

Resource Summary Report

Generated by SPARC SAWG on Aug 9, 2026

ISCA Consortium

RRID:SCR_006168

Type: Tool

Proper Citation

ISCA Consortium (RRID:SCR_006168)

Resource Information

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

Proper Citation: ISCA Consortium (RRID:SCR_006168)

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: data or information resource, community building portal, database, consortium, 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

Record Creation Time: 20220129T080234+0000

Record Last Update: 20260808T190234+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 78 mentions in open access literature.

Listed below are recent publications. The full list is available at [SPARC SAWG](#) .

Yin Y, et al. (2025) A Prospective Evaluation of the Diagnostic Utility for Low-Coverage Genome Sequencing in Prenatal Samples: A Comparison With Chromosomal Microarray Analysis. *Prenatal diagnosis*, 45(12), 1525.

Zhang Y, et al. (2025) Genetic and clinical features of microcephaly in a prenatal cohort. *BMC pregnancy and childbirth*, 26(1), 65.

Pan Y, et al. (2025) 16p13.11 microduplication in 14 fetuses: prenatal diagnosis and postnatal follow-up. *Molecular cytogenetics*, 18(1), 30.

Liu S, et al. (2025) 5' UTR variant in the NDP gene leads to incorrect splicing and familial exudative vitreoretinopathy. *Orphanet journal of rare diseases*, 20(1), 553.

Xie W, et al. (2025) Ultrasound and genetic findings in a case series of fetuses presenting vertebral defects. *BMC pregnancy and childbirth*, 25(1), 858.

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

Xu T, et al. (2024) Prenatal detection of distal 18p deletion by chromosomal microarray analysis: Three case reports and literature review. *Medicine*, 103(30), e39046.

Zhou Y, et al. (2023) Prenatal diagnosis of ultrasound soft markers in a single medical center of mainland China. *Molecular cytogenetics*, 16(1), 3.

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

Wang Y, et al. (2023) Should Prenatal Chromosomal Microarray Analysis Be Offered for Pulmonary Atresia? A Single-Center Retrospective Study in China. *Genes*, 14(3).

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.

Wang Y, et al. (2023) Genetic diagnosis of fetal microcephaly at a single tertiary center in China. *Frontiers in genetics*, 14, 1112153.

Cao L, et al. (2023) Advanced maternal age: copy number variations and pregnancy outcomes. *Frontiers in genetics*, 14, 1206855.

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. (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.

Sennsfelder L, et al. (2023) Description of Copy Number Variations in a Series of Children and Adolescents with FASD in Reunion Island. *Children (Basel, Switzerland)*, 10(4).

Yi T, et al. (2022) Genetic and Clinical Features of Heterotaxy in a Prenatal Cohort. *Frontiers in genetics*, 13, 818241.

Li D, et al. (2022) Case report: Fetal cervical immature teratoma and copy number variations. *Frontiers in oncology*, 12, 843268.

Wu CW, et al. (2022) Copy Number Variation Analysis Facilitates Identification of Genetic Causation in Patients with Congenital Anomalies of the Kidney and Urinary Tract. *European urology open science*, 44, 106.

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