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


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

**Availability:** This resource is no longer in service

**Resource Name:** ISCA Consortium

**Resource ID:** SCR_006168

**Alternate IDs:** nlx_151670

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**Ratings and Alerts**

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

No alerts have been found for ISCA Consortium.

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**Data and Source Information**

**Source:** [SciCrunch Registry](https://www.sci.crunch.org)

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**Usage and Citation Metrics**
We found 55 mentions in open access literature.

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

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.


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.


Huang H, et al. (2021) SNP Array as a Tool for Prenatal Diagnosis of Congenital Heart Disease Screened by Echocardiography: Implications for Precision Assessment of Fetal Prognosis. Risk management and healthcare policy, 14, 345-355.


biosciences, 8, 666115.


Song Y, et al. (2021) Application of Copy Number Variation Detection to Fetal Diagnosis of Echogenic Intracardiac Focus During Pregnancy. Frontiers in genetics, 12, 626044.


Zhang H, et al. (2021) Prenatal detection of distal 1q21.1q21.2 microduplication with abnormal ultrasound findings: Two cases report and literature review. Medicine, 100(1), e24227.