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 repository, service resource, storage service resource, database, data or information resource

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

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

Record Creation Time: 20220129T080238+0000

Record Last Update: 20250428T053255+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 50 mentions in open access literature.

Listed below are recent publications. The full list is available at FDI Lab - SciCrunch.org.

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.

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.

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.

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

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.

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.

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.

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).

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.

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.

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.

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

Kharrat M, et al. (2019) Identification of two additional novel mutations in the AR gene associated with severe forms of androgen insensitivity syndrome. Steroids, 152, 108489.

Liu S, et al. (2019) Severe forms of complete androgen insensitivity syndrome caused by a p.Q65X novel mutation in androgen receptor: Clinical manifestations, imaging findings and molecular genetics. Steroids, 144, 47.

Malcher A, et al. (2019) Novel Mutations Segregating with Complete Androgen Insensitivity

Syndrome and their Molecular Characteristics. International journal of molecular sciences, 20(21).

Vaidyanathan P, et al. (2018) Partial androgen insensitivity syndrome presenting as pubertal gynecomastia: clinical and hormonal findings and a novel mutation in the androgen receptor gene. Endocrinology, diabetes & metabolism case reports, 2018.

Nadal M, et al. (2017) Structure of the homodimeric androgen receptor ligand-binding domain. Nature communications, 8, 14388.

Pritsini F, et al. (2017) Psychological Aspects of Androgen Insensitivity Syndrome: Two Cases Illustrating Therapeutical Challenges. Case reports in endocrinology, 2017, 8313162.

Li L, et al. (2017) A missense mutation in the androgen receptor gene causing androgen insensitivity syndrome in a Chinese family. Asian journal of andrology, 19(2), 260.