledge to expedite the understanding of copy number variation (CNV) in an abnormal population. The ISCA database contains whole genome array data from a subset of the ISCA Consortium clinical diagnostic laboratories. Array analysis was carried out on individuals with phenotypes including intellectual disability, autism, and developmental delay. Efforts of the Consortium include: # Clinical Utility: The ISCA Consortium has made recommendations regarding the appropriate clinical indications for cytogenetic array testing (Miller et al. AJHG 2010, PMID: 20466091). Currently, discussions are focused on pediatric applications for children with unexplained developmental delay, intellectual disability, autism and other developmental disabilities. A separate committee has been developed to address appropriate cancer genetic applications (http://www.urmc.rochester.edu/ccmc/). # Evidence-based standards for cytogenomic array design: The Consortium will develop recommendations for standards for the design, resolution and content of cytogenomic arrays using an evidence-based process and an international panel of experts in clinical genetics, clinical laboratory genetics (cytogenetics and molecular genetics), genomics and bioinformatics. This design is intended to be platform and vendor-neutral (common denominator is genome sequence coordinates), and is a dynamic process with input from the broader genetics community and evidence-based review by the expert panel (which will evolve into a Standing Committee with international representation). # Public Database for clinical and research community: It is essential that

publicly available databases be created and maintained for cytogenetic array data generated in clinical testing laboratories. The ISCA data will be held in dbGaP and dbVar at NCBI/NIH and curated by a committee of clinical genetics laboratory experts. The very high quality of copy number data (i.e., deletions and duplications) coming from clinical laboratories combined with expert curation will produce an invaluable resource to the clinical and research communities. # Standards for interpretation of cytogenetic array results: Using the ISCA Database, along with other genomic and genetics databases, the Consortium will develop recommendations for the interpretation and reporting of pathogenic vs. benign copy number changes as well as imbalances of unknown clinical significance.

Abbreviations: ISCA Consortium, ISCA

Synonyms: ISCA Consortium and Public Database, International Standards for Cytogenomic Arrays (ISCA) Consortium, International Standards For Cytogenomic Arrays Consortium

Resource Type: community building portal, organization portal, database, data or information resource, consortium, portal

Keywords: clinical, cytogenetics, molecular genetics, genetic testing, molecular cytogenetic technology, array comparative genomic hybridization, quantitative snp analysis, microarray, bead chip, genome, array, phenotype, copy number, deletion, duplication, copy number variation, FASEB list

Related Condition: Intellectual disability, Developmental delay, Etc., Autism

Funding:

Availability: This resource is no longer in service

Resource Name: ISCA Consortium

Resource ID: SCR_006168

Alternate IDs: nlx_151670

Record Creation Time: 20220129T080234+0000

Record Last Update: 20250519T204320+0000

Ratings and Alerts

No rating or validation information has been found for ISCA Consortium.

No alerts have been found for ISCA Consortium.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 59 mentions in open access literature.

Listed below are recent publications. The full list is available at dkNET.

Zhao Y, et al. (2024) Prenatal diagnosis and postnatal follow-up of 15 fetuses with 16p13.11 microduplication syndrome. Frontiers in genetics, 15, 1486974.

Yi T, et al. (2023) Genetic aetiology distribution of 398 foetuses with congenital heart disease in the prenatal setting. ESC heart failure, 10(2), 917.

Que Y, et al. (2023) Ultrasonographic characteristics, genetic features, and maternal and fetal outcomes in fetuses with omphalocele in China: a single tertiary center study. BMC pregnancy and childbirth, 23(1), 679.

Cai M, et al. (2023) Pathogenic copy number variations are associated with foetal short femur length in a tertiary referral centre study. Journal of cellular and molecular medicine, 27(16), 2354.

Yue F, et al. (2023) Prenatal phenotypes and pregnancy outcomes of fetuses with recurrent 1q21.1 microdeletions and microduplications. Frontiers in medicine, 10, 1207891.

Cai M, et al. (2022) Novel homozygous nonsense mutation associated with Bardet-Biedl syndrome in fetuses with congenital renal malformation. Medicine, 101(32), e30003.

Kowalczyk K, et al. (2022) Application of array comparative genomic hybridization (aCGH) for identification of chromosomal aberrations in the recurrent pregnancy loss. Journal of assisted reproduction and genetics, 39(2), 357.

Liu Y, et al. (2022) Chromosomal microarray analysis of 410 Han Chinese patients with autism spectrum disorder or unexplained intellectual disability and developmental delay. NPJ genomic medicine, 7(1), 1.

Lu F, et al. (2022) Estimating the frequency of causal genetic variants in foetuses with congenital heart defects: a Chinese cohort study. Orphanet journal of rare diseases, 17(1), 2.

Yue F, et al. (2022) Prenatal detection of pure proximal 6q14.1 microduplication encompassing LCA5 gene: A variant of likely benign. Medicine, 101(24), e29369.

Li M, et al. (2022) Prenatal diagnosis and molecular cytogenetic characterization of an inherited microdeletion of chromosome 16p11.2. The Journal of international medical research, 50(7), 3000605221109400.

Huang H, et al. (2021) Chromosomal Microarray Analysis for the Prenatal Diagnosis in

Fetuses with Nasal Bone Hypoplasia: A Retrospective Cohort Study. Risk management and healthcare policy, 14, 1533.

Greenbaum L, et al. (2021) Chromosomal Microarray Analysis in Pregnancies With Corpus Callosum or Posterior Fossa Anomalies. Neurology. Genetics, 7(3), e585.

Cai M, et al. (2021) Clinical Utility and the Yield of Single Nucleotide Polymorphism Array in Prenatal Diagnosis of Fetal Central Nervous System Abnormalities. Frontiers in molecular biosciences, 8, 666115.

Huang H, et al. (2021) Effectiveness of Chromosomal Microarray Analysis for Prenatal Diagnosis of Fetal Echogenic Intracardiac Focus: A Single-Center Experience. International journal of general medicine, 14, 1991.

She Q, et al. (2021) Prenatal genetic testing in 19 fetuses with corpus callosum abnormality. Journal of clinical laboratory analysis, 35(11), e23971.

Yue F, et al. (2021) Prenatal detection of terminal 9p24.3 microduplication encompassing DOCK8 gene: A variant of likely benign. Medicine, 100(3), e23967.

Fan X, et al. (2021) Performance of Chromosomal Microarray Analysis for Detection of Copy Number Variations in Fetal Echogenic Bowel. Risk management and healthcare policy, 14, 1431.

Zhang X, et al. (2021) Prenatal detection and molecular cytogenetic characterization of 19q13.42 microduplication: three reported cases and literature review. Molecular cytogenetics, 14(1), 5.

Li L, et al. (2021) Ultrasonographic findings and prenatal diagnosis of complete trisomy 17p syndrome: A case report and review of the literature. Journal of clinical laboratory analysis, 35(1), e23582.