# ABBREVIATIONS

## General Abbreviations

<table>
<thead>
<tr>
<th>Abbreviation</th>
<th>Description</th>
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<tbody>
<tr>
<td>ACES</td>
<td>Aravind Comprehensive Eye Survey</td>
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<tr>
<td>ACG</td>
<td>Angle closure glaucoma</td>
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<tr>
<td>aCGH</td>
<td>Array based comparative genomic hybridization</td>
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<td>ADHD</td>
<td>Attention deficit hyperactivity disorder</td>
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<td>AKAP13</td>
<td>A kinase (PRKA) anchor protein 13</td>
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<tr>
<td>AKT</td>
<td>v-akt murine thymoma viral oncogene homolog 1</td>
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<tr>
<td>AMD</td>
<td>Age related macular degeneration</td>
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<tr>
<td>APEDS</td>
<td>Andhra Pradesh Eye Disease Study</td>
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<td>AS</td>
<td>de novo assembly of a genome</td>
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<td>ASTN2</td>
<td>Astrotactin 2</td>
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<td>ATOH7</td>
<td>Atonal homolog 7 (Drosophila)</td>
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<tr>
<td>AVGR8</td>
<td>Homo sapiens mRNA for autogenous vein graft remodeling associated protein 8</td>
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<td>BAF</td>
<td>B allele frequency</td>
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<tr>
<td>BBS</td>
<td>Bardet Biedl Syndrome</td>
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<td>BLAST</td>
<td>Basic local alignment search tool</td>
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<td>BLAT</td>
<td>Blast like alignment tool</td>
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<tr>
<td>BPD</td>
<td>Bipolar disorder</td>
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<td>C/D</td>
<td>Cup to disk</td>
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<td>C245-C433</td>
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<td>CARD10</td>
<td>Caspase recruitment domain family, member 10</td>
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<td>Caveolin 1</td>
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<td>CAV2</td>
<td>Caveolin 2</td>
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<td>CBS</td>
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<td>Chemokine (C-C motif) ligand 3-like 1</td>
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<td>CCT</td>
<td>Central corneal thickness</td>
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<td>Cdkn1c</td>
<td>Cyclin-dependent kinase inhibitor 1C (p57, Kip2)</td>
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<td>CDKN2B-AS1</td>
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<tr>
<td>CEAS</td>
<td>Cis regulatory element annotation system</td>
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<tr>
<td>CFHR1</td>
<td>Complement factor H-related 1</td>
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<td>CFHR3</td>
<td>Complement factor H-related 3</td>
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<td>CGS</td>
<td>Chennai Glaucoma Study</td>
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<td>CGS</td>
<td>Contiguous gene syndrome</td>
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<td>CHARGE</td>
<td>Coloboma of the eye, heart defects, atresia of the nasal choanae, retardation of growth and/or development, genital and/or urinary abnormalities, and ear abnormalities and deafness</td>
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<td>CHEK2</td>
<td>checkpoint kinase 2</td>
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<tr>
<td>ChIP-Seq</td>
<td>Chromatin immunoprecipitation and sequencing</td>
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<td>CKAP4</td>
<td>Cytoskeleton Associated Protein 4</td>
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<td>CNAG</td>
<td>Copy Number Analyzer for GeneChip</td>
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<td>Copy number polymorphism</td>
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<td>CNTN4</td>
<td>Contactin 4</td>
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<td>CNV</td>
<td>COPY number variation</td>
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<td>CNVRs</td>
<td>Copy number variantion regions</td>
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<td>COL5A1</td>
<td>Collagen, type V, alpha 1</td>
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<td>COL8A2</td>
<td>Collagen, type VIII, alpha 2</td>
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<td>CTNND2</td>
<td>Catenin (cadherin-associated protein), delta 2</td>
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<td>CYP1B1</td>
<td>Cytochrome P4501B1</td>
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<td>CYP21</td>
<td>Cytochrome P450, family 21</td>
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<td>DAVID</td>
<td>Database for Annotation, Visualization and Integrated Discovery</td>
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<td>Doublecortin-like kinase 1</td>
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<td>DE</td>
<td>Differentially expressed</td>
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<td>DECIPHER</td>
<td>Database of Chromosomal Imbalance and Phenotype in Humans Using Ensembl Resources</td>
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<td>DGV</td>
<td>Database of Genomic Variants</td>
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<td>Dulbecco's Modified Eagle Medium</td>
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<td>Dimethyl sulfoxide</td>
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<td>Dmx-like 1</td>
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<td>DNA</td>
<td>Deoxyribonucleic acid</td>
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<td>DSBs</td>
<td>DNA double strand breaks</td>
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<td>DUSP7</td>
<td>Dual specificity phosphatise</td>
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<td>dyI</td>
<td>Dysgenetic lens</td>
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<td>DZ</td>
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<td>ELOVL5</td>
<td>ELOVL fatty acid elongase 5</td>
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<td>Foxe3 binding region</td>
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<td>FBS</td>
<td>Fetal bovine serum</td>
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<td>FDR</td>
<td>False discovery rate</td>
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<td>FISH</td>
<td>Fluorescence in situ hybridization</td>
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<td>GO</td>
<td>Gene ontology</td>
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<td>GWAS</td>
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<td>Hidden Markov model</td>
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<tr>
<td>HMM</td>
<td>Hidden Markov Model</td>
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<td>HSD17B10</td>
<td>hydroxysteroid (17-beta) dehydrogenase 10</td>
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<td>High-testion glaucoma</td>
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<td>HUGO</td>
<td>Human genome organization</td>
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<td>ID</td>
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<td>-----------------------------------------------</td>
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<td>ID</td>
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<td>interleukin 5 receptor, alpha</td>
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<td>IOP</td>
<td>Intra ocular pressure</td>
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<td>JOAG</td>
<td>Juvenile open angle glaucoma</td>
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<tr>
<td>kb</td>
<td>Kilobase</td>
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<td>KEGG</td>
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<td>LCR</td>
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<td>MAQ</td>
<td>Mapping and Assembly with Qualities</td>
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<td>Mb</td>
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<td>MCODE</td>
<td>Molecular complex detection</td>
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<td>MGB</td>
<td>minor groove binder</td>
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<td>MiMI</td>
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<td>MLP A</td>
<td>Multiplex Ligation-dependent Probe Amplification</td>
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<td>mm</td>
<td>Mili meter</td>
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<td>Neurexin 1</td>
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<td>Ocular hypertension</td>
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<td>ONH</td>
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<td>OPTN</td>
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**Amino acids**

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<th>Amino Acid</th>
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<tr>
<td>G</td>
<td>Glycine (Gly)</td>
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<td>P</td>
<td>Proline (Pro)</td>
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<td>A</td>
<td>Alanine (Ala)</td>
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<tr>
<td>V</td>
<td>Valine (Val)</td>
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<tr>
<td>L</td>
<td>Leucine (Leu)</td>
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<td>I</td>
<td>Isoleucine (Ile)</td>
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<td>C</td>
<td>Cysteine (Cys)</td>
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<td>Letter</td>
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<td>S</td>
<td>Serine (Ser)</td>
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<td>T</td>
<td>Threonine (Thr)</td>
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