Resource Summary Report

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Androgen Receptor Gene Mutations Database

RRID:SCR_006887
Type: Tool

Proper Citation

Androgen Receptor Gene Mutations Database (RRID:SCR_006887)

Resource Information

URL: http://androgendb.mcgill.ca/

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Description: Comprehensive listing of androgen receptor gene mutations published in journals and meetings proceedings. The majority of mutations are point mutations identified in patients with androgen insensitivity syndrome. Information is included regarding the phenotype, the nature and location of the mutations, as well as the effects of the mutations on the androgen binding activity of the receptor. In light of the difficulty in getting new AR mutations published the curator will now accept new mutations that have not been published, provided that it is from a reputable research or clinical laboratory. The database incorporates information on the exon 1 CAG repeat expansion disease, spinobulbar muscular atrophy (SBMA), as well as CAG repeat length variations associated with risk for female breast, uterine endometrial, colorectal, and prostate cancer, as well as for male infertility. The possible implications of somatic mutations, as opposed to germline mutations, in the development of future locus-specific mutation databases (LSDBs) is discussed.
The database now provides information on the external genitalia and on sex - of - rearing. Additionally, the new version of the database has an entry to show if pathogenicity has been proven. A pdf and fully searchable version of the Database is available for download.

Abbreviations: AR Mutation DB, AndrogenDB

Synonyms: Androgen Receptor Gene Mutations Database World Wide Web Server

Resource Type: data or information resource, database, service resource, storage service resource, data repository

Defining Citation: PMID:22334387, PMID:15146455, PMID:7937057

Keywords: genitalia, gene, androgen, androgen insensitivity syndrome, androgen receptor, biochemical property, disease, kinetic property, mutation, phenotype, sex-of-rearing, genotype, FASEB list

Related Condition: Androgen insensitivity syndrome

Funding: Canadian Institutes of Health Research

Availability: The community can contribute to this resource

Resource Name: Androgen Receptor Gene Mutations Database

Resource ID: SCR_006887

Alternate IDs: nif-0000-02547, r3d100012038

Alternate URLs: https://doi.org/10.17616/R3PH1J

Old URLs: http://www.mcgill.ca/androgendb/

Record Creation Time: 20220129T080238+0000

Record Last Update: 20260808T042625+0000

Ratings and Alerts

No rating or validation information has been found for Androgen Receptor Gene Mutations Database.

No alerts have been found for Androgen Receptor Gene Mutations Database.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 57 mentions in open access literature.

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

Liao B, et al. (2026) A novel androgen receptor gene splice site mutation induces aberrant mRNA splicing and internal in-frame deletion in androgen insensitivity syndrome. BMC medical genomics, 19(1).

Sun HY, et al. (2025) A novel androgen resistance gene mutation (p.G590W) in complete androgen insensitivity syndrome: Emphasizing the need for early gonadectomy and integrated patient care. The Journal of international medical research, 53(6), 3000605251350626.

Gaikwad AS, et al. (2025) Functional and clinical insights into nuclear receptor variants for advancing precision diagnostics in male infertility. EBioMedicine, 119, 105899.

Pedroza-Torres A, et al. (2025) Deep Sequencing Reveals Novel Mutations in Androgen Receptor-Related Genes in Prostate Cancer. International journal of molecular sciences,

26(18).

Ferrante R, et al. (2024) A Very Early Diagnosis of Complete Androgen Insensitivity Syndrome Due to a Novel Variant in the AR Gene: A Neonatal Case Study. Biomedicines, 12(8).

Chen DL, et al. (2024) Complete androgen insensitivity syndrome coexisting with müllerian duct remnants: a case report and literature review. Frontiers in pediatrics, 12, 1400319.

Lee NY, et al. (2023) Clinical outcomes and genotype-phenotype correlations in patients with complete and partial androgen insensitivity syndromes. Annals of pediatric endocrinology & metabolism, 28(3), 184.

Mansour M, et al. (2023) Urethral reconstruction using amniotic membrane allograft in hereditary androgen insensitivity syndrome: a case series. Journal of surgical case reports, 2023(12), rjad652.

Liu Q, et al. (2023) Clinical characteristics, AR gene variants, and functional domains in 64 patients with androgen insensitivity syndrome. Journal of endocrinological investigation, 46(1), 151.

Estébanez-Perpiñá E, et al. (2021) Eighty Years of Targeting Androgen Receptor Activity in Prostate Cancer: The Fight Goes on. Cancers, 13(3).

Tajouri A, et al. (2021) In vitro functional characterization of androgen receptor gene mutations at arginine p.856 of the ligand-binding-domain associated with androgen insensitivity syndrome. The Journal of steroid biochemistry and molecular biology, 208, 105834.

Oduwole OO, et al. (2021) The Roles of Luteinizing Hormone, Follicle-Stimulating Hormone and Testosterone in Spermatogenesis and Folliculogenesis Revisited. International journal of molecular sciences, 22(23).

Poon KS, et al. (2021) A novel de novo androgen receptor nonsense mutation in a sex-reversed 46,XY infant. Human genome variation, 8(1), 35.

Marino L, et al. (2021) Testosterone-induced increase in libido in a patient with a loss-of-function mutation in the AR gene. Endocrinology, diabetes & metabolism case reports, 2021.

Coelho ML, et al. (2021) Complete Androgen Insensitivity Syndrome: A Rare Case of Prenatal Diagnosis. Revista brasileira de ginecologia e obstetricia : revista da Federacao Brasileira das Sociedades de Ginecologia e Obstetricia, 43(9), 710.

Wang C, et al. (2021) Wnt/?-catenin signal transduction pathway in prostate cancer and associated drug resistance. Discover oncology, 12(1), 40.

Rocca MS, et al. (2020) Comparison of NGS panel and Sanger sequencing for genotyping CAG repeats in the AR gene. Molecular genetics & genomic medicine, 8(6), e1207.

Liu Q, et al. (2020) Clinical, hormonal and genetic characteristics of androgen insensitivity syndrome in 39 Chinese patients. Reproductive biology and endocrinology : RB&E, 18(1), 34.

Villagomez DAF, et al. (2020) Androgen Receptor Gene Variants in New Cases of Equine Androgen Insensitivity Syndrome. Genes, 11(1).

Chen F, et al. (2020) Computational analysis of androgen receptor (AR) variants to decipher the relationship between protein stability and related-diseases. Scientific reports, 10(1), 12101.