

REGIONES ASOCIADAS A AUTISMO INCLUIDAS

AUTISM LOCI

Syndromic and Recognized Loci

The following table includes loci for which multiple examples exist supporting a role in autism. In some cases, an established syndrome is associated with the gene or locus and autistic features may or may not be a consistent feature of the syndrome. If a copy number abnormality (CNA) encompassing one of the loci below is found in an individual with autistic features, a plausible (though not necessarily definitive) conclusion is that the abnormality contributes to the phenotype. If the tested individual is a fetus or very young child without obvious autistic features, there is a presumed increased risk of the development of autistic features, although the magnitude of that risk varies greatly among the different loci.

CHROMOSOME BAND	GENE(S)/ SYNDROME	REFERENCES	HYPERLINK – Please click on PMID # below, or type http://www.ncbi.nlm.nih.gov/pubmed/ followed by the individual PMID #	CHROMOSOME BAND	GENE(S)/ SYNDROME	REFERENCES	HYPERLINK – Please click on PMID # below, or type http://www.ncbi.nlm.nih.gov/pubmed/ followed by the individual PMID #
1p36.31p36.23	<i>CAMTA1</i>	Mikhail et al. 2011 Thevenon et al. 2012	22031302 22693284	6p21.32	<i>SYNGAP1</i>	Pinto et al. 2010 Hamdan et al. 2011	20531469 21237447
1q21.1		Szatmari et al. 2007 Brunetti-Pierri et al. 2008 Mefford et al. 2008 Rosenfeld et al. 2012	17322880 19029900 18784092 22317977	6q25.3	<i>ARID1B</i>	Nord et al. 2011 Halgren et al. 2012	21448237 21801163
2p25.3	<i>SNTG2</i>	Rosenfeld et al. 2010 Talkowski et al. 2012	20808228 22521361	7q11.22	<i>AUTS2</i>	Sultana et al. 2002 Huang et al. 2010 Talkowski et al. 2012 Beunders et al. 2013	12160723 20635338 22521361 23332918
2p16.3	<i>NRXN1</i>	Ching et al. 2010 Schaaf et al. 2012 Dabell et al. 2013	20468056 22617343 23495017	7q11.23	Williams	Berg et al. 2007 Sanders et al. 2011 Tordjman et al. 2012 Dixit et al. 2013	17666889 21658581 22412832 22369319
2p16.1p15		Rajcan-Separovic et al. 2007 Qiao et al. 2009	16963482 19625284	8q12.1q12.2	<i>CHD7/CHARGE</i>	Johansson et al. 2006 Johansson et al. 2010 O’Roak et al. 2012	16359593 19709852 22495309
2q23.1	<i>MBD5</i>	van Bon et al. 2010 Williams et al. 2010 Talkowski et al. 2011 Chung et al. 2012	19809484 19904302 21981781 22085900	9q34.11	<i>STXBP1</i>	Milh et al. 2011 Campbell et al. 2012	21770924 22722545
2q33.1	<i>SATB2</i>	Marshall et al. 2008 Talkowski et al. 2012	18252227 22521361	9q34.13	<i>TSC1/Tuberous sclerosis</i>	Smalley 1998 Wiznitzer 2004 Wong 2006	9813776 15563013 16901420
2q36.2	<i>CUL3</i>	Kong et al. 2012 O’Roak et al. 2012	22914163 22495309	9q34.3	<i>EHMT1/Kleefstra</i>	Stewart & Kleefstra 2007 Talkowski et al. 2012	17910072 22521361
2q37.3	<i>HDAC4</i>	Falk & Casas 2007 Galasso et al. 2008 Williams et al. 2010 Leroy et al. 2012 Mazzone et al. 2012	17910077 18658079 20691407 23073310 2222775	10q11.22q11.23		Stankiewicz et al. 2012 Girirajan et al. 2013	21948486 23375656
3p13	<i>FOXP1</i>	Hamdan et al. 2010 O’Roak et al. 2011	20950788 21572417	10q22.3q23.2		Balciuniene et al. 2007 Alliman et al. 2010 van Bon et al. 2011	17436248 20345475 21248748
3q24	<i>SLC9A9</i>	Morrow et al. 2008	18621663	10q23.31	<i>P TEN</i>	Butler et al. 2005 Buxbaum et al. 2007 O’Roak et al. 2012	15805158 17427195 22495309
3q29		Willatt et al. 2005 Ballif et al. 2008 Cobb et al. 2010 Quintero-Rivera et al. 2010	15918153 18471269 20832509 20830797	11p14.1	<i>BDNF/WAGRO</i>	Xu et al. 2008 Shinawi et al. 2011 Ernst et al. 2012	19096215 21567907 23044507
5p13.2	<i>NIPBL/Cornelia de Lange</i>	Krantz et al. 2004 Nakanishi et al. 2012	15146186 22740374	11p13	<i>PAX6/WAGR</i>	Davis et al. 2008 Xu et al. 2008	18322702 19096215
5q14.3	<i>MEF2C</i>	Novara et al. 2010 Nowakowska et al. 2010 Paciorkowski et al. 2013	20412115 20333642 23389741	11q13.3q13.4	<i>SHANK2</i>	Berkel et al. 2010 Pinto et al. 2010 Sanders et al. 2012	20473310 20531469 22495306
				11q23.3	<i>MLL/Wiedemann-Steiner</i>	Jones et al. 2012	22795537

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12p13.33	<i>ERC1</i>	Thevenon et al. 2013	22713806	17q21.31		Grisart et al. 2009	19502243
12p13.1	<i>GRIN2B</i>	Tarabeux et al. 2011 O'Roak et al. 2012 Talkowski et al. 2012	22833210 23160955 22521361	19q13.33	<i>SHANK1</i>	Sato et al. 2012	22503632
12p12.1	<i>SOX5</i>	Lamb et al. 2012	22290657	21 (trisomy)	Down	Kent et al. 1999 Molloy et al. 2009 Moss et al. 2012	10210247 19198037 22589453
13q13.3	<i>NBEA</i>	Smith et al. 2002 Castermans et al. 2003	12826745 12746398	21q22.13	<i>DYRK1A</i>	van Bon et al. 2011 Iossifov et al. 2012 O'Roak et al. 2012	21294719 22542183 23160955
14q11.2	<i>CHD8</i>	O'Roak et al. 2012 O'Roak et al. 2012 Talkowski et al. 2012	22495309 23160955 22521361	22q11.21	<i>GNB1L/</i> DiGeorge, Velocardiofacial	Fine et al. 2005 Vorstman et al. 2006 Antshel et al. 2007 Chen et al. 2012 Van Campenhout et al. 2012	16134031 16926618 17180713 22095694 22876571
14q12	<i>FOXP1</i>	Kortum et al. 2011	21441262	22q13.33	<i>SHANK3/Phelan- McDermid</i>	Durand et al. 2007 Peeters et al. 2008 Philippe et al. 2008 Phelan & McDermid 2012	17173049 19239079 18625665 22670140
14q24.3q31.1	<i>NRXN3</i>	Griswold et al. 2012 Vaags et al. 2012	22543975 22209245	Xp22.32p22.31	<i>NLGN4X</i>	Jamain et al. 2003 Macarov et al. 2007 Kent et al. 2008 Willemsen et al. 2012 Yu et al. 2013	12669065 17391250 18413370 22796527 23352163
15q11.2		Doombos et al. 2009 Burnside et al. 2011 von der Lippe et al. 2011 Madrigal et al. 2012	19328872 21359847 21187176 22842191	Xp22.13	<i>CDKL5</i>	Weaving et al. 2004 Erez et al. 2009 Russo et al. 2009	15492925 19471977 19241098
15q11q13		Cook et al. 1997 Peters et al. 2004 Battaglia et al. 2010 Hogart et al. 2010 Simon et al. 2010	9106540 15521981 20981774 18840528 19548260	Xp22.11	<i>PTCHD1, DDX53</i>	Noor et al. 2010 Pinto et al. 2010 Filges et al. 2011	20844286 20531469 21091464
15q13.3	<i>CHRNA7</i>	Ben-Shachar et al. 2009 Miller et al. 2009 Hoppman-Chaney et al. 2013	19289393 18805830 22775350	Xp21.3p21.2	<i>IL1RAPL1</i>	Bhat et al. 2008 Piton et al. 2008 Awadalla et al. 2010 Pinto et al. 2010	18005360 18801879 20797689 20531469
15q24		Sharp et al. 2007 McInnes et al. 2010 Mefford et al. 2012	17360722 20678247 22180641	Xp11.23	<i>SYN1</i>	Garcia et al. 2004 Fassio et al. 2011	14985377 21441247
16p13.3	<i>TSC2/Tuberous sclerosis</i>	Smalley 1998 Wiznitzer 2004 Wong 2006 Numis et al. 2011 O'Roak et al. 2012	9813776 15563013 16901420 21403110 22495309	Xp11.23	<i>PQBP1/</i> Renpenning	Germanaud et al. 2011	20950397
16p13.3	<i>CREBBP/</i> Rubinstein-Taybi	Schorry et al. 2008	18792986	Xp11.23p11.22		Bonnet et al. 2006 Giorda et al. 2009 Chung et al. 2011 Edens et al. 2011	16900295 19716111 21689796 21418194
16p13.11		Ullmann et al. 2007 Hannes et al. 2009	17480035 18550696	Xp11.22	<i>PHF8</i>	Qiao et al. 2008 Nava et al. 2012	18498374 23092983
16p12.2		Girirajan et al. 2010	20154674	Xq22.1	<i>PCDH19</i>	Piton et al. 2011 Camacho et al. 2012 Marini et al. 2012	20479760 22504056 22946748
16p11.2	<i>KCTD13</i>	Kumar et al. 2008 Weiss et al. 2008 Hanson et al. 2010 Rosenfeld et al. 2010 Golzio et al. 2012 Zufferey et al. 2012	18156158 18184952 20613623 21731881 22596160 23054248	Xq24	<i>UPF3B</i>	Laumonnier et al. 2010 Addington et al. 2011 Lynch et al. 2012	19238151 20479756 22609145
16q24.3	<i>ANKRD11</i>	Marshall et al. 2008 Willemsen et al. 2010 Youngs et al. 2011	18252227 19920853 21527850	Xq26.3	<i>SLC9A6/</i> Christianson	Garbern et al. 2010 Mignot et al. 2013	20395263 22541666
17p13.3		Bi et al. 2009 Bruno et al. 2010 Capra et al. 2012	19136950 20452996 23035971	Xq27.3	<i>FMR1/Fragile X</i>	Reddy 2005 Chaste et al. 2012 Smith et al. 2012 Wolff et al. 2012	15655077 22738402 22264109 23200289
17p11.2	<i>RAI1/Smith- Magenis, Potocki-Lupski</i>	Hicks et al. 2008 Nakamine et al. 2008 Treadwell-Deering et al. 2010	18301319 17334992 20110824	Xq28	<i>AFF2/Fragile X E</i>	Abrams et al. 1997 Sahoo et al. 2011 Stettner et al. 2011 Mondal et al. 2012	9034011 22065534 21739600 22773736
17q11.2	<i>NF1/Neurofibro- matosis</i>	Garg et al. 2013 Walsh et al. 2013	23163236 23163951	Xq28	<i>SLC6A8/Creatine deficiency</i>	Poo-Arguelles et al. 2006	16601898
17q12	Renal cysts and diabetes	Loirat et al. 2010 Moreno-De-Luca et al. 2010 Brandt et al. 2012 Dixit et al. 2012	20587423 21055719 22488896 22887843	Xq28	<i>RAB39B</i>	Giannandrea et al. 2010	20159109

According to hg19 genome build

Loci with Limited Evidence for Involvement in Autism

The following table includes loci for which the evidence of a role in autism is less certain, or for which copy number evaluation may not be conclusive. Reasons for the uncertainty include the following: 1) there may be only single examples of a CNA or null mutation; 2) multiple examples of CNAs exist, but a well-defined critical region remains unknown; 3) multiple examples exist, but a control population was not tested; 4) examples in both cases and controls have been reported, but the increase in cases was not statistically significant; 5) only missense mutations have been reported, and therefore the effect of a copy number change is unclear. As new information becomes available, the status of many genes in this list will likely change. Signature Genomics may not always comment in results reports about copy number changes involving these loci; the decision to do so will depend on the details of the individual case and the nature of the published evidence, as well Signature Genomics' experience.

CHROMOSOME BAND	GENE(S)/ SYNDROME	REFERENCES	HYPERLINK – Please click on PMID # below, or type http://www.ncbi.nlm.nih.gov/pubmed/ followed by the individual PMID #	CHROMOSOME BAND	GENE(S)/ SYNDROME	REFERENCES	HYPERLINK – Please click on PMID # below, or type http://www.ncbi.nlm.nih.gov/pubmed/ followed by the individual PMID #
1p36.23	<i>CA6</i>	Bucan et al. 2009	19557195	2q14.3	<i>CNTNAP5</i>	Pagnamenta et al. 2010	20346443
1p36.12	<i>EPHB2</i>	Kong et al. 2012 Sanders et al. 2012	22914163 22495306	2q21.1		Dharmadhikari et al. 2012	22543972
1p34.2	<i>RIMS3</i>	Kumar et al. 2010	19546099	2q22.1q22.3		Mulatinho et al. 2012	22686481
1p31.3	<i>NFIA</i>	Iossifov et al. 2012	22542183	2q23.1	<i>KIF5C</i>	Awadalla et al. 2010	20797689
1p21.1p13.2		Piccione et al. 2010	20101695	2q23.3q24.1	<i>GALNT13</i>	Bucan et al. 2009	19557195
1p13.3	<i>NTNG1</i>	Borg et al. 2005	15870826	2q24		Sebat et al. 2007 Newbury et al. 2009 Chen et al. 2010 Krepischi et al. 2010 Traylor et al. 2012	17363630 19267418 20346423 21204806 23112752
1q21.3	<i>POGZ</i>	Iossifov et al. 2012 Neale et al. 2012	22542183 22495311	2q24.2	<i>TBR1</i>	O'Roak et al. 2012 O'Roak et al. 2012	22495309 23160955
1q23.3	<i>NOS1AP</i>	Delorme et al. 2010	20602773	2q24.3	<i>SCN2A</i>	Weiss et al. 2003 Iossifov et al. 2012 Sanders et al. 2012	12610651 22542183 22495306
1q23.3q24.2		Della Monica et al. 2007	17937430	2q24.3	<i>SCN1A</i>	Weiss et al. 2003	12610651
1q42.2	<i>DISC1</i>	Williams et al. 2009 Crepel et al. 2010 Sanders et al. 2012	19606485 20002455 22495306	2q31.1	<i>LRP2</i>	Ionita-Laza et al. 2012 Iossifov et al. 2012	22578327 22542183
1q43	<i>CHRM3</i>	Perrone et al. 2012 Petersen et al. 2013	22186213 23253743	2q31.1	<i>RAPGEF4</i>	Bacchelli et al. 2003	14593429
1q44	<i>OR1C1</i>	Bucan et al. 2009	19557195	2q31.2	<i>TTN</i>	Iossifov et al. 2012	22542183
2p25.3		Meyer et al. 2012	22157634	2q34	<i>MAP2</i>	Pescucci et al. 2003 Bisgaard et al. 2006 Brandau et al. 2008 Rosenfeld et al. 2010	14986829 16955412 18203195 20808228
2p25.2	<i>SOX11</i>	Lo-Castro et al. 2009	18992374	2q34	<i>ERBB4</i>	Pinto et al. 2010	20531469
2p22.3	<i>SPAST</i>	Talkowski et al. 2012	22521361	2q36		Borg et al. 2002	12070244
2p21		Matsunami et al. 2013	23341896	2q36.3	<i>TRIP12</i>	Chahrouh et al. 2012 Iossifov et al. 2012	22511880 22542183
2p15		Matsunami et al. 2013	23341896	3p26.3p26.2	<i>CNTN4</i>	Glessner et al. 2009 Roohi et al. 2009 Cottrell et al. 2011 Guo et al. 2012	19404257 18349135 21308999 22750301
2p13.2	<i>RAB11FIP5</i>	Roohi et al. 2008	18384058	3p26.1		Celestino-Soper et al. 2011	21865298
2q14.1	<i>DPP10</i>	Marshall et al. 2008 Girirajan et al. 2013 Matsunami et al. 2013	18252227 23375656 23341896				

Loci with Limited Evidence for Involvement in Autism

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3p26.1p25.3	<i>OXTR</i>	Gregory et al. 2009 Gunnarsson & Foy Bruun 2010	19845972 21082655	4q23	<i>EIF4E</i>	Neves-Pereira et al. 2009 Kelleher et al. 2012	19556253 22558107
3p24.3	<i>TBC1D5</i>	Pinto et al. 2010	20531469	4q32.1		Wang et al. 2009	19764020
3p24.1	<i>RBMS3</i>	O'Roak et al. 2011	21572417	4q32q34		Ramanathan et al. 2004	15090072
3p22.2	<i>XIRP1</i>	O'Roak et al. 2011	21572417	4q35.2		Gilling et al. 2008 Chien et al. 2010 Neale et al. 2012	18183041 20236125 22495311
3p21.1	<i>CACNA1D</i>	Iossifov et al. 2012 O'Roak et al. 2012	22542183 22495309	5q13.1q13.2	<i>SLC30A5</i>	O'Roak et al. 2011	21572417
3p14.2	<i>FHIT</i>	Sebat et al. 2007 Girirajan et al. 2013	17363630 23375656	5q14.1	<i>HOMER1</i>	Kelleher et al. 2012	22558107
3p14.2p14.1		Tao et al. 2011	21276947	5q22.2	<i>YTHDC2</i>	O'Roak et al. 2012	22495309
3p14.1	<i>ATXN7</i>	van Daalen et al. 2011	21837366	5q23.1		Matsunami et al. 2013	23341896
3p14.1	<i>SUCLG2</i>	Bucan et al. 2009 Matsunami et al. 2013	19557195 23341896	5q31.3	<i>PCDHGA11</i>	Sato et al. 2012	22503632
3p12.3	<i>CNTN3</i>	Vaags et al. 2012	22209245	6p22.3		Celestino-Soper et al. 2012 Baroy et al. 2013 Di Benedetto et al. 2013	22480366 23294540 23324214
3q11.2	<i>EPHA6</i>	Pinto et al. 2010	20531469	6p22.2		Rosenfeld et al. 2010	20808228
3q13.2q13.31		Molin et al. 2012 Wisniewicka-Kowalik et al. 2012	22180640 23032108	6p12.1	<i>KIAA1586</i>	Bucan et al. 2009 O'Roak et al. 2012 Girirajan et al. 2013	19557195 22495309 23375656
3q21.3	<i>MBD4</i>	Cukier et al. 2010	19921286	6q16.1	<i>GPR63</i>	Derwinska et al. 2009	18717687
3q26.31	<i>NLGN1</i>	Glessner et al. 2009 O'Roak et al. 2012	19404257 22495309	6q16.3	<i>GRIK2</i>	Jamain et al. 2002 Strutz-Seebohm et al. 2006 Griswold et al. 2012	11920157 17167233 22543975
3q26.32	<i>TBL1XR1</i>	O'Roak et al. 2012 O'Roak et al. 2012	22495309 23160955	6q22.31	<i>PLN</i>	Marshall et al. 2008	18252227
4p16.3	<i>TNIP2</i>	Salyakina et al. 2011	22016809	6q25.1q25.3		Sukumar et al. 1999	10528241
4p16.1	<i>WFS1</i> Wolfram-like	Rendtorff et al. 2011	21538838	6q26	<i>PARK2</i>	Glessner et al. 2009 O'Roak et al. 2012 Girirajan et al. 2013	19404257 22495309 23375656
4p12	<i>GABRA4</i> , <i>GABRB1</i>	Vincent et al. 2006 Kakinuma et al. 2008	16556609 18163449	7p22.3		Vulto-van Silfhout et al. 2012	22822384
4q12	<i>SCFD2</i>	Pinto et al. 2010	20531469	7p21.3	<i>ICA1</i>	Salyakina et al. 2011 Iossifov et al. 2012	22016809 22542183
4q13.3		Matsunami et al. 2013	23341896	7p21.3	<i>NXP1</i>	Cukier et al. 2011 Salyakina et al. 2011 Gai et al. 2012	21480499 22016809 21358714
4q22.1q22.2	<i>GRID2</i>	Schaaf et al. 2011 He et al. 2012	21624971 22678785				

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7p21.2	<i>AGMO</i>	Sebat et al. 2007	17363630	8q21.3	<i>MMP16</i>	Borg et al. 2002 Debost-Legrand et al. 2013	12070244 23239647
7p14.3	<i>PDE1C</i>	Vaags et al. 2012	22209245	8q22.1		Jain et al. 2010	20074678
7q11.23	<i>HIP1</i>	Edelmann et al. 2007 Ramocki et al. 2010 Girirajan et al. 2013	16971481 21109226 23375656	8q22.1	<i>SDC2</i>	Ishikawa-Brush et al. 1997	9259269
7q21.3	<i>DLX6</i>	Nakashima et al. 2010	19195802	8q23.3	<i>CSMD3</i>	Floris et al. 2008	18270536
7q22.1	<i>RELN</i>	Iossifov et al. 2012 Neale et al. 2012	22542183 22495311	8q24.11	<i>EXT1</i>	Li et al. 2002 Irie et al. 2012	12032595 22411800
7q31.1	<i>FOXP2/</i> Developmental verbal dyspraxia	Schaaf et al. 2011 Bacon & Rappold 2012 O’Roak et al. 2012	21624971 22736078 23160955	9p24.3	<i>SMARCA2/</i> Nicolaidis- Baraitser	Gana et al. 2011 Wolff et al. 2012	20802310 22822383
7q31.2	<i>ST7</i>	Vincent et al. 2000	10889047	9p24.3p24.2		Vinci et al. 2007 Pinto et al. 2010 Levy et al. 2011	17644778 20531469 21658582
7q31.2	<i>WNT2</i>	Wassink et al. 2001	11449391	9p13.2	<i>FBXO10</i>	O’Roak et al. 2012	22495309
7q31.32	<i>CADPS2</i>	Sadakata et al. 2007 Okamoto et al. 2011	17380209 21626674	9q21.2	<i>PRUNE2</i>	Vaags et al. 2012	22209245
7q32.2	<i>CEP41</i>	Korvatska et al. 2011	21438139	9q21.2	<i>GNA14</i>	Talkowski et al. 2012	22521361
7q35q36.1	<i>CNTNAP2</i>	Bakkaloglu et al. 2008 Poot et al. 2010 Rosenfeld et al. 2010	18179895 19582487 20808228	9q31.2q33.1		Gamerding et al. 2008	18386807
7q36.2	<i>DPP6</i>	Marshall et al. 2008 O’Roak et al. 2012	18252227 22495309	9q33.1	<i>ASTN2</i>	Glessner et al. 2009	19404257
8p23.3	<i>DLGAP2</i>	Marshall et al. 2008 Pinto et al. 2010	18252227 20531469	10p14		Verri et al. 2004 Lindstrand et al. 2010	15337474 20425828
8p23.3p23.1		Glancy et al. 2009 Ozgen et al. 2009 Chien et al. 2010 Nuvaro et al. 2011 van Daalen et al. 2011	18716609 19793310 20236125 21371014 21837366	10q11.23q21.2		Sebat et al. 2007	17363630
8p23.1	<i>PINX1</i>	Pinto et al. 2010	20531469	10q21.2	<i>ANK3</i>	Bi et al. 2012	22865819
8p22	<i>MSR1</i>	O’Roak et al. 2012	22495309	10q21.3	<i>JMJD1C, REEP3</i>	Castermans et al. 2007 Rosenfeld et al. 2010 Neale et al. 2012	17290275 20808228 22495311
8p22	<i>PSD3</i>	Pinto et al. 2010	20531469	10q21.3	<i>CTNNA3</i>	Levy et al. 2011 O’Roak et al. 2012 Girirajan et al. 2013	21658582 22495309 23375656
8p21.3p21.1		Ozgen et al. 2009	18696223	10q22.3	<i>KCNMA1</i>	Laumonnier et al. 2006 Neale et al. 2012	16946189 22495311
8q11.23	<i>RB1CC1</i>	Marshall et al. 2008	18252227	10q23.32	<i>BTA1F1, FGFBP3</i>	Salyakina et al. 2011	22016809
8q21.13	<i>FABP5</i>	Maekawa et al. 2010	20057506	10q25.2	<i>SMC3/Cornelia de Lange</i>	Deardorff et al. 2007 Moss et al. 2012	17273969 22490014
				10q25.3	<i>ATRNL1</i>	Stark et al. 2010	20670697

Loci with Limited Evidence for Involvement in Autism

CHROMOSOME BAND	GENE(S)/ SYNDROME	REFERENCES	HYPERLINK – Please click on PMID # below, or type http://www.ncbi.nlm.nih.gov/pubmed/ followed by the individual PMID #	CHROMOSOME BAND	GENE(S)/ SYNDROME	REFERENCES	HYPERLINK – Please click on PMID # below, or type http://www.ncbi.nlm.nih.gov/pubmed/ followed by the individual PMID #
10q26.2qter		Yatsenko et al. 2009	19558528	15q15.3	<i>CASC4</i>	Pinto et al. 2010	20531469
11p15.5	<i>SCT</i>	Yamagata et al. 2002	12160732	15q24.1	<i>UBL7</i>	Cukier et al. 2011 Salyakina et al. 2011	21480499 22016809
11p11.2	Potocki-Shaffer	Swarr et al. 2010	20140962	15q25.1	<i>ARNT2</i>	Vaags et al. 2012	22209245
11q13.1	<i>NRXN2</i>	Gauthier et al. 2011	21424692	15q25.2q25.3		Cooper et al. 2011	21841781
11q13.2	<i>KLC2</i>	Awadalla et al. 2010	20797689	16p13.3	<i>CACNA1H</i>	Splawski et al. 2006	16754686
11q13.2	<i>SUV420H1</i>	Iossifov et al. 2012 Sanders et al. 2012	22542183 22495306	16p13.3	<i>RBFOX1</i>	Martin et al. 2007 Sebat et al. 2007 Davis et al. 2012	17503474 17363630 22678932
11q14.2q14.3	<i>GRM5</i>	Iossifov et al. 2012 Kelleher et al. 2012	22542183 22558107	16p13.2	<i>USP7</i>	Celestino-Soper et al. 2011 Levy et al. 2011	21865298 21658582
11q22.1	<i>CNTN5</i>	van Daalen et al. 2011	21837366	16p12.3	<i>GPR139</i>	O'Roak et al. 2011 Sanders et al. 2012	21572417 22495306
11q23.3	<i>CADM1</i>	Zhiling et al. 2008	18957284	16q21	<i>CDH8</i>	Pagnamenta et al. 2011	20972252
11q24.2	<i>HEPACAM</i>	Lopez-Hernandez et al. 2011	21419380	16q22.1q22.3		Girirajan et al. 2013	23375656
11q24.2q24.3		Guerin et al. 2012	22965935	16q23.2q23.3	<i>CMIP</i>	Newbury et al. 2010 Celestino-Soper et al. 2011 Van der Aa et al. 2012	20193051 21865298 22689534
12q13.13	<i>SCN8A</i>	Veeramah et al. 2012	22365152	16q23.3		Matsunami et al. 2013	23341896
12q13.3q14.2		Lynch et al. 2011	21267005	16q24.2		Handrigan et al. 2013	23335808
12q14.3	<i>GRIP1</i>	Mejias et al. 2011	21383172	17q21.32	<i>ITGB3</i>	Weiss et al. 2006 O'Roak et al. 2012	16724005 22495309
13q31.3q32.1	<i>GPC6</i>	Pinto et al. 2010	20531469	17q22	<i>BZRAP1</i>	Bucan et al. 2009	19557195
14q23.2q23.3		Griswold et al. 2011	21360829	17q23.2	<i>TLK2</i>	O'Roak et al. 2011	21572417
14q32.2	<i>EML1</i>	Schaaf et al. 2011	21624971	17q25.3	<i>BAIAP2</i>	Celestino-Soper et al. 2011	21865298
14q32.3		Merritt et al. 2005	15657934	17q25.3	<i>SLC16A3</i>	Pinto et al. 2010	20531469
15q13.1	<i>APBA2</i>	Babatz et al. 2009	20029827	17q25.3	<i>CSNK1D</i>	Pinto et al. 2010	20531469
15q13.2	<i>ARHGAP11B</i>	Leblond et al. 2012	22346768	18q12		Gilling et al. 2008	18183041
15q13.3	<i>TRPM1</i>	Girirajan et al. 2013	23375656	18q21.1	<i>KATNAL2</i>	O'Roak et al. 2012 Sanders et al. 2012	22495309 22495306

Loci with Limited Evidence for Involvement in Autism

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18q21.1	<i>MBD1</i>	Li et al. 2005 Cukier et al. 2010	15967618 19921286	Xp11.23	<i>CACNA1F</i>	Hope et al. 2005 Myers et al. 2011	15807819 21383861
18q21.33	<i>PIGN</i>	Wisniewicka-Kowalnik et al. 2012	23032108	Xp11.23	<i>PPP1R3F</i>	Piton et al. 2011	20479760
18q22.3	<i>FBXO15</i>	Christian et al. 2008	18374305	Xp11.22	<i>IQSEC2</i>	Shoubridge et al. 2010	20473311
19p13.3	<i>ODF3L2</i>	Bucan et al. 2009	19557195	Xp11.22	<i>SMC1A</i> /Cornelia de Lange	Musio et al. 2006 Moss et al. 2012	16604071 22490014
19p13.3	<i>MBD3</i>	Cukier et al. 2010	19921286	Xq12	<i>OPHN1</i>	Celestino-Soper et al. 2011 Piton et al. 2011 Kaya et al. 2012	21865298 20479760 22213401
20p13	<i>TGM3</i>	O'Roak et al. 2011	21572417	Xq13.1	<i>DGAT2L6</i>	Lim et al. 2013	23352160
20p13		Sebat et al. 2007	17363630	Xq13.1	<i>NLGN3</i>	Jamain et al. 2003 Sanders et al. 2011 Kaya et al. 2012	12669065 21658581 22213401
20p12.3	<i>PLCB1</i>	Christian et al. 2008 Girirajan et al. 2013 Matsunami et al. 2013	18374305 23375656 23341896	Xq13.1	<i>CXCR3</i>	Piton et al. 2011 Chahrouh et al. 2012	20479760 22511880
20p12.1p11.23		Michaelis et al. 1997 Kamath et al. 2009	9268100 19058200	Xq13.1	<i>HDAC8</i> /Cornelia de Lange	Deardorff et al. 2012 Moss et al. 2012	22885700 22490014
20q11.22		Matsunami et al. 2013	23341896	Xq22.1	<i>HNRNP2</i>	Piton et al. 2011	20479760
20q12q13.11	<i>PTPRT</i>	Christian et al. 2008	18374305	Xq22.1		Grillo et al. 2010	20096387
20q13.33		Beri-Deixheimer et al. 2007 Traylor et al. 2010	17290276 20805988	Xq22.3	<i>PSMD10</i>	Piton et al. 2011	20479760
22q13.2	<i>EP300</i> Rubinstein-Taybi	Vaags et al. 2012	22209245	Xq23	<i>LUZP4</i>	Lim et al. 2013	23352160
Xp22.2	<i>FRMPD4</i>	Piton et al. 2011	20479760	Xq23	<i>AGTR2</i>	Vervoort et al. 2002 Takeshita et al. 2012	12089445 22269148
Xp22.2	<i>GLRA2</i>	Piton et al. 2011 Iossifov et al. 2012	20479760 22542183	Xq25	<i>GRIA3</i>	Wu et al. 2007	17989220
Xp22.2	<i>GRPR</i>	Ishikawa-Brush et al. 1997 Seidita et al. 2008	9259269 18393381	Xq25q26.1		Schroer et al. 2012	22965764
Xp22.2p22.13		Liao et al. 2011	20882036	Xq27.2	<i>MAGEC3</i>	Lim et al. 2013	23352160
Xp22.12	<i>SH3KBP1</i>	Piton et al. 2011	20479760	Xq28	<i>PDZD4</i>	Piton et al. 2011	20479760
Xp21.3	<i>ARX</i>	Stromme et al. 2002 Turner et al. 2002	12142061 12376946	Xq28	<i>HCFC1</i>	Gedeon et al. 1991 Huang et al. 2012	1870093 23000143
Xp11.23		Alesi et al. 2012	22634100	Xq28	<i>RPL10</i>	Klauck et al. 2006 Chiocchetti et al. 2011	16940977 21567917
Xp11.23	<i>FTSJ1</i>	Froyen et al. 2007	17333282	Xq28	<i>TMLHE</i>	Celestino-Soper et al. 2012 Nava et al. 2012	22566635 23092983

According to hg19 genome build

Loci Associated with Recessive Inheritance of Autistic Traits

The following table includes genes for which an autosomal recessive pattern of inheritance of autism susceptibility has been reported. A deletion or homozygous state would not by itself indicate autism susceptibility but would increase its likelihood. Sequence analysis of the relevant gene could be considered to evaluate this risk.

CHROMOSOME BAND	GENE(S)/ SYNDROME	REFERENCES	HYPERLINK – Please click on PMID # below, or type http://www.ncbi.nlm.nih.gov/pubmed/ followed by the individual PMID #	CHROMOSOME BAND	GENE(S)/ SYNDROME	REFERENCES	HYPERLINK – Please click on PMID # below, or type http://www.ncbi.nlm.nih.gov/pubmed/ followed by the individual PMID #
1p34.1	<i>POMGNT1</i> Muscle-eye-brain disease	Yu et al. 2013	23352163	11q13.4	<i>DHCR7</i> /Smith-Lemli-Opitz	Tierney et al. 2001 Sikora et al. 2006	11223857 16761297
1p21.3	<i>DPYD</i> /DPYD deficiency	Wadman et al. 1984 Carter et al. 2011 Willemsen et al. 2011	6720361 21114665 22003227	12p13.33	<i>CACNA2D4</i>	Lewy et al. 2011	21658582
1q43	<i>RGS7</i>	Pinto et al. 2010 Najmabadi et al. 2011	20531469 21937992	12q13.12	<i>NCKAP5L</i>	Chahrouh et al. 2012	22511880
2q35	<i>PECR</i>	Najmabadi et al. 2011	21937992	12q24.11	<i>UBE3B</i>	Chahrouh et al. 2012	22511880
3p25.1	<i>XPC</i> /Xeroderma pigmentosum	Khan et al. 1998 Quackenbush et al. 1999	9804340 10604143	12q24.33	<i>EP400</i>	Chahrouh et al. 2012 Iossifov et al. 2012	22511880 22542183
3p22.2	<i>SLC22A14</i>	Lim et al. 2013	23352160	13q21.2	<i>DIAPH3</i>	Vorstman et al. 2011	20308993
3p21.31	<i>AMT</i> /Nonketotic hyperglycinemia	Yu et al. 2013	23352163	14q32.12	<i>UBR7</i>	Najmabadi et al. 2011	21937992
4q28.3	<i>PCDH10</i>	Morrow et al. 2008	18621663	15q21.1	<i>GATM</i> /GATM deficiency	Battini et al. 2002	12468279
5p15.31	<i>NSUN2</i>	Martinez et al. 2012	22577224	16p11.2	<i>BCKDK</i> /BCKDK deficiency	Novarino et al. 2012	22956686
6p22.3	<i>ALDH5A1</i> /SSADH deficiency	Pearl et al. 2003	12743223	17p12	<i>ZNF18</i>	Chahrouh et al. 2012	22511880
6q23.3	<i>PEX7</i> /Rhizomelic chondrodysplasia punctata	Yu et al. 2013	23352163	19p13.3	<i>GAMT</i> /GAMT deficiency	Lion-Francois et al. 2006 Nasrallah et al. 2012	17101918 22783571
6q25.1q25.2	<i>SYNE1</i>	O'Roak et al. 2011 Yu et al. 2013	21572417 23352163	22q11.21	<i>PRODH1</i> Hyperprolinemia	Afenjar et al. 2007 Di Rosa et al. 2008	17412540 18197084
8q22.2	<i>VPS13B</i> /Cohen	Kolehmainen et al. 2003 Howlin et al. 2005	12730828 15793684	22q11.21	<i>CLTCL1</i>	Chahrouh et al. 2012	22511880
10q22.2	<i>ADK</i>	Najmabadi et al. 2011	21937992	22q13.1	<i>ADSL</i> /ADSL deficiency	Stone et al. 1992 Sivendran et al. 2004 Perez-Duenas et al. 2012	1302001 15471876 21903433

According to hg19 genome build