## Bharti Morar

## List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

42 1,959 21 44 g-index

47 2,226 6.2 3.51 ext. papers ext. citations avg, IF L-index

| #  | Paper   | IF      | Citations |
|----|---|---------|-----------|
| 42 | A patient with homozygous nonsense variants in two Leigh syndrome disease genes: Distinguishing a dual diagnosis from a hypomorphic protein-truncating variant. <i>Human Mutation</i> , <b>2019</b> , 40, 893-898                             | 4.7     | 7         |
| 41 | Whole genome sequencing of 91 multiplex schizophrenia families reveals increased burden of rare, exonic copy number variation in schizophrenia probands and genetic heterogeneity. <i>Schizophrenia Research</i> , <b>2018</b> , 197, 337-345 | 3.6     | 10        |
| 40 | The longevity gene Klotho is differentially associated with cognition in subtypes of schizophrenia. <i>Schizophrenia Research</i> , <b>2018</b> , 193, 348-353  | 3.6     | 8         |
| 39 | A Roma founder mutation causes a novel phenotype of centronuclear myopathy with rigid spine. <i>Neurology</i> , <b>2018</b> , 91, e339-e348   | 6.5     | 12        |
| 38 | Longevity Klotho gene polymorphism and the risk of dementia in older men. <i>Maturitas</i> , <b>2017</b> , 101, 1-5   | 5       | 11        |
| 37 | Exome array analysis suggests an increased variant burden in families with schizophrenia. <i>Schizophrenia Research</i> , <b>2017</b> , 185, 9-16   | 3.6     | 17        |
| 36 | founder mutation in the Roma population causes recessive variant of H-ABC. <i>Neurology</i> , <b>2017</b> , 89, 1821  | -168-28 | 19        |
| 35 | Transcriptome-wide effects of a POLR3A gene mutation in patients with an unusual phenotype of striatal involvement. <i>Human Molecular Genetics</i> , <b>2016</b> , 25, 4302-4314   | 5.6     | 30        |
| 34 | Integrity of genome-wide genotype data from low passage lymphoblastoid cell lines. <i>Genomics Data</i> , <b>2016</b> , 9, 18-21  |         | 5         |
| 33 | The AQP1 del601G mutation in different European Romani (Gypsy) populations. <i>Blood Transfusion</i> , <b>2016</b> , 14, 580-581  | 3.6     | 1         |
| 32 | Integrin Alpha 8 Recessive Mutations Are Responsible for Bilateral Renal Agenesis in Humans. <i>American Journal of Human Genetics</i> , <b>2014</b> , 94, 799  | 11      | 78        |
| 31 | Integrin alpha 8 recessive mutations are responsible for bilateral renal agenesis in humans. <i>American Journal of Human Genetics</i> , <b>2014</b> , 94, 288-94   | 11      | 71        |
| 30 | Founder p.Arg 446* mutation in the PDHX gene explains over half of cases with congenital lactic acidosis in Roma children. <i>Molecular Genetics and Metabolism</i> , <b>2014</b> , 113, 76-83  | 3.7     | 16        |
| 29 | Roma (Gypsies): Genetic Studies <b>2013</b> ,   |         | 5         |
| 28 | Promoter polymorphisms in two overlapping 6p25 genes implicate mitochondrial proteins in cognitive deficit in schizophrenia. <i>Molecular Psychiatry</i> , <b>2012</b> , 17, 1328-39  | 15.1    | 14        |
| 27 | Autosomal-recessive congenital cerebellar ataxia is caused by mutations in metabotropic glutamate receptor 1. <i>American Journal of Human Genetics</i> , <b>2012</b> , 91, 553-64  | 11      | 67        |
| 26 | Deleterious GRM1 mutations in schizophrenia. <i>PLoS ONE</i> , <b>2012</b> , 7, e32849  | 3.7     | 45        |

## (2003-2012)

| 25 | Impact of the Reelin signaling cascade (ligands-receptors-adaptor complex) on cognition in schizophrenia. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , <b>2012</b> , 159B, 392-                               | -40 <sup>345</sup> | 23  |
|----|--|--------------------|-----|
| 24 | Polymorphisms associated with normal memory variation also affect memory impairment in schizophrenia. <i>Genes, Brain and Behavior</i> , <b>2011</b> , 10, 410-7   | 3.6                | 35  |
| 23 | LTBP2 and CYP1B1 mutations and associated ocular phenotypes in the Roma/Gypsy founder population. <i>European Journal of Human Genetics</i> , <b>2011</b> , 19, 326-33   | 5.3                | 50  |
| 22 | Neuregulin 3 (NRG3) as a susceptibility gene in a schizophrenia subtype with florid delusions and relatively spared cognition. <i>Molecular Psychiatry</i> , <b>2011</b> , 16, 860-6   | 15.1               | 60  |
| 21 | Focal epilepsy of probable temporal lobe origin in a Gypsy family showing linkage to a novel locus on 7p21.3. <i>Epilepsy Research</i> , <b>2011</b> , 96, 101-8   | 3                  | 8   |
| 20 | A novel GEFS+ locus on 12p13.33 in a large Roma family. <i>Epilepsy Research</i> , <b>2011</b> , 97, 198-207   | 3                  | 5   |
| 19 | Psychosis susceptibility gene ZNF804A and cognitive performance in schizophrenia. <i>Archives of General Psychiatry</i> , <b>2010</b> , 67, 692-700  |                    | 120 |
| 18 | Partial epilepsy syndrome in a Gypsy family linked to 5q31.3-q32. <i>Epilepsia</i> , <b>2009</b> , 50, 1679-88   | 6.4                | 15  |
| 17 | KIBRA genetic polymorphism influences episodic memory in later life, but does not increase the risk of mild cognitive impairment. <i>Journal of Cellular and Molecular Medicine</i> , <b>2008</b> , 12, 1672-6                                   | 5.6                | 71  |
| 16 | Evaluation of association of SNPs in the TNF alpha gene region with schizophrenia. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , <b>2007</b> , 144B, 318-24  | 3.5                | 18  |
| 15 | A newly discovered founder population: the Roma/Gypsies. <i>BioEssays</i> , <b>2005</b> , 27, 1084-94  | 4.1                | 73  |
| 14 | Vlax Roma history: what do coalescent-based methods tell us?. <i>European Journal of Human Genetics</i> , <b>2004</b> , 12, 285-92   | 5.3                | 21  |
| 13 | The effective mutation rate at Y chromosome short tandem repeats, with application to human population-divergence time. <i>American Journal of Human Genetics</i> , <b>2004</b> , 74, 50-61  | 11                 | 320 |
| 12 | Mutation history of the roma/gypsies. American Journal of Human Genetics, 2004, 75, 596-609  | 11                 | 106 |
| 11 | Mutation screening of the N-myc downstream-regulated gene 1 (NDRG1) in patients with Charcot-Marie-Tooth Disease. <i>Human Mutation</i> , <b>2003</b> , 22, 129-35   | 4.7                | 41  |
| 10 | A standard protocol for single nucleotide primer extension in the human genome using matrix-assisted laser desorption/ionization time-of-flight mass spectrometry. <i>Rapid Communications in Mass Spectrometry</i> , <b>2003</b> , 17, 1195-202 | 2.2                | 19  |
| 9  | Genealogy and genes: tracing the founding fathers of Tristan da Cunha. <i>European Journal of Human Genetics</i> , <b>2003</b> , 11, 705-9   | 5.3                | 15  |
| 8  | Partial deficiency of the C-terminal-domain phosphatase of RNA polymerase II is associated with congenital cataracts facial dysmorphism neuropathy syndrome. <i>Nature Genetics</i> , <b>2003</b> , 35, 185-9                                    | 36.3               | 116 |

| 7 | Genetic substructure in South African Bantu-speakers: evidence from autosomal DNA and Y-chromosome studies. <i>American Journal of Physical Anthropology</i> , <b>2002</b> , 119, 175-85 | 2.5 | 46  |  |
|---|--|-----|-----|--|
| 6 | Y-chromosomal evidence for a strong reduction in male population size of Yakuts. <i>Human Genetics</i> , <b>2002</b> , 110, 198-200  | 6.3 | 21  |  |
| 5 | The Human Genome as Archive: Some Illustrations from the South <b>2002</b> , 179-192   |     | 1   |  |
| 4 | Origins and divergence of the Roma (gypsies). American Journal of Human Genetics, <b>2001</b> , 69, 1314-31  | 11  | 156 |  |
| 3 | A global survey of haplotype frequencies and linkage disequilibrium at the DRD2 locus. <i>Human Genetics</i> , <b>1998</b> , 103, 211-27   | 6.3 | 188 |  |
| 2 | The molecular characterization of Gaucher disease in South Africa. <i>Clinical Genetics</i> , <b>1996</b> , 50, 78-84  | 4   | 8   |  |
| 1 | Alpha-1-antitrypsin variation in Southern Africa. <i>Human Heredity</i> , <b>1986</b> , 36, 238-42   | 1.1 | 7   |  |