

**Evidence that autosomal recessive spastic cerebral palsy-1
(CPSQ1) is caused by a missense variant in HPDL**

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4 **Evidence that autosomal recessive spastic cerebral palsy-1 (CPSQ1) is caused by a missense variant**
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6 **in *HPDL***
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Abstract

A subset of individuals diagnosed with cerebral palsy will have an underlying genetic diagnosis. Previously a missense variant in *GAD1* was described as a candidate mutation in a single family diagnosed with autosomal recessive spastic cerebral palsy-1 (CPSQ1; OMIM 603513). Following the ascertainment of a further branch of the CPSQ1 kindred, we found that the previously reported *GAD1* variant did not segregate with the neurological disease phenotype in the recently ascertained branch of the kindred. Following genetic linkage studies to map autozygous regions and whole exome sequencing a missense variant (c.527 T>C; p.Leu176Pro, rs773333490) in the *HPDL* gene was detected and found to segregate with disease status in both branches of the kindred. *HPDL* encodes a 371 amino acid protein, 4-Hydroxyphenylpyruvate Dioxygenase Like, that localises to mitochondria but whose function is uncertain. Recently biallelic loss of function variants and missense substitution-causing variants in *HPDL* were reported to cause a childhood onset progressive spastic movement disorder with a variable presentation. These findings suggest that *HPDL*-related neurological disease may mimic spastic cerebral palsy and that *GAD1* should not be included in diagnostic gene panels for inherited cerebral palsy.

Abbreviated Summary:

Morgan et al report that a novel candidate mutation in the *HPDL* gene (but not *GAD1*) segregates with disease status within a kindred in which a locus for autosomal recessive spastic cerebral palsy-1 (CPSQ1; OMIM 603513, *GAD1*) was mapped previously (in part of the kindred).

Introduction

Cerebral palsy is a common cause of childhood neurodisability that is characterised clinically by permanent abnormalities of motor activity and posture. It typically includes spasticity and often intellectual disability and epilepsy, and results from non-progressive disturbances to the fetal or infantile brain (MacLennan *et al.*, 2019; Pearson *et al.*, 2019). In most cases initial symptoms occur in infancy and the diagnosis of cerebral palsy is made by age two years (Pearson *et al.*, 2019). Cerebral palsy is an umbrella term covering a heterogeneous group of conditions (MacLennan *et al.*, 2019). Traditionally cerebral palsy was often attributed to acute hypoxia at birth but this now thought to account for only a minority of cases. Instead, prenatal insults are a more prominent cause, although in 25-50% of cases no cause is identified (Michael-Asalu *et al.*, 2019). However, there has been increasing realisation that inherited disorders can present as cerebral palsy. Both copy number abnormalities and pathogenic variants in single genes have been described as causes of a cerebral palsy phenotype and inherited causes of apparent cerebral palsy may be transmitted in an autosomal dominant, autosomal recessive, X-linked or mitochondrial inheritance fashion (McMichael *et al.*, 2015; Oskoui *et al.*, 2015; Takezawa *et al.*, 2018; Michael-Asalu *et al.*, 2019). Autosomal recessively inherited disorders are a common cause of familial cerebral palsy and consanguinity has been identified as a risk factor for cerebral palsy (Erkin *et al.*, 2008; Moreno de Luca *et al.*, 2012).

Autosomal recessively inherited disorders are an important cause of mortality and morbidity, particularly in communities with parental consanguinity and/or common founder mutations. In the UK, the overall frequency of consanguinity is <1% but is much higher in some communities (Bundey *et al.*, 1991; Sheridan *et al.*, 2013). In one UK centre, consanguinity was associated with an increased risk of perinatal mortality from lethal malformations attributed to autosomal recessive disorders and in another centre parental-relatedness was associated with a ~2.2-fold increased risk for congenital anomaly (Bundey *et al.*, 1991; Sheridan *et al.*, 2013). For families in which the pathogenic alleles causing a recessive disorder are characterised, carrier parents can be provided with counselling regarding their reproductive options and their at risk relatives can be tested to determine their carrier risk. Strategies for preconception genetic testing have been developed to offer prospective parents the opportunity to determine their carrier status for autosomal recessive disease mutations (Bell *et al.*, 2011, Capalbo *et al.*, 2019).

The availability of genetic testing for recessive disorders is based on the reliable identification of the relevant disease gene. Lander and Botstein (1987) proposed that mapping regions of homozygosity by descent (autozygosity) in the affected children of consanguineous marriages could provide a

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3 highly efficient strategy for mapping recessive traits. With improvements in genetic maps and
4 identification of polymorphic DNA markers the strategy of homozygosity/autozygosity mapping was
5 widely adopted to localise recessive genes even in the presence of locus heterogeneity (Mueller and
6 Bishop, 1993; Sheffield *et al.*, 1995). Though for a child whose parents are first cousins, ~6% (1/16) of
7 the genome should be homozygous/autozygous, in communities in which consanguinity has been
8 practiced over multiple generations, more extensive autozygosity often occurs. In consanguineous
9 families with an autosomal recessive disorder, Woods *et al.*, (2006) reported that children with first
10 cousin parents had an average of 11% homozygosity with 20 homozygous segments >3 cM.
11 Additionally, the chance of the longest segment of homozygosity harbouring the disease gene was
12 estimated at ~1 in 6 (Woods *et al.*, 2006). In older autozygosity mapping studies, candidate genes in
13 an autozygous region would be sequenced serially until a candidate pathogenic variant was
14 identified (Hartley *et al.*, 2010). However, the advent of exome sequencing (Bolze *et al.*, 2010; Dang
15 *et al.*, 2016) enabled simultaneous sequencing of all genes in the linked region and interrogation of
16 genes in smaller autozygous segments. This is helpful because many autosomal recessive disease
17 families with parental consanguinity that are recruited to research studies will be from non-white
18 ethnicities and therefore will be less represented in global databases of genetic variation, making the
19 interpretation of the pathogenicity of rare genetic variants in such cases challenging (Manolio,
20 2019).

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34 Over twenty years ago, Mitchell and Bunday (1997) described seven families with parental
35 consanguinity in which children presented with a spastic cerebral palsy-like syndrome. There was
36 clinical heterogeneity between the families (e.g. variable degrees of learning disability and
37 microcephaly in some cases) suggesting different underlying diagnoses but the presence of
38 symmetrical lower limb spasticity was noted in all cases. The authors concluded that the symmetry
39 was consistent with an underlying, probably autosomal recessively inherited, genetic origin (Mitchell
40 and Bunday, 1997). Subsequently, genetic linkage studies were reported in a some of these families
41 using 290 polymorphic DNA markers and a common region of autozygosity was identified at
42 chromosome 2q24-q25 (~5 cM interval) in three families, suggesting the presence of a locus for
43 autosomal recessive symmetrical spastic cerebral palsy (McHale DP *et al.*, 1999). One of the families
44 studied (Family 4 in Mitchell and Bunday (1997) and McHale DP *et al.*, 1999) contained four affected
45 children and further investigations identified a rare homozygous missense substitution (c. 36G>C;
46 p.Ser12Cys) in the *GAD1* gene (Lynex *et al.*, 2004). This suggested that autosomal recessive spastic
47 cerebral palsy-1 (CPSQ1; OMIM 603513) resulted from mutation in the glutamate decarboxylate-1
48 gene (*GAD1*; OMIM 605363). Here we provide further genetic evidence to demonstrate that CPSQ1
49 is not caused by *GAD1* but by a missense substitution in a recently described gene, *HPDL*.

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For Review Only

METHODS

Patients

Molecular genetic studies were undertaken in 15 individuals from a large consanguineous British Pakistani kindred that was initially ascertained as two distinct families but later established to be part of a larger kindred. One of the families (Branch A) was originally described in the clinical report by Mitchell and Bunday (1997) (referred to as Family 4) and in molecular genetic studies reported by McHale DP et al., (1999) (Family 4) and Lynex et al., (2004) (Family B). Subsequently a family with a similar phenotype (herein referred to as Branch B) was identified independently and then shown to have shared ancestors with Branch A (see Figure 1). All individuals gave written informed consent. The study was approved by the South Birmingham Research Ethics Committee and performed in accordance with the ethical standards laid down in the 1964 Declaration of Helsinki. Genomic DNA was extracted from the buffy coat of all individuals where possible.

Molecular genetic analysis

Linkage analysis

Linkage to the chromosome 2 and *GAD1* candidate region reported in Lynex et al., (2004) was further investigated here in 15 individuals (see supplementary figure 1) from branches A and B of the kindred using microsatellite markers in the previously identified candidate region at chromosome 2q24-q31 and Sanger sequencing of the *GAD1* missense substitution was performed. Subsequently a genome-wide linkage scan was carried out using Affymetrix Genome-Wide Human SNP 5.0 microarrays in a total of 6 affected individuals (5 affected individuals from Branch A (V:1, V:2, V:5, V:7, V:8) and 1 affected individual from Branch B (V:10)). Homozygosity mapping analysis was then performed and homozygous regions were identified using HomozygosityMapper (<http://homozygositymapper.org>) (Seelow et al., 2009).

Exome Sequencing

Exome sequencing was performed in two stages. Initially Patient V:5 was subjected to whole exome sequencing using the SureSelect human All Exon 50Mb kit (Agilent Technologies) and sequencing on the HiSeq 2500 (Illumina) with 100 bp paired-end reads. The sequences were aligned to the reference genome (GRCh37), with Novoalign (Novocraft Technologies Sdn Bhd). Duplicate reads, resulting from PCR clonality or optical duplicates, and reads mapping to multiple locations were excluded from downstream analysis. Depth and breadth of sequence coverage was calculated with custom scripts and the BedTools package. Single nucleotide variants (SNVs) and small insertions and deletions

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3 (InDels) were identified and quality filtered within the SamTools software package and in-house
4 software tools. All calls with a read coverage <4 and a phred-scaled SNP quality of <20 were filtered
5 out. Variants were annotated with respect to genes and transcripts with the Annovar tool.
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7 Subsequently exome sequencing was performed in 5 individuals (IV:3; IV:4; V:10; V:11; V:12) using
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9 Illumina Nextera assays. The samples were sequenced on Illumina's HiSeq 4000 platform with 150bp
10 paired end reads. Raw Illumina BCL files were demultiplexed and converted to fastq format using
11
12 Illumina's bcl2fastq 2.19, which also trimmed from the reads the indexes and the adaptors used for
13
14 sequencing. All sample pairs were aligned to the hg38 version of the reference human genome using
15
16 bwa 0.7.15 in alt contig aware mode as described by the authors (Li and Durbin, 2009). Further details
17
18 of the bioinformatic pipeline are detailed in supplementary methods.
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20 21 22 **Sanger Sequencing**

23 Amplification of genomic DNA for Sanger sequencing for 3 individuals (IV:2; V:1; V:5) was performed
24 by standard PCR methods. PCR clean-up was performed with MicroClean™. Sanger sequencing was
25 then performed using the same primers, primer sequences are available on request. Sanger
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27 sequencing was performed using BigDye Terminator Cycle Sequencing Kit, version 3.1 (Applied
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29 Biosystems) and analysed on an ABI 3130XL DNA analyser (Applied Biosystems).
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RESULTS

Clinical features

The kindred pedigree is shown in figure 1. The sibship labelled Branch A was described previously (Mitchell and Bunday, 1997; McHale DP *et al.*, 1999; Lynex *et al.*, 2004). Subsequently the sibship labelled Branch B was ascertained independently and found to have a common ancestor with Branch A (see Figure 1). The clinical details of the affected individuals from both branches are summarised in Table 1. The details for four affected individuals (V:1; V:2; V:5; V:7) in Branch A were described previously (Mitchell and Bunday, 1997; McHale DP *et al.*, 1999; Lynex *et al.*, 2004) but since the last report (Lynex *et al.*, 2004) a further younger male sibling (V:8) had been diagnosed with spastic cerebral palsy. Branch A included two sets of dizygotic twins with one affected twin (both male) in each case. Branch B comprised four siblings. At the time of ascertainment, two of the siblings were clinically affected (V:10; V:11), one sibling was asymptomatic and judged to be unaffected (V:12) and the clinical status of the youngest sibling had not been established (V:13). After clinical assessment of V:10 and V:11 and review of the clinical descriptions of affected individuals in Family A the working diagnosis was that both branches of the family were affected by the same neurological disorder.

Molecular Genetic Studies

Previous linkage studies and sequencing analysis in Branch A (McHale DP *et al.*, 1999; Lynex *et al.*, 2004) had identified a common region of homozygosity at chromosome 2q24-q25 in four affected individuals and a rare homozygous missense substitution within *GAD1* as a candidate mutation (c.36G>C; p.Ser12Cys). Following the ascertainment of Branch B, we performed extended segregation analysis using microsatellite markers mapping to 2q24-q31 and genotyped the c.36G>C variant across both branches. This confirmed the finding that c.36G>C variant segregated with disease status in Branch A (including the newly diagnosed sibling V:8) but that in Branch B two affected individuals (V:10 and V:11) were either heterozygous (V:10) or homozygous wild-type (V:11) and the father (iV:3) was homozygous wild-type (see supplementary Figure 1). Unless the autosomal recessive neurological disorder diagnosed in Branch A and Branch B were different, this excluded *GAD1* as the cause of the disease in this kindred.

To investigate the evidence of a single genetic cause for the autosomal recessive spastic cerebral palsy segregating in Branches A and B, we performed autozygosity mapping using Affymetrix SNP gene arrays in 6 affected individuals (V:1, V:2, V:5, V:7, V:8) from Branch A and V:10 from Branch B). Analysis using homozygosityMapper revealed 2 areas of extended homozygosity shared between all

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3 affected individuals. A 551,761 bp region on chromosome 10 between SNPs rs2036919
4 (chr10:23,636,106, GRCh37) and rs11013778 (chr10:24,187,867, GRCh37) and a much larger
5 17,596,867 bp region on chromosome 1 between SNP markers rs1046988 (chr1:40,219,065,
6 GRCh37) and rs6687842 (chr1:57,815,932, GRCh37) (Figure 2A).
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10 To identify a causative pathogenic variant we proceeded with exome sequencing in one affected
11 individual (V:5) from Branch A. Exome sequencing revealed 23,469 variations, of which 9,869 were
12 novel or rare (<0.01) sequence changes. At the time of initial analysis (May 2011;
13 <https://etheses.bham.ac.uk/id/eprint/3068/>) comparisons with dbSNP build 134, the 1000
14 Genomes Project database ([The 1000 Genomes Project Consortium](http://evs.gs.washington.edu/EVS/)), Exome Variant Server (ESV)
15 (<http://evs.gs.washington.edu/EVS/>) and our in-house dataset , identified 61 homozygous variants
16 in the affected individual (Figure 2B). Of these one homozygous non-synonymous missense variant
17 (c.527 T>C; p.Leu176Pro, rs773333490) mapped to one of the identified regions of extended
18 homozygosity on chromosome 1p34.1 in the *HPDL* gene (Figure 2B). To verify this candidate
19 mutation, Sanger sequencing/exome sequencing was performed on the DNA of three available
20 members of the family from Branch A (IV:2; V:1; V:5) and was found to segregate with the disease
21 (Figure 2C and supplementary Table 1). The unaffected parent (IV:2) was heterozygous for the
22 variant (T/C) and the two affected individuals (V:1 and V:5) were homozygous for the mutant allele
23 (C/C). Interrogation of genomic databases at the time of the initial exome sequencing (May 2011)
24 did not identify homozygous individuals in any control data sets (see above). Subsequently, further
25 exome sequencing undertaken in 2 affected individuals from Branch B (V:10 and V:11) and 3
26 unaffected family members (IV:3, IV:4 and V:12) demonstrated that V:10 and V:11 were homozygous
27 for the mutant allele and the unaffected parents and sibling were heterozygous for the mutant allele
28 either directly (IV:4), or inferred from flanking SNPs (IV:3 and V:12). Finally three affected individuals
29 (V:2; V:7; V:8) from Branch A in whom DNA was not available for sequencing were inferred (from
30 flanking homozygous SNPs) to be homozygous for the mutant allele (C/C) from the SNP array data
31 (Supplementary Table 1 and Supplementary Table 3). Overall these findings were consistent with the
32 candidate pathogenic *HPDL* variant segregating with the disease in both branches of the family.
33 Subsequent to the initial exome sequencing analysis and identification of the *HPDL* variant ((c.527
34 T>C; p.Leu176Pro, rs773333490), independent analysis of the exome sequencing results for two
35 affected individuals (V:10 and V:11) in Branch B also prioritised this variant as the best candidate
36 mutation within the autozygous segment.
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40 We inferred identity by descent (IBD) to estimate familial relationships and calculated pairwise
41 PIHAT between six family members who underwent exome sequencing (see Supplementary
42 Methods). This confirmed parental relatedness between IV:3 and IV:4 (PIHAT 0.15) and that V:5 from
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3 Branch A was closely related to the two affected individuals in Branch B (V:10 and V:11; PIHAT 0.28
4 and 0.26 respectively).

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6 *In silico* variant pathogenicity prediction tools categorised the missense substitution as
7 possibly_damaging in Polyphen (<http://genetics.bwh.harvard.edu/pph2/>) and deleterious in SIFT
8 (<https://sift.bii.a-star.edu.sg>). The affected Leucine residue (ancestral T allele) is conserved across
9 multiple species (Figure 2D). The variant was heterozygous in one individual of South Asian origin in
10 gnomAD (<https://gnomad.broadinstitute.org> giving an allele frequency of 1 in 244,766, and never
11 reported in a homozygous state).. Variant classification according to ACMG criteria and assigning
12 moderate evidence for pathogenicity for the results of segregation of the c.527 T>C; p.Leu176Pro
13 substitution in both branches of the family resulted in a likely pathogenic classification for this
14 variant (Evidence for pathogenicity PM1 PM2 PP1; Intervar, Li and Wang, 2017)

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21 Data Availability: The authors confirm that all the data supporting the findings of this study are
22 available within the article and readily available upon request.
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DISCUSSION

The identification of a genetic cause for cerebral palsy has important implications enabling clarification of the recurrence risk and risks to relatives, provides parents with an explanation for their child's disorder and may occasionally identify a treatable condition (Moreno de Luca *et al.*, 2012). Genetic causes of cerebral palsy are extremely heterogeneous and may overlap with those from other neurological disorders. Clinically cerebral palsy has been subdivided into subtypes including spastic, athetoid, ataxic and mixed types but the clinical diagnosis of spastic cerebral palsy and hereditary spastic paraparesis (HSP) or of ataxic cerebral palsy and hereditary cerebellar ataxias (HCA) may be blurred and both HSP and HCA are highly genetically heterogeneous (Fahey *et al.*, 2017, Parodi *et al.*, 2018). The key clinical distinction between spastic diplegic cerebral palsy and HSP is the progressive nature of the latter, but this may not be obvious without long-term follow up and there are previous examples of a molecular diagnosis of HSP being made in children who were initially labelled as having cerebral palsy (Ranier *et al.*, 2006).

Previously a homozygous *GAD1* missense substitution (c. 36G>C; p.Ser12Cys) was identified in four affected siblings with symmetrical spastic cerebral palsy (Lynex *et al.*, 2004). At that time the variant was not present in 100 control individuals. Recently (accessed August 2020) this variant was found to have a frequency of 6 in 251302 alleles (allele frequency = 0.00002388) in gnomAD with no homozygotes reported (in South Asians the allele frequency was 0.0001633 (5 in 30,616)). However, the *GAD1* variant did not segregate with disease status in Branch B of the large consanguineous kindred in which the *GAD1* variant was identified in Branch A. Therefore, unless there were two separate genetic diagnoses in the kindred, *GAD1* was not the cause of the autosomal recessively inherited spastic cerebral palsy disorder within the kindred. To date, though *GAD1* mutation has been widely cited as a cause of autosomal recessively inherited cerebral palsy (Lynex *et al.*, 2004,), this kindred is the only reported example of CPSQ1 (OMIM 603513).

To search for other candidate pathogenic variants we undertook whole exome sequencing and identified a SNV leading to a missense substitution (c.527T>C, p.Leu176Pro) in *HPDL*, which encodes a 371 amino acid protein of unknown function. When we initially detected this variant the *HPDL* gene had not been linked to human disease but recently, Husain *et al.*, (2020) described biallelic variants in *HPDL* in a childhood onset progressive spastic movement disorder with a variable presentation. They reported 13 families containing 17 affected individuals (5 kindreds with parental consanguinity) with biallelic *HPDL* mutations (loss of function variants in 5 individuals from 4 kindreds). The rare c.527T>C, p.Leu176Pro variant detected in both branches of the family reported here has not been reported previously but other candidate pathogenic missense variants described

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3 by Husain et al., (2020) were p.Gly50Asp (detected in 3 kindreds); p.Trp157Arg; p.Leu217Pro;
4 p.Gly250Glu; p.His251Gln; p.Ile266Thr and p.Tyr287His. *HPDL* encodes the 4-Hydroxyphenylpyruvate
5 Dioxygenase Like protein that does not currently have a known function. *HPDL* transcript levels are
6 high in the central and peripheral nervous system, a mitochondrial localisation signal was predicted
7 in the first 37 amino acids and exogenous *HPDL* was found to co-localise with mitochondria (Husain
8 et al., 2020). However fibroblasts and muscle from affected individuals did not show clear evidence
9 of aberrant mitochondrial function or morphology (Husain et al., 2020), thus further evidence is
10 required before *HPDL* can be unequivocally placed in the group of HSP genes that affect
11 mitochondrial function (Hensiek et al., 2015).
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19 Though we report a single kindred, the large number of affected individuals (n=7) adds significantly
20 to the number of subjects with *HPDL*-associated disease currently in the literature (n=17). In the
21 original descriptions of affected children in Branch A, Bunday and Mitchell (1997) described how,
22 after a normal pregnancy, the four affected individuals showed signs in the first year of life and all
23 had a non-progressive symmetrical paraparesis (upper limbs affected in some cases). A degree of
24 learning disability was present in all cases (severe in two) but upper limb ataxia was variably present.
25 By the third decade, affected individuals were wheelchair-dependent with fixed flexion deformities.
26 The additional individuals (V:8 from Branch A and V:10 and V:11 from Branch B) who were
27 diagnosed since Lynex et al., (2004) generally showed similar features though prenatal growth
28 retardation was present in one individual. The affected individuals had not been extensively
29 investigated but, when performed, blood lactate levels were normal and brain MRI scans in three
30 individuals were mostly unremarkable though one scan was reported to show demyelination in the
31 medulla. In the series of patients (n=17) reported by Husein et al., (2020) the clinical phenotype was
32 more variable ranging from a severe spastic neurological disorder to uncomplicated hereditary
33 spastic paraparesis. Features present in more than a half of patients included progression of
34 neurological signs, intellectual disability, developmental motor delay, microcephaly and strabismus
35 (which were present in our series except for microcephaly). A subgroup of their cohort presented in
36 the neonatal period with severe disease and acute respiratory failure. In many cases blood lactate
37 levels were increased (at the time of neurological deterioration but not at routine follow ups) and
38 MRI scans showed deficiency in myelination in most cases (present in one of our series). Thus the
39 phenotype of autosomal recessive *HPDL*-related neurological disorder is variable and ranges from a
40 severe congenital-onset disorder without neurological development to a milder later-onset
41 hereditary spastic paraparesis phenotype, and the clinical characteristics of the affected individuals
42 in Branch A and Branch B would appear to fall within this spectrum. Though the *GAD1* (c. 36G>C;
43 p.Ser12Cys) detected previously in Branch A could not account for the disease phenotype in both
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3 branches of the kindred, we note that mice homozygous null for *Gad1* die at birth with cleft palate
4 and hypoxia, with omphalocele in about 50% (Asada et al 1997; Condie et al 1997; Saito et al 2010).
5 Furthermore, Chatron et al (2020) have recently reported that biallelic pathogenic (mostly loss of
6 function) variants in *GAD1* were associated with a phenotype of early onset epileptic
7 encephalopathy in 11 individuals from six independent kindreds. Additional, variable,
8 clinical features included cleft palate (64%), omphalocele (18%) and joint contractures
9 (55%). Though it is possible that *GAD1* variant status might modify clinical presentation within the
10 two branches of the family described here, we note that these clinical features, including, epilepsy
11 were not reported within the family.
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19 In summary we propose that the evidence for an autosomal recessive spastic cerebral palsy-1
20 (CPSQ1; OMIM 603513) locus at 2q31.1 resulting from a mutation in the *GAD1* gene is insecure and
21 that the single family reported to have a *GAD1*-related neurological disorder are most likely to have
22 a *HPDL*-related autosomal recessive disorder. Our findings support the contention that biallelic
23 mutations in *HPDL* can present with a variety of clinical phenotypes and the neurological phenotype
24 is progressive. They also underscore the need to consider a genetic etiology in patients labelled as
25 having cerebral palsy.
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Figure Legends:

Figure 1. Family structure showing how two branches of the kindred were found to be connected (Branch A previously reported and Branch B subsequently ascertained (see text for details).

Figure 2: Mapping and Sequence identification of a missense substitution in *HPDL*. (A) Genomewide SNP arrays performed in 6 affected individuals from family branch A and B, shows areas of extended homozygosity that are shared between all affected individuals and highlighted as red peaks on chromosome 1 (containing *HPDL*) and a smaller region on chromosome 10. (B) Initial whole Exome sequencing analysis in patient V:5 revealed 41 candidate homozygous rare and novel sequence variants through filtering steps and only 1 of these variants mapped to the chromosome 1 candidate region (c.527T>C, p.Leu176Pro in *HPDL*). (C) Sanger sequencing traces showing the *HPDL* missense substitution. Upper panel shows mother of branch A (IV:2) in a heterozygous state (T/C) and lower panel the affected individual V:1 in a homozygous state (C/C). (D) A comparison of sequence alignments of *HPDL* in 11 species showing the conservation of the p.Leu176 amino acid position where the variant p.Leu176Pro occurs.

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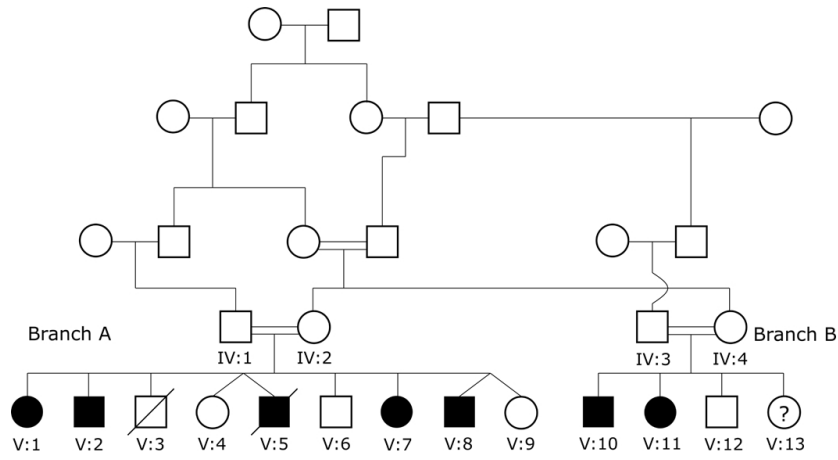


Figure 1: Family structure showing how two branches of the kindred were found to be connected (Branch A previously reported and Branch B subsequently ascertained (see text for details).

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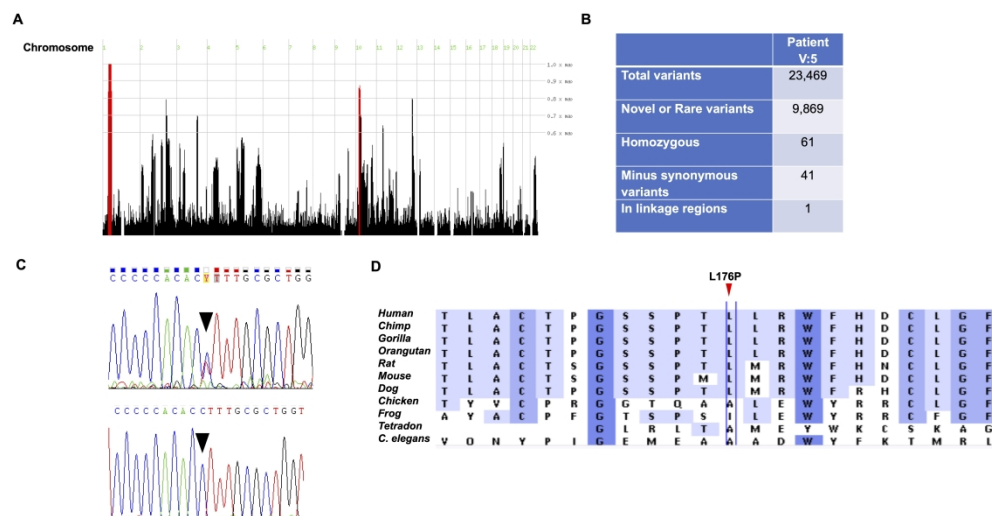
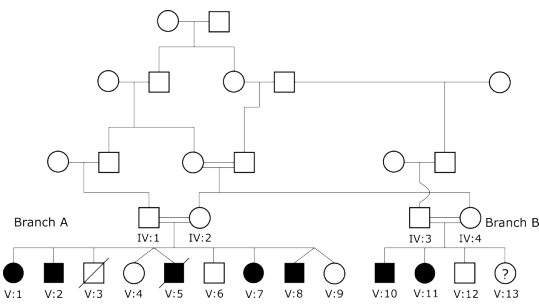


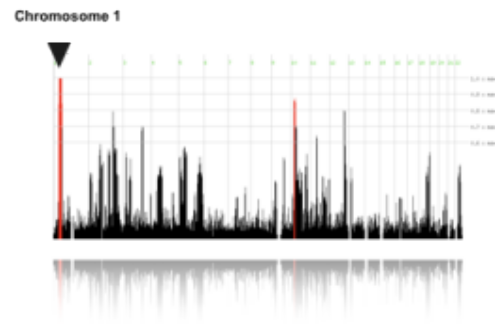
Figure 2: Mapping and Sequence identification of a missense substitution in HPDL. (A) Genomewide SNP arrays performed in 6 affected individuals from family branch A and B, shows areas of extended homozygosity that are shared between all affected individuals and highlighted as red peaks on chromosome 1 (containing HPDL) and a smaller region on chromosome 10. (B) Initial whole Exome sequencing analysis in patient V:5 revealed 41 candidate homozygous rare and novel sequence variants through filtering steps and only 1 of these variants mapped to the chromosome 1 candidate region (c.527T>C, p.Leu176Pro in HPDL). (C) Sanger sequencing traces showing the HPDL missense substitution. Upper panel shows mother of branch A (IV:2) in a heterozygous state (T/C) and lower panel the affected individual V:1 in a homozygous state (C/C). (D) A comparison of sequence alignments of HPDL in 11 species showing the conservation of the p.Leu176 amino acid position where the variant p.Leu176Pro occurs.

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autosomal recessive cerebral spastic palsy-1 (CPSQ1)



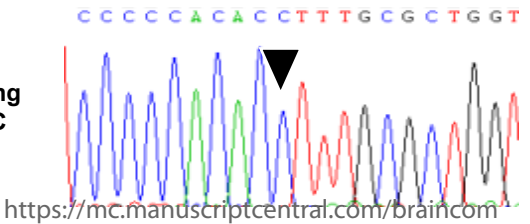
SNP array
Genomewide scan



exome sequencing
- filtering analysis

	Patient V:5
Total variants	23,469
Novel or Rare variants	9,869
Homozygous	61
- synonymous variants	41
In linkage regions	1 variant (<i>HPDL</i>) chromosome 1

Sanger sequencing
HPDL: c.527 T>C
p.Leu176Pro



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Supplementary Material

Evidence that autosomal recessive spastic cerebral palsy-1 (CPSQ1) is caused by a missense variant in *HPDL*

Neil V Morgan (1), Bryndis Yngvadottir (2), Mary O'Driscoll (3), Graeme R Clark (2), Diana Walsh (4), Ezequiel Martin (2,5), Louise Tee (6), Evan A Reid (2, 7), Hannah V Titheradge (3), Eamonn R Maher (2)

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Supplementary Methods

Exome Sequencing and Bioinformatics Analysis for Branch B participants

Exome sequencing was performed in 5 individuals (IV:3; IV:4; V:10; V:11; V:12), using the Nextera Rapid Capture Enrichment assay for V:10 and V:11 and the Nextera DNA Exome Enrichment assay for V:3, V:4 and V:12. The samples were sequenced on Illumina's HiSeq 4000 platform with 150bp paired end reads. Raw Illumina BCL files were demultiplexed and converted to fastq format using Illumina's bcl2fastq 2.19, which also trimmed from the reads the indexes and the adaptors used for sequencing. All sample pairs were aligned to the hg38 version of the reference human genome using bwa 0.7.15 in alt contig aware mode as described by the authors (Li and Durbin, 2009). The generated SAM file was compressed into a BAM file and sorted by genomic position using samtools 1.3.1 (Li *et al.*, 2009). The sorted BAM files were subject to Base Quality Score Recalibration and Indel Realignment followed by variant calling using the Haplotype Caller algorithm as specified in the Genome-Analysis Toolkit (GATK version 3.8 (McKenna *et al.*, 2010)) best practices (DePristo *et al.*, 2001; Auwera *et al.*, 2014). The only default parameter from best practices that was altered was the threshold for mapping quality (mapQ) during variant calling which was set to 30. The resulting VCF files were filtered for a minimum depth of 20 reads and a Genotype Quality (GQ) of 30 using vcftools 0.1.15 (Danecek *et al.*, 2011). VCF files were then annotated with ANNOVAR (Wang *et al.*, 2010) using all default databases available for hg38.

Variants were then filtered based on the following exclusion criteria. A rare variant cut off or minor allele frequency (MAF) of <0.01 in each dataset was used and synonymous and intron variants +/- 5 base pairs away from the exon-intron boundaries were excluded. Non-shared variants between the same affected family members were also eliminated. Following exclusion of variants based on these criteria, *in silico* pathogenicity prediction tools were employed for further analysis.

Identity by descent (IBD)

We used PLINK (version 1.90, <http://pngu.mgh.harvard.edu/purcell/plink/>, Purcell *et al.* (2007)) to infer familial relationships (IBD) by calculating pairwise PIHAT values between all individuals in Branch B (IV:3, IV:4, V:10, V:11 and V:12) and one individual in Branch A (V:5). To this end we filtered out variants with missing call rates above 0.01, as well variants which have a Hardy-Weinberg equilibrium exact test p-value below 0.000005 and variants with minor allele frequency below 0.05.

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Supplementary Table 1: Summary of genotyping results for HPDL variant (c.527 T>C; p.Leu176Pro, rs773333490) in family members in who sufficient DNA for testing was available

	Branch A							Branch B				
	Father	Mother	Child 1	Child 2	Child 5	Child 7	Child 8	Father	Mother	Child 1	Child 2	Child 3
Individual ID	IV:1	IV:2	V:1	V:2	V:5	V:7	V:8	IV:3	IV:4	V:10	V:11	V:12
Clinical Status	Unaffected	Unaffected	Affected	Affected	Affected	Affected	Affected	Unaffected	Unaffected	Affected	Affected	Unaffected
SNP Array results ¹			Predicted homozygous for <i>HPDL</i> c.527 T>C by SNP array data	Predicted homozygous for <i>HPDL</i> c.527 T>C by SNP array data	Predicted homozygous for <i>HPDL</i> c.527 T>C by SNP array data	Predicted homozygous for <i>HPDL</i> c.527 T>C by SNP array data	Predicted homozygous for <i>HPDL</i> c.527 T>C by SNP array data			Predicted homozygous for <i>HPDL</i> c.527 T>C by SNP array data		
Exome sequencing ²					Homozygous for <i>HPDL</i> c.527 T>C by exome sequencing			Heterozygous by SNPs flanking the <i>HPDL</i> gene by exome sequencing (poor coverage of the <i>HPDL</i> c.527 T>C variant)	Heterozygous <i>HPDL</i> c.527 T>C by exome sequencing	Homozygous <i>HPDL</i> c.527 T>C by exome sequencing; Predicted homozygous by SNP array data	Homozygous <i>HPDL</i> c.527 T>C by exome sequencing; Predicted homozygous by SNP array data	Heterozygous by SNPs flanking the <i>HPDL</i> gene by exome sequencing (poor coverage of the <i>HPDL</i> c.527 T>C variant)

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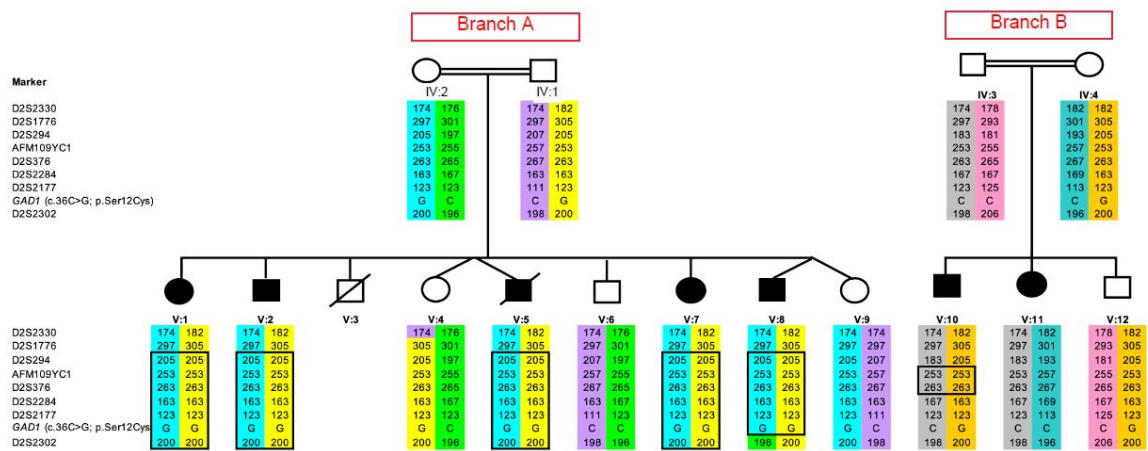
Sanger sequencing results		Heterozygous for <i>HPDL</i> c.527 T>C by Sanger sequencing	Homozygous for <i>HPDL</i> c.527 T>C by Sanger sequencing		Homozygous for <i>HPDL</i> c.527 T>C by Sanger sequencing							
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¹ :see supplementary figure 2

² :see supplementary table 2

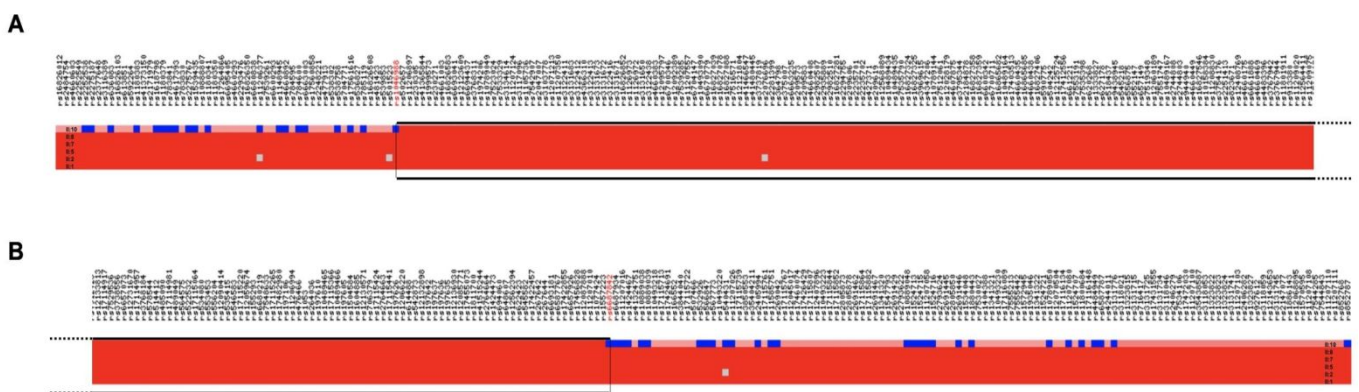
For Review Only

Supplementary Figure 1: results of microsatellite genotyping and *GAD1* variant analysis in Branch A and Branch B



Review Only

Supplementary Figure 2: Results of Affymetrix SNP 5 array genotyping showing the boundaries of the homozygous extended region on chromosome 1 containing *HPDL* for 6 affected individuals from Branch A (V:1, V:2, V:5, V:7, V:8) and B (V:10). The minimal candidate region of overlap between the 6 genotyped affected patients is shown, a 17,596,867 bp region on chromosome 1 between SNP markers rs1046988 (chr1:40,219,065, GRCh37) (panel A) and rs6687842 (chr1:57,815,932, GRCh37) (panel B), both highlighted in red. Homozygous SNP calls are shaded in red, heterozygous SNP calls in pink or blue and no calls shown in grey. Homozygous (minimal candidate region) is shown by borderlines. Larger homozygous region is not shown due to size but contains the *HPDL* gene at genomic position 45,792,545 - 45,794,347 bp. Genomic build shown is version 37.



View Only

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3 **Supplementary Table 2:** This table contains the genotypes called from whole exome sequencing of
4 three affected individuals (V:5, V:10, V:11), and three unaffected individuals (IV:3, IV:4 (parents of
5 V:10, V:11 and V:12) and V:12 (sibling of V:10 and V:11) for informative (IV:3 and IV:4 both
6 heterozygous) SNPs flanking the HPDL variant of interest ((rs773333490, highlighted in yellow in the
7 table). We filtered out variants with missing data in any individual in Branch B. Grey cells depict
8 missing data in V:5 (from Branch A). Green cells depict SNPs where affected individuals (V:5, V:10
9 and V:11) were homozygous and unaffected individuals were heterozygous (IV:3, IV:4 and IV:12). A
10 shared minimal region of autozygosity (5,753,368 bp between markers rs117337557 and rs785467
11 (shaded blue)) common to the three affected individuals is apparent and this region includes the
12 causative variant (yellow) which has inadequate coverage for IV:3 and IV:12. However the flanking
13 SNP genotypes are consistent with the IV:3 being homozygous and IV:12 being heterozygous for the
14 HPDL c.527 T>C; p.Leu176Pro, rs773333490 variant.
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20 Key:

21 Genotypes (GT) are reported for each sample along with the read depth at each position for the
22 sample (DP).

23 GT are coded as 0/0 (homozygous for the reference allele), 1/1 (homozygous for the alternative
24 allele), 0/1 (heterozygous), and ./ (missing).
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30 **Supplementary Table 3:** This table contains the genotypes from the genomewide SNP array v5.0 of
31 six affected individuals from Branch A (V:1, V:2, V:5, V:7, V:8) and Branch B (V:10), in the autozygous
32 region on chromosome 1 containing the *HPDL* gene. Genotypes are indicated as homozygous (AA, BB
33 (highlighted in green)) or heterozygous (AB). The minimal autozygous region is shaded in blue as
34 determined by individual V:10. The number of consecutive homozygous SNPs is shown in brackets
35 after the SNP genotype. The *HPDL* gene at genomic position 45,792,545 - 45,794,347 bp. Genomic
36 build shown is version 37.
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40 **Supplementary Table 4:** This table contains all rare variants that were homozygous for the
41 alternative allele (1/1) in both affected individuals V:10 and V:11 identified by exome sequencing
42 (WES). We filtered out variants with that had a maximum allele frequency (MAX-AF) higher than
43 0.01, which is based on the highest reported allele frequency observed in any population from 1000
44 genomes, ESP or gnomAD. Variants in the autozygous region containing the *HPDL* gene and the
45 causative variant (rs773333490) are highlighted in pink in the table. Genotypes (GT) are reported for
46 each sample along with the read depth at each position for the sample (DP). Biallelic germline
47 mutations in C8A (encoding complement component 8, alpha subunit) are associated with
48 complement component 8 deficiency type I which typically presents with recurrent neisserial
49 infections (particularly meningococcus) after age 10 years. There was no evidence to suggest this
50 diagnosis in the family and the C8A variant was also homozygous (1/1) in the unaffected sibling
51 (V:12) leaving the *HPDL* as the best candidate gene.
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CHROM	POS	ID	REF	ALT	QUAL	V:5		V:10		V:11		V:12		IV:4		IV:3	
						GT	DP	GT	DP	GT	DP	GT	DP	GT	DP	GT	DP
chr1	39753393	rs1046988	C	T	1324.2	./.	29	0/1	48	0/1	47	0/0	20	0/1	30	0/1	48
chr1	39760374	rs12094291	G	A	781.03	0/0	34	0/1	18	0/1	17	0/0	13	0/1	20	0/1	28
chr1	39763696	rs2463260	A	G	2882.03	0/0	44	0/1	127	0/1	148	0/0	33	0/1	47	0/1	52
chr1	40302519	rs117337557	G	A	1617.17	./.	0	1/1	15	1/1	32	0/1	14	0/1	20	0/1	19
chr1	40303513	rs74366749	C	G	9791.54	1/1	15	1/1	89	1/1	156	0/1	147	0/1	119	0/1	164
chr1	40311052	rs202027418	C	T	11952.5	1/1	37	1/1	93	1/1	130	0/1	169	0/1	169	0/1	159
chr1	40495777	rs3795348	T	C	9972.54	1/1	56	1/1	106	1/1	74	0/1	102	0/1	122	0/1	139
chr1	40515558	rs3795344	G	A	12691.5	1/1	34	1/1	83	1/1	99	0/1	236	0/1	273	0/1	247
chr1	40817247	rs13374844	C	T	17163.5	1/1	9	1/1	70	1/1	116	0/1	180	0/1	190	0/1	205
chr1	40817249	rs13374845	C	T	18923.5	1/1	11	1/1	72	1/1	117	0/1	180	0/1	190	0/1	205
chr1	40819913	rs12117176	G	A	33481.5	1/1	88	1/1	184	1/1	261	0/1	144	0/1	209	0/1	236
chr1	40819919	rs12143503	A	G	33152.5	1/1	79	1/1	187	1/1	246	0/1	131	0/1	185	0/1	226
chr1	41382624	rs4359027	G	A	9904.66	./.	5	1/1	169	1/1	179	0/1	24	0/1	26	0/1	38
chr1	42449856	rs12142723	C	A	2974.54	1/1	30	1/1	22	1/1	26	0/1	8	0/1	14	0/1	13
chr1	42450056	rs12047095	C	G	1375.54	0/0	54	0/0	33	0/0	43	0/1	46	0/1	52	0/1	78
chr1	42450082	rs11210647	T	C	780.54	0/0	54	0/0	33	0/0	43	0/1	19	0/1	36	0/1	49
chr1	42556139	rs12038786	C	T	485.54	0/0	53	0/0	11	0/0	30	0/1	35	0/1	33	0/1	29
chr1	42558042	rs71798684	G	GA	209.13	./.	0	0/0	37	0/0	37	0/1	12	0/1	9	0/1	6
chr1	42577032	rs12049046	C	T	1327.54	0/0	42	0/0	37	0/0	34	0/1	41	0/1	55	0/1	44
chr1	42581386	rs12037518	A	G	1134.54	0/0	48	0/0	35	0/0	36	0/1	36	0/1	43	0/1	35
chr1	42747432	rs3738498	A	C	3681.54	1/1	45	1/1	18	1/1	27	0/1	13	0/1	22	0/1	27
chr1	42750259	rs11581921	C	T	13512.5	1/1	28	1/1	138	1/1	197	0/1	134	0/1	120	0/1	155
chr1	42755516	rs3738502	C	T	14564.5	1/1	50	1/1	135	1/1	166	0/1	103	0/1	127	0/1	149
chr1	42837300	rs1466548	C	G	3128.54	1/1	24	1/1	21	1/1	30	0/1	11	0/1	15	0/1	13
chr1	42840082	rs12034000	G	A	28975.5	1/1	212	1/1	278	1/1	363	0/1	107	0/1	148	0/1	163
chr1	42927574	rs11282849	G	GGGT	3177.49	0/0	26	0/0	35	0/0	36	0/1	66	0/1	60	0/1	80
chr1	43569801	rs943513	G	A	4057.54	1/1	19	1/1	38	1/1	62	0/1	13	0/1	13	0/1	14
chr1	43605020	rs17371903	A	G	5561.54	1/1	47	1/1	34	1/1	56	0/1	31	0/1	29	0/1	39
chr1	43605550	rs631248	G	A	7254.54	1/1	16	1/1	71	1/1	91	0/1	111	0/1	78	0/1	141
chr1	43612713	rs499257	T	C	9488.54	1/1	38	1/1	111	1/1	114	0/1	62	0/1	56	0/1	68
chr1	43613740	rs603542	T	C	8743.54	1/1	41	1/1	73	1/1	128	0/1	73	0/1	70	0/1	54

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chr1	43617439	rs571862	C	T	7994.54	1/1	71	1/1	47	1/1	62	0/1	61	0/1	73	0/1	79
chr1	43617836	rs641365	T	C	5656.54	1/1	30	1/1	36	1/1	85	0/1	55	0/1	53	0/1	82
chr1	43617848	rs641351	G	A	5629.54	1/1	33	1/1	33	1/1	81	0/1	53	0/1	51	0/1	81
chr1	43619068	rs1143701	C	T	7562.54	1/1	60	1/1	72	1/1	72	0/1	40	0/1	51	0/1	41
chr1	43621160	rs1143702	C	T	7275.54	1/1	21	1/1	74	1/1	61	0/1	81	0/1	67	0/1	81
chr1	44130707	rs34057178	CCAA	C	7112.5	0/0	80	0/0	34	0/0	34	0/1	150	0/1	125	0/1	151
chr1	44220650	rs12419	T	C	2309.54	0/0	19	0/0	41	0/0	33	0/1	80	0/1	115	0/1	89
chr1	44775702	rs3806398	G	A	2742.54	1/1	23	1/1	16	1/1	32	0/1	18	0/1	13	0/1	23
chr1	44802710	rs12404160	G	A	2402.54	1/1	27	1/1	10	1/1	22	0/1	16	0/1	16	0/1	16
chr1	44827194	rs2295996	G	A	18191.5	1/1	44	1/1	141	1/1	221	0/1	239	0/1	250	0/1	319
chr1	44827846	rs7525308	A	G	8670.54	1/1	17	1/1	108	1/1	137	0/1	51	0/1	53	0/1	67
chr1	44828484	rs2273940	T	C	9084.54	1/1	16	1/1	102	1/1	115	0/1	66	0/1	104	0/1	98
chr1	44841834	rs3795720	G	A	11686.5	1/1	55	1/1	93	1/1	111	0/1	162	0/1	155	0/1	184
chr1	44874610	rs9988451	C	T	3247.55	1/1	26	1/1	34	1/1	29	0/1	6	0/1	17	0/1	6
chr1	44879774	rs3738248	T	A	13883.5	1/1	30	1/1	138	1/1	195	0/1	142	0/1	117	0/1	142
chr1	45003483	rs11548990	C	T	5572.54	1/1	33	1/1	44	1/1	66	0/1	37	0/1	62	0/1	54
chr1	45003914	rs41312020	G	A	3805.54	1/1	31	1/1	30	1/1	44	0/1	23	0/1	19	0/1	42
chr1	45013827	rs2234480	G	C	13317.5	1/1	62	1/1	103	1/1	171	0/1	96	0/1	107	0/1	148
chr1	45014037	rs2228084	A	G	21636.5	1/1	50	1/1	185	1/1	200	0/1	245	0/1	326	0/1	374
chr1	45327675	rs773333490	T	C	4090.95	1/1	13	1/1	54	1/1	62	./.	4	0/1	15	./.	8
chr1	45330597	rs3219493	G	C	3262.66	./.	6	0/0	36	0/0	33	0/1	128	0/1	129	0/1	143
chr1	45332883	rs3219487	T	C	8631.54	0/0	59	0/0	36	0/0	37	0/1	304	0/1	315	0/1	363
chr1	45499668	rs7546268	T	C	879.14	./.	1	0/0	31	0/0	39	0/1	24	0/1	21	0/1	24
chr1	45613182	rs2230658	A	G	8527.54	1/1	46	1/1	78	1/1	106	0/1	81	0/1	94	0/1	108
chr1	45613212	rs2230659	C	T	9267.54	1/1	49	1/1	85	1/1	110	0/1	100	0/1	111	0/1	134
chr1	45616288	rs2253862	C	T	6541.54	1/1	46	1/1	73	1/1	75	0/1	35	0/1	42	0/1	49
chr1	45616759	rs2991988	C	A	6165.54	1/1	44	1/1	64	1/1	58	0/1	31	0/1	22	0/1	45
chr1	45618034	rs41310432	C	G	5630	./.	3	1/1	69	1/1	106	0/1	50	0/1	58	0/1	61
chr1	45620405	rs3014246	C	T	4438.54	1/1	60	1/1	33	1/1	20	0/1	27	0/1	24	0/1	18
chr1	45620657	rs11590549	C	T	3027.54	1/1	25	1/1	17	1/1	28	0/1	29	0/1	28	0/1	6
chr1	45621905	rs3014242	G	A	3528.54	1/1	17	1/1	33	1/1	55	0/1	26	0/1	16	0/1	37
chr1	45622207	rs3014241	A	G	6135.54	1/1	27	1/1	14	1/1	20	0/1	110	0/1	104	0/1	120
chr1	45623553	rs3014240	C	G	5325.78	./.	4	1/1	63	1/1	72	0/1	42	0/1	80	0/1	71

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2	chr1	45628355	rs1250	A	G	3867.54	0/0	35	0/0	35	0/0	34	0/1	84	0/1	137	0/1	120
3	chr1	45629600	rs1135812	T	C	4240.54	0/0	42	0/0	40	0/0	39	0/1	157	0/1	159	0/1	202
4	chr1	45694588	rs6658700	C	T	715.54	0/0	12	0/0	35	0/0	38	0/1	22	0/1	21	0/1	29
5	chr1	45729539	rs139068713	TAAC	T	2138.5	0/0	39	0/0	10	0/0	25	0/1	30	0/1	40	0/1	34
6	chr1	45729597	rs768635994	T	C	9100.54	1/1	97	1/1	31	1/1	53	0/1	70	0/1	102	0/1	81
7	chr1	45729703	rs28375469	T	C	2757.54	0/0	47	0/0	33	0/0	43	0/1	97	0/1	104	0/1	123
8	chr1	45913846	rs4256763	A	G	1004.17	./.	0	0/0	36	0/0	37	0/1	47	0/1	18	0/1	45
9	chr1	46008230	rs3922886	A	T	745.54	0/0	12	0/0	49	0/0	38	0/1	21	0/1	27	0/1	18
10	chr1	46010915	rs11211247	T	G	2962.54	0/0	50	0/0	33	0/0	36	0/1	68	0/1	83	0/1	87
11	chr1	46032703	rs1622208	A	G	5349.54	1/1	35	1/1	51	1/1	62	0/1	29	0/1	48	0/1	53
12	chr1	46034941	rs17855317	T	C	8911.54	1/1	83	1/1	62	1/1	70	0/1	41	0/1	62	0/1	75
13	chr1	46055845	rs785468	G	A	13044.5	1/1	123	1/1	72	1/1	99	0/1	80	0/1	99	0/1	119
14	chr1	46055887	rs785467	A	T	14137.5	1/1	83	1/1	108	1/1	147	0/1	94	0/1	115	0/1	162
15	chr1	46131824	rs751868703	CT	C	375.02	0/0	124	0/0	44	0/1	68	0/1	33	0/1	30	0/1	34
16	chr1	46511165	rs1886116	C	T	9463.54	1/1	24	1/1	102	1/1	124	0/1	129	0/1	124	0/1	131
17	chr1	46594828	rs183190014	T	C	3063.17	./.	0	1/1	45	1/1	55	0/1	31	0/1	18	0/1	28
18	chr1	46643263	rs763191064	A	G	8689.54	1/1	27	1/1	80	1/1	127	0/1	71	0/1	65	0/1	100
19	chr1	46813503	rs4646487	C	T	11437.5	1/1	42	1/1	91	1/1	96	0/1	181	0/1	176	0/1	188
20	chr1	46813886	rs4646489	CCCT	C	7722.5	1/1	79	1/1	32	1/1	31	0/1	32	0/1	21	0/1	31
21	chr1	46818854	rs3766196	C	A	2949.54	1/1	31	1/1	23	1/1	18	0/1	13	0/1	12	0/1	14
22	chr1	46932824	rs1126742	A	G	9072.54	1/1	105	1/1	69	1/1	83	0/1	73	0/1	98	0/1	132
23	chr1	47029933	rs17102977	A	G	4491.54	1/1	42	1/1	33	1/1	35	0/1	11	0/1	16	0/1	21
24	chr1	47031365	rs11211420	G	A	9379.54	1/1	138	1/1	36	1/1	47	0/1	23	0/1	29	0/1	37
25	chr1	47106230	rs4926802	C	T	8175	1/1	81	0/1	41	0/1	45	0/1	67	0/1	56	0/1	92
26	chr1	47106233	rs28463559	C	T	8154	1/1	78	0/1	40	0/1	45	0/1	65	0/1	48	0/1	91
27	chr1	47140798	rs56258513	C	A	11611.5	1/1	85	1/1	89	1/1	105	0/1	99	0/1	104	0/1	136
28	chr1	47141609	rs12564525	C	T	10920.5	1/1	72	1/1	103	1/1	164	0/1	44	0/1	52	0/1	38
29	chr1	47142090	rs2056901	T	C	5219.54	1/1	42	1/1	27	1/1	54	0/1	41	0/1	44	0/1	55
30	chr1	47142113	rs2056900	G	A	7188.54	1/1	49	1/1	42	1/1	71	0/1	68	0/1	65	0/1	93
31	chr1	47143311	rs4926581	G	T	5130.54	1/1	19	1/1	49	1/1	69	0/1	48	0/1	48	0/1	57
32	chr1	47143817	rs10789501	T	C	7609.54	1/1	88	1/1	43	1/1	45	0/1	69	0/1	37	0/1	62
33	chr1	47144393	rs6661132	A	C	9129.54	1/1	59	1/1	62	1/1	76	0/1	110	0/1	94	0/1	132
34	chr1	47145926	rs2405599	T	C	14617.5	1/1	227	1/1	42	1/1	52	0/1	67	0/1	80	0/1	124
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2	chr1	47148762	rs4926600	C	T	7202.54	1/1	43	1/1	74	1/1	61	0/1	59	0/1	63	0/1	67
3	chr1	47189927	rs11801079	C	T	645.54	0/0	14	0/0	38	0/0	40	0/1	26	0/1	29	0/1	51
4	chr1	48232079	rs61997212	C	T	6921.54	1/1	41	1/1	65	1/1	77	0/1	64	0/1	77	0/1	94
5	chr1	48298747	rs1056042	C	T	12608.5	1/1	161	1/1	72	1/1	72	0/1	59	0/1	74	0/1	68
6	chr1	48472019	rs2275309	A	T	1729	./.	3	1/1	11	1/1	35	0/1	8	0/1	14	0/1	20
7																		
8	chr1	50491837	rs1149790	A	T	563.54	0/0	38	0/0	34	0/0	40	0/1	24	0/1	22	0/1	36
9	chr1	50655526	rs11205753	T	C	8191.54	1/1	62	1/1	67	1/1	87	0/1	37	0/1	56	0/1	70
10	chr1	51302240	rs375305601	GC	G	469.41	./.	28	0/1	91	0/1	80	0/1	98	0/1	81	0/1	94
11	chr1	51361249	rs17567	T	C	15398.5	1/1	84	1/1	154	1/1	180	0/1	125	0/1	105	0/1	130
12	chr1	51402391	rs202067917	C	CT	3424.5	./.	9	1/1	58	1/1	45	0/1	6	0/1	22	0/1	12
13																		
14	chr1	51447088	rs34704431	T	G	14656.5	1/1	94	1/1	153	1/1	203	0/1	78	0/1	59	0/1	71
15	chr1	51458554	rs6672300	C	T	2489.17	./.	0	1/1	36	1/1	44	0/1	31	0/1	29	0/1	32
16	chr1	51463832	rs5774101	TA	T	6347.5	1/1	43	1/1	37	1/1	73	0/1	13	0/1	26	0/1	14
17	chr1	51825345	rs370502514	C	T	10346.5	1/1	69	1/1	79	1/1	88	0/1	58	0/1	95	0/1	83
18	chr1	52340059	chrchr1bp523	C	T	6231.54	1/1	15	1/1	47	1/1	59	0/1	78	0/1	92	0/1	116
19	chr1	52784978	rs6588443	G	A	1880.76	0/0	46	0/0	62	0/0	37	0/0	33	0/1	110	0/1	131
20	chr1	52805553	rs487453	T	C	1117.42	./.	0	0/0	37	0/0	40	0/0	39	0/1	41	0/1	60
21	chr1	52987968	rs7528565	A	G	284.76	0/0	18	0/0	34	0/0	35	0/0	14	0/1	22	0/1	19
22	chr1	52988011	rs7516939	C	T	1158.76	0/0	34	0/0	34	0/0	35	0/0	33	0/1	67	0/1	70
23	chr1	52988042	rs7538935	G	T	2023.76	0/0	43	0/0	34	0/0	35	0/0	33	0/1	117	0/1	121
24	chr1	53047751	rs11206079	C	T	305.76	./.	9	0/0	35	0/0	41	0/0	10	0/1	12	0/1	21
25	chr1	53069827	rs17107831	C	T	581.37	./.	2	0/0	26	0/0	24	0/0	20	0/1	16	0/1	42
26	chr1	53210776	rs1799821	G	A	1755.76	0/0	52	0/0	35	0/0	35	0/0	33	0/1	92	0/1	102
27	chr1	53247055	rs5174	C	T	1031.76	0/0	47	0/0	34	0/0	34	0/0	36	0/1	55	0/1	65
28	chr1	53250744	rs3737983	G	A	4883.76	0/0	59	0/0	35	0/0	36	0/0	37	0/1	210	0/1	254
29	chr1	53266643	rs2297660	G	T	679.76	0/0	68	0/0	36	0/0	35	0/0	44	0/1	56	0/1	45
30	chr1	53464878	rs943514	G	A	16359.8	1/1	53	1/1	124	1/1	173	1/1	146	0/1	172	0/1	198
31	chr1	53878609	rs2272928	C	A	7849.76	1/1	42	1/1	44	1/1	67	1/1	65	0/1	66	0/1	73
32	chr1	53904591	rs2294511	A	T	250.76	0/0	83	0/0	12	0/0	22	0/0	18	0/1	11	0/1	17
33	chr1	54594551	rs1755591	G	A	4694.76	1/1	20	1/1	43	1/1	52	1/1	29	0/1	34	0/1	32
34	chr1	54594689	rs1702003	G	A	4351.76	1/1	32	1/1	31	1/1	35	1/1	21	0/1	25	0/1	30
35	chr1	54597287	rs2304305	G	A	11621.8	1/1	25	1/1	85	1/1	78	1/1	156	0/1	159	0/1	179
36	chr1	54610440	rs4926650	G	A	10993.8	1/1	97	1/1	61	1/1	69	1/1	77	0/1	74	0/1	106
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2	chr1	54653471	rs11206407	A	G	11881.8	1/1	80	1/1	102	1/1	135	1/1	62	0/1	56	0/1	57
3	chr1	54653616	rs9332417	G	C	25034.8	1/1	72	1/1	210	1/1	219	1/1	236	0/1	291	0/1	339
4	chr1	54653842	rs1655519	T	G	2789.76	0/0	78	0/0	51	0/0	69	0/0	84	0/1	139	0/1	156
5	chr1	54665258	rs2078273	C	T	9914.76	1/1	44	1/1	86	1/1	127	1/1	54	0/1	53	0/1	70
6	chr1	54665273	rs6703113	G	C	7471.76	1/1	38	1/1	55	1/1	91	1/1	40	0/1	34	0/1	46
7																		
8	chr1	54668814	rs478513	G	A	780.76	0/0	51	0/0	19	0/0	27	0/0	35	0/1	43	0/1	43
9	chr1	54670856	rs480963	T	C	1871.76	0/0	15	0/0	34	0/0	33	0/0	38	0/1	94	0/1	121
10	chr1	54716630	rs145266091	A	G	27128.8	1/1	63	1/1	212	1/1	219	1/1	81	0/1	65	0/1	69
11	chr1	54721252	rs3737825	G	A	1183.76	0/0	33	0/0	45	0/0	40	0/0	38	0/1	48	0/1	62
12	chr1	54722709	rs11550481	C	T	1960.76	0/0	46	0/0	34	0/0	33	0/0	35	0/1	90	0/1	115
13	chr1	54722819	rs201524669	CAATT	C	1311.71	0/0	46	0/0	34	0/0	33	0/0	35	0/1	30	0/1	37
14	chr1	54731476	rs3815226	T	C	6665.76	1/1	31	1/1	35	1/1	60	1/1	69	0/1	42	0/1	66
15	chr1	54741543	rs41297863	T	C	594.76	0/0	53	0/0	40	0/0	34	0/0	34	0/1	48	0/1	38
16	chr1	54801172	rs1147984	C	T	7903.76	1/1	11	1/1	31	1/1	31	1/1	111	0/1	129	0/1	142
17	chr1	54807907	rs9782980	G	T	8621.76	1/1	101	1/1	67	1/1	54	1/1	18	0/1	27	0/1	30
18	chr1	55005149	rs41297879	T	G	6145.76	1/1	17	1/1	56	1/1	65	1/1	35	0/1	62	0/1	63
19	chr1	55052188	rs613855	C	G	551.76	0/0	58	0/0	35	0/0	34	0/0	38	0/1	38	0/1	40
20	chr1	55052210	rs624612	C	G	1929.76	0/0	62	0/0	35	0/0	34	0/0	38	0/1	81	0/1	97
21	chr1	55052794	rs2495477	A	G	11328.8	1/1	19	1/1	72	1/1	76	1/1	147	0/1	164	0/1	177
22	chr1	55052855	rs494198	C	A	1891.76	0/0	15	0/0	29	0/0	36	0/0	60	0/1	74	0/1	109
23	chr1	56753958	rs11404547	G	GA	1455.71	0/0	40	0/0	34	0/0	33	0/0	48	0/1	72	0/1	84
24	chr1	56755880	rs1774808	G	T	3001.76	0/0	47	0/0	33	0/0	34	0/0	35	0/1	55	0/1	83
25	chr1	56755881	rs1738401	A	G	3001.76	0/0	47	0/0	33	0/0	34	0/0	35	0/1	55	0/1	83
26	chr1	56758787	rs857103	G	C	662.76	0/0	14	0/0	33	0/0	33	0/0	35	0/1	42	0/1	64
27	chr1	56875054	rs652785	C	A	23419.8	1/1	18	1/1	218	1/1	321	1/1	186	0/1	170	0/1	261
28	chr1	56883734	rs6694643	A	T	1026.76	0/0	27	0/0	32	0/0	25	0/0	33	0/1	33	0/1	59
29	chr1	56886031	rs147513844	G	T	18991.8	1/1	64	1/1	187	1/1	234	1/1	113	0/1	109	0/1	136
30	chr1	56951960	rs138791380	G	GGGA	1211.71	0/0	32	0/0	35	0/0	34	0/0	39	0/1	29	0/1	33
31	chr1	57033602	rs17417626	A	G	17564.8	1/1	36	1/1	163	1/1	208	1/1	138	0/1	106	0/1	127
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Supplementary Table 3

dbSNP	pos on chr 1	V:10	V:8	V:7	V:5	V:2	V:1
rs16826012	39719519	BB (4)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs2484754	39728964	AA (4)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
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rs2252538	39743929	AB (0)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs2275187	39763242	AB (0)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs2377648	39780802	AA (2)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs3116389	39785641	BB (2)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs636083	39821681	AB (0)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs16826103	39821705	AA (3)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs613851	39823866	AA (3)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs592264	39845622	BB (3)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs1180383	39846267	AB (0)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs41373150	39859900	AA (2)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs2171979	39885977	BB (2)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs1618798	39886940	AB (0)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs1180379	39915035	AB (0)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs783841	39922303	AB (0)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs4617393	39940223	AB (0)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs783830	39945177	AA (1)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs2275767	39945297	AB (0)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs1539435	39968824	AB (0)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs10888807	39970705	AA (1)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs1180341	39992808	AB (0)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs1180350	40007013	BB (7)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs17264866	40022777	AA (7)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs6695405	40025080	AA (7)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs4660293	40028180	BB (7)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs2293476	40036847	BB (7)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs16826350	40039611	BB (7)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs6680838	40040635	BB (7)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs11206377	40049104	AB (0)	BB (3236)	BB (3236)	BB (3236)	-3290	BB (2800)
rs7539261	40050503	BB (2)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs6600293	40083819	AA (2)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs1148945	40096053	AB (0)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs4660892	40104584	AB (0)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs784595	40135017	BB (1)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs784600	40139553	AB (0)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs6663003	40143327	AB (0)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs17560858	40145850	AA (4)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs2248221	40152117	AA (4)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs477913	40153141	BB (4)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs538302	40153748	AA (4)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs538758	40155188	AB (0)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs704771	40161013	AA (1)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs17561616	40168865	AB (0)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs536827	40169724	BB (1)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)

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2	rs785119	40178268	AB (0)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
3	rs16826508	40179431	AA (4)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
4	rs481423	40183922	AA (4)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
5	rs539853	40194903	BB (4)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
6	rs501523	40216993	BB (4)	BB (3236)	BB (3236)	BB (3236)	-3290	BB (2800)
8	rs1046988	40219065	AB (0)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
9	rs502657	40221855	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
10	rs11206897	40264146	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
11	rs230305	40283051	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
12	rs41365844	40284187	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
13	rs1999573	40289812	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
14	rs230271	40290597	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
15	rs4661003	40290696	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
16	rs16826583	40291192	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
17	rs6693041	40302463	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
18	rs12123609	40303627	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
19	rs6694437	40304647	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
20	rs3768311	40308017	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
21	rs1974206	40308918	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
22	rs10789049	40343432	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
23	rs7523024	40346001	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
24	rs7533329	40346129	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
25	rs3134614	40363054	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
26	rs11207124	40381453	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
27	rs3117096	40389966	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
28	rs1105736	40390532	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
29	rs7542907	40391752	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
30	rs2047007	40427975	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
31	rs3103778	40433771	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
32	rs12071213	40445667	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
33	rs11207350	40491765	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
34	rs3122411	40503779	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
35	rs3131683	40513710	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
36	rs3122412	40515277	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
37	rs4366310	40515346	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
38	rs3122413	40519523	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
39	rs3131673	40522971	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
40	rs3131672	40523157	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
41	rs3122416	40523328	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
42	rs3131671	40524690	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
43	rs16826852	40531094	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
44	rs1126973	40539076	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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47	rs3122438	40555802	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
48	rs4660383	40559248	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
49	rs12095797	40588288	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
50	rs6700346	40588548	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
51	rs12092389	40593127	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
52	rs7539285	40594472	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)

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2	rs11207557	40595283	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
3	rs6704147	40595353	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
4	rs10493090	40626918	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
5	rs6670779	40657449	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
6	rs16827039	40665502	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
7	rs16827078	40702570	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
8	rs16827088	40714034	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
9	rs7516571	40733658	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
10	rs12121804	40761848	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
11	rs41466547	40766542	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
12	rs41480445	40766618	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
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17	rs375007	40799989	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
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20	rs209583	40847491	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
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22	rs2982507	40865801	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
23	rs2935889	40872356	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
24	rs2982510	40872623	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
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27	rs209606	40881041	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
28	rs2235701	40883095	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
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31	rs209619	40904757	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
32	rs12568089	40921238	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
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36	rs16827324	40927959	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
37	rs16827325	40930155	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
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39	rs4347206	40938751	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
40	rs10789144	40955156	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
41	rs7512061	40966183	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
42	rs12028179	40966458	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
43	rs4607875	40975260	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
44	rs3795344	40981230	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
45	rs2184878	40994050	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
46	rs16827358	40999475	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
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48	rs6600342	41004021	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
49	rs6700711	41005205	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
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51	rs10489164	41011140	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	

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rs4660438	41050952	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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rs17412556	41088577	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
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rs1333827	41111970	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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rs11209020	41317020	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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3	rs11209145	41372237	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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6	rs6698251	41383838	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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26	rs3819842	41563086	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
27	rs721651	41563329	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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30	rs3766323	41566096	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
31	rs2300648	41569488	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
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39	rs6672914	41666929	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
40	rs12121121	41684456	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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43	rs12123384	41705018	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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51	rs12037129	41794301	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)

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40	rs2485992	44502650	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
41	rs17413167	44507773	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
42	rs17413277	44510128	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
43	rs2057968	44515696	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
44	rs2158822	44519447	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
45	rs6429646	44523706	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
46	rs16831607	44534252	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
47	rs942755	44537401	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
48	rs7531328	44541316	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
49	rs6429648	44566221	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
50	rs509261	44568322	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
51	rs4660776	44568478	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	

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rs663818	44578559	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs7513563	44585616	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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rs17414232	44623428	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs11210971	44635188	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs1291169	44681113	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs12419	44686322	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs1152030	44697673	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs17414912	44699848	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs325168	44731068	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs369493	44740024	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs10890295	44745450	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs325164	44751390	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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rs325155	44764547	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs6429535	44766071	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs325150	44767913	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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rs161721	44788398	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
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rs2524353	44807164	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs226067	44820319	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs6429539	44860224	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
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rs1340647	44889509	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
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rs7555873	44912147	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs1417371	44920992	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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rs7550326	44946187	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs7549585	44947616	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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rs2792597	44954342	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs6429541	44965723	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs272531	44988433	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs270724	44991278	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs272537	44992714	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs10890308	44998464	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs746733	44999614	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
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2	rs7554123	45004204	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
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5	rs156256	45018915	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
6	rs156255	45018937	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
7	rs2924869	45019168	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
8	rs12568345	45021601	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
9	rs69141	45039205	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
10	rs11810526	45040118	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
11	rs11211015	45042741	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
12	rs16832091	45045989	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
13	rs270707	45049458	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
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16	rs156650	45056293	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
17	rs17842391	45064759	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
18	rs17383073	45070142	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
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23	rs3768439	45113326	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
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27	rs1321703	45157738	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
28	rs2355710	45166513	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
29	rs2180765	45167143	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
30	rs2983713	45168726	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
31	rs10890313	45175925	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
32	rs11211026	45187285	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
33	rs6689911	45219952	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
34	rs17391430	45234389	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
35	rs6676749	45264545	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
36	rs11573588	45293090	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
37	rs16832263	45299050	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
38	rs7517439	45312867	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
39	rs11211046	45321563	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
40	rs16832301	45327716	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
41	rs17390398	45333256	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
42	rs17392632	45333719	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
43	rs263975	45358527	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
44	rs263977	45360287	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
45	rs3767490	45360658	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
46	rs10890315	45362445	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
47	rs263978	45362992	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
48	rs608649	45363887	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
49	rs7531019	45365834	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
50	rs1572379	45369086	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
51	rs3903059	45392955	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	

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rs264002	45423943	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs11211059	45430218	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs11556200	45444038	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
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rs7535347	45466511	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs7541207	45474008	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs2236576	45477675	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs6678726	45496395	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs346732	45585348	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs1226757	45592886	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs346696	45596447	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs11585488	45617312	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs1698295	45635802	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs2997465	45641454	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs3009973	45665580	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
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rs6688723	45763542	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs7548931	45766192	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs7535487	45791189	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs3219487	45798555	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs9429072	45810091	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs2153608	45813941	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs2185549	45814103	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs4660849	45819872	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs9326141	45828943	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs7543428	45839643	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
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rs1826691	45881262	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs1771551	45883643	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs41343546	45908263	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
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rs2275276	45973928	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs4660306	45978675	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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rs6699702	45981834	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs10736426	45984353	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs7522705	45992300	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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rs487174	46009536	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs868351	46009818	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs669417	46026737	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs3014216	46035760	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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2	rs3014210	46066608	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
3	rs17410308	46078191	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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5	rs2991986	46081282	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
6	rs1547924	46085112	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
7	rs1547925	46085356	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
8	rs2275086	46085879	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
9	rs3014245	46086913	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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11	rs5021934	46109304	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
12	rs4660313	46110841	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
13	rs4660880	46111040	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
14	rs11211150	46112693	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
15	rs6662999	46119351	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
16	rs11211176	46223086	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
17	rs6702764	46252717	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
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19	rs7539800	46262129	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
20	rs4660316	46273183	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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22	rs10789477	46306583	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
23	rs10749859	46314170	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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33	rs6700322	46409176	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
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37	rs4660334	46462126	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
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40	rs11579634	46466391	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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44	rs946526	46487168	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
45	rs785480	46492164	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
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47	rs1768809	46502836	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
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50	rs1707321	46505309	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
51	rs4388641	46522326	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)

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rs785495	46587887	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs41378047	46602011	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs2486445	46605369	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs41401147	46608657	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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rs41534051	46828734	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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rs4141964	46865040	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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rs1150066	46901546	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
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rs12562748	46961914	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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2	rs2404611	46997127	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
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31	rs41394646	48467450	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
32	rs6695419	48479075	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
33	rs303916	48479525	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
34	rs2492791	48484327	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
35	rs1413330	48485047	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
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40	rs6588642	48507037	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
41	rs748142	48509053	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
42	rs748141	48509159	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
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51	rs2270973	48520777	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	

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rs592072	48543158	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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rs17468972	48568400	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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rs6670286	48709019	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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rs498502	48725096	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs6704171	48725431	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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rs743065	48734366	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs212968	48734666	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
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rs7536253	48775943	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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rs11205476	48820294	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
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rs3767616	48857313	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs1416043	48863365	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
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rs2485911	48910976	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs2255264	48911430	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs2803251	48911793	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs2787873	48932488	AB (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs2245912	48939678	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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2	rs7527001	48976856	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
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36	rs7517510	49107979	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
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38	rs17373849	49113247	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
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51	rs7532350	49141251	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	

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51	rs1766377	49917084	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	

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rs2762819	52798033	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs2820337	52801061	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs7517338	52811581	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs9633423	52836139	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)

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3	rs6672059	52855047	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
4	rs3087477	52861871	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
5	rs6665054	52874114	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
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7	rs10127490	52874787	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
8	rs17373044	52887953	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
9	rs10493166	52897395	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
10	rs12239281	52915793	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
11	rs7554003	52916371	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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13	rs13376564	52928448	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
14	rs6588429	52972639	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
15	rs997915	52973210	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
16	rs7553627	52986978	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
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31	rs2485917	53112410	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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33	rs269324	53126155	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
34	rs6672879	53137112	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
35	rs12067454	53137328	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
36	rs448788	53161620	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
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39	rs387436	53179376	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
40	rs425520	53179805	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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44	rs12095247	53193796	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
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46	rs6588439	53209442	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
47	rs4926928	53210163	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
48	rs10157120	53210888	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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51	rs10888755	53240829	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
52	rs2065477	53250213	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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54	rs12731800	53286967	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)

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rs512723	53343880	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
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rs12094972	53385292	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs17107484	53410388	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
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rs11206061	53455899	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
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rs11581638	53459690	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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rs34418279	53608425	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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rs1481043	53619502	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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5	rs1288333	53630914	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
6	rs1288332	53630960	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
7	rs2046140	53632424	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
8	rs4543741	53636177	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
9	rs3125253	53637106	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
10	rs7531337	53641911	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
11	rs3102047	53642449	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
12	rs6704459	53643199	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
13	rs6588471	53644967	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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16	rs869987	53723190	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
17	rs869988	53723349	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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19	rs2297657	53746595	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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22	rs884586	53782093	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
23	rs41360049	53788279	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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6	rs2073109	54827554	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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11	rs213481	54861540	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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13	rs4244644	54883163	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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31	rs300271	55046039	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
32	rs11811633	55046130	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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38	rs732509	55067992	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
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40	rs10736390	55103945	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
41	rs11206403	55106933	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
42	rs10888858	55108978	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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44	rs1701999	55115608	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
45	rs1655519	55119515	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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49	rs571199	55139618	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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51	rs498831	55144100	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)

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rs13046	55207754	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs6698255	55212719	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
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7	rs7524689	55416974	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
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9	rs7520033	55418263	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
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13	rs7523658	55428397	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
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18	rs1015979	55449273	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
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27	rs2479393	55488369	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
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31	rs2479409	55504650	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
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33	rs12066265	55513169	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
34	rs7552841	55518752	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
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44	rs1165282	55567143	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
45	rs615652	55567410	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
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48	rs640238	55593546	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
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50	rs1475702	55596587	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
51	rs1475703	55596716	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	

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rs880386	55820264	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs41438246	55820441	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs2179853	55820713	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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15	rs11800773	56382975	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
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54	rs6703713	56616961	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	

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rs778417	56644567	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
rs778420	56647914	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs17362438	56654191	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
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rs697581	56689365	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
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rs2793669	56741692	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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5	rs3005896	56785831	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
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7	rs3005907	56790785	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
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9	rs7525535	56799799	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
10	rs10493206	56819151	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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12	rs4477326	56821054	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
13	rs17113783	56823378	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
14	rs4912230	56826300	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
15	rs17113792	56829396	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
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17	rs4600084	56844621	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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47	rs4532850	56996104	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
48	rs11206836	56996741	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
49	rs10888979	57010711	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
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rs41446844	57031938	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
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30	rs545582	57767050	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
31	rs17422457	57780247	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
32	rs267642	57780985	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
33	rs267644	57790529	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
34	rs6693181	57799785	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
35	rs6587767	57800285	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
36	rs17422855	57801345	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
37	rs6657926	57801950	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
38	rs17456828	57802255	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
39	rs12087888	57802759	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
40	rs11207010	57802886	BB (2385)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)
41	rs1077424	57810304	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
42	rs1557223	57810618	AA (2385)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
43	rs6687842	57815932	AB (0)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
44	rs6687934	57816011	AB (0)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
45	rs11207016	57816224	AB (0)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
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4	rs3844040	57854451	AA (7)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
5	rs17115722	57855219	BB (7)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
6	rs507466	57855425	AA (7)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
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9	rs567773	57857390	AB (0)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
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17	rs17115761	57872860	AA (1)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
18	rs11585751	57872987	AB (0)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
19	rs269042	57874048	AB (0)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
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27	rs17115844	57934206	BB (19)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
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35	rs1341320	57966528	AA (19)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
36	rs1547793	57966812	AA (19)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
37	rs2691429	57969583	AA (19)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
38	rs2793622	57971846	AA (19)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
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40	rs1524713	58003131	AB (0)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
41	rs1524715	58003409	AB (0)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
42	rs11207058	58003804	AB (0)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
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44	rs2793624	58006318	AA (3)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
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46	rs2805850	58006699	AA (3)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
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51	rs1404388	58043416	AA (11)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	

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2	rs1341321	58044168	AA (11)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
3	rs10493230	58050139	BB (11)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
4	rs17116089	58051878	AA (11)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
5	rs7555213	58062496	BB (11)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
6	rs7555442	58062765	BB (11)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
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8	rs1935046	58075285	AA (11)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
9	rs1341322	58079143	AA (11)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
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12	rs12070504	58084901	AA (2)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
13	rs17116134	58093163	BB (2)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
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15	rs1524707	58102540	BB (1)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
16	rs11800684	58103010	AB (0)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
17	rs17116148	58105811	BB (1)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
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19	rs6587787	58125050	AB (0)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
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21	rs3131776	58162616	AB (0)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
22	rs3118021	58173935	AA (35)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
23	rs1323815	58177003	BB (35)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
24	rs3131715	58178278	AA (35)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
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26	rs3131725	58191387	BB (35)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
27	rs17431555	58208277	BB (35)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
28	rs3131734	58209220	AA (35)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
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31	rs7534106	58215034	AA (35)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
32	rs17472030	58221074	AA (35)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
33	rs11207100	58224212	BB (35)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
34	rs35438587	58224332	BB (35)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
35	rs3118053	58226522	BB (35)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
36	rs1323822	58226592	BB (35)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
37	rs1323823	58226667	BB (35)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
38	rs1323824	58226786	AA (35)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
39	rs3131741	58227195	AA (35)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
40	rs11207103	58232061	AA (35)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
41	rs2406282	58232329	BB (35)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
42	rs1323827	58232720	AA (35)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
43	rs927612	58234759	BB (35)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
44	rs3118059	58235497	BB (35)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
45	rs12143653	58238545	BB (35)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
46	rs3131745	58238692	BB (35)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
47	rs2147077	58244952	AA (35)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
48	rs1886143	58258134	AA (35)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)	
49	rs12068895	58259108	BB (35)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	
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51	rs11207108	58259705	BB (35)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)	

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rs7544285	58263118	AA (35)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
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rs852768	58287622	AA (35)	AA (3236)	AA (3236)	AA (3236)	AA (3290)	AA (2800)
rs852787	58302102	AB (0)	BB (3236)	BB (3236)	BB (3236)	BB (3290)	BB (2800)

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Individual

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Supplementary Table 4

CHROM	POS	ID	REF	ALT	QUAL	Consequence	IMPACT	SYMBOL
chr1	44806175	rs759039462	GGGCTG	G	6255.13	downstream_gene_variant	MODIFIER	PLK3
chr1	45206192	rs768634679	G	T	8826.54	synonymous_variant	LOW	ZSWIM5
chr1	45327675	rs773333490	T	C	4090.95	missense_variant	MODERATE	HPDL
chr1	45729597	rs768635994	T	C	9100.54	intron_variant	MODIFIER	IPP
chr1	46191982	rs528667376	C	G	3633.66	intron_variant	MODIFIER	POMGNT1
chr1	46643263	rs763191064	A	G	8689.54	synonymous_variant	LOW	ATPAF1
chr1	51825345	rs370502514	C	T	10346.5	synonymous_variant	LOW	NRDC
chr1	56886031	rs147513844	G	T	18991.8	missense_variant	MODERATE	C8A
chr1	120814790	chrchr1bp12081	C	T	9890.93	intron_variant	MODIFIER	NBPF26
chr1	150230711	rs782288239	G	GA	7673.89	intron_variant	MODIFIER	ANP32E
chr10	17849701	rs606231248	G	A	37253.9	missense_variant	MODERATE	MRC1
chr10	50210866	chrchr10bp5021	T	C	37082.9	synonymous_variant	LOW	ASAH2
chr11	36271570	rs543326317	G	GACTT	20957	downstream_gene_variant	MODIFIER	COMMD9
chr11	46317341	rs577462367	C	T	3885.01	intron_variant	MODIFIER	CREB3L1
chr11	46726097	rs138260543	C	T	11664	synonymous_variant	LOW	F2
chr11	55665389	rs201388016	G	A	10138	missense_variant	MODERATE	OR4C6
chr11	55827767	rs374078582	T	C	7663.01	synonymous_variant	LOW	OR5L2
chr11	61342488	rs200763152	G	A	13041	missense_variant	MODERATE	TKFC
chr11	63095960	rs184268615	T	A	4177.17	intron_variant	MODIFIER	SLC22A24
chr11	63671373	rs528988637	AGAG	A	5656.05	5_prime_UTR_variant	MODIFIER	ATL3
chr11	65035224	rs761464534	G	A	1165.01	intron_variant	MODIFIER	SNX15
chr11	66334581	rs780118610	C	A	4516.16	synonymous_variant	LOW	RIN1
chr11	66704877	rs144746828	C	T	14035	missense_variant	MODERATE	SPTBN2
chr11	68780769	rs545149834	A	G	11691	intron_variant	MODIFIER	CPT1A
chr11	69054039	rs150671549	C	T	5615.01	missense_variant	MODERATE	TPCN2
chr11	69773347	rs752416500	G	C	10430	missense_variant	MODERATE	FGF4
chr12	95062595	rs149368820	C	T	16630	synonymous_variant	LOW	NR2C1
chr19	39891610	rs2260134	A	T	9890.93	intron_variant	MODIFIER	FCGBP
chr19	39891613	rs78638893	C	T	9666.15	intron_variant	MODIFIER	FCGBP

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chr19	39906217	chrchr19bp3990	T	C	4204.93	synonymous_variant	LOW	FCGBP
chr19	39906221	rs782328768	C	T	4024.93	missense_variant	MODERATE	FCGBP
chr19	39906229	chrchr19bp3990	C	T	3394.93	synonymous_variant	LOW	FCGBP
chr19	39906242	rs782103783	T	C	9912.93	missense_variant	MODERATE	FCGBP
chr4	186628707	rs568366597	C	T	17546	synonymous_variant	LOW	FAT1

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HGVS _c	HGVS _p	MAX_AF	MAX_AF_P	V:10		V:11	
				GT	DP	GT	DP
		0.000318	gnomAD_S	1/1	83	1/1	100
ENST00000359600.5:c.159C>A	ENSP00000352614.5:p.Val53%3D	3.46E-05	gnomAD_S	1/1	136	1/1	189
ENST00000334815.4:c.527T>C	ENSP00000335060.3:p.Leu176Pro	3.28E-05	gnomAD_S	1/1	54	1/1	62
ENST00000396478.3:c.880+17A>G		7.23E-05	gnomAD_S	1/1	31	1/1	53
ENST00000371992.1:c.1539+116G>C		0.001	EUR&SAS	1/1	57	1/1	51
ENST00000576409.5:c.792T>C	ENSP00000460964.1:p.Tyr264%3D	0.000163	gnomAD_S	1/1	80	1/1	127
ENST00000354831.11:c.1182G>A	ENSP00000346890.7:p.Lys394%3D	0.002813	gnomAD_S	1/1	79	1/1	88
ENST00000361249.3:c.960G>T	ENSP00000354458.3:p.Glu320Asp	0.006205	gnomAD_S	1/1	187	1/1	234
ENST00000620612.5:c.1878-39C>T		0.004572	gnomAD_E	1/1	55	1/1	118
ENST00000583931.5:c.205-19dup		0.000235	AA	1/1	40	1/1	49
ENST00000569591.2:c.1186G>A	ENSP00000455897.1:p.Gly396Ser	0.000138	gnomAD_A	1/1	109	1/1	139
ENST00000395526.8:c.1371A>G	ENSP00000378897.3:p.Ala457%3D	0.000129	gnomAD_S	1/1	214	1/1	231
		0.0051	SAS	1/1	126	1/1	154
ENST00000621158.4:c.1132-20C>T		0.0041	SAS	1/1	47	1/1	45
ENST00000311907.9:c.798C>T	ENSP00000308541.5:p.Asp266%3D	0.0051	SAS	1/1	98	1/1	144
ENST00000641251.1:c.223G>A	ENSP00000493334.1:p.Val75Ile	0.0051	SAS	1/1	102	1/1	107
ENST00000378397.1:c.549T>C	ENSP00000367650.1:p.Pro183%3D	0.0051	SAS	1/1	55	1/1	64
ENST00000394900.7:c.683G>A	ENSP00000378360.3:p.Arg228Gln	0.003	EUR	1/1	129	1/1	167
ENST00000612278.4:c.1070+31A>T		0.003392	gnomAD_S	1/1	48	1/1	74
ENST00000398868.7:c.-41_-39del		0.0072	SAS	1/1	36	1/1	55
ENST00000377244.7:c.520+18G>A		0.000265	gnomAD_S	1/1	16	1/1	13
ENST00000311320.8:c.1218G>T	ENSP00000310406.4:p.Leu406%3D	0.000272	gnomAD_S	1/1	41	1/1	59
ENST00000533211.5:c.2399G>A	ENSP00000432568.1:p.Arg800Gln	0.000233	EA	1/1	121	1/1	157
ENST00000265641.9:c.1353-24T>C		0.0031	SAS	1/1	142	1/1	175
ENST00000294309.7:c.116C>T	ENSP00000294309.3:p.Ala39Val	0.00871	gnomAD_S	1/1	63	1/1	56
ENST00000168712.2:c.583C>G	ENSP00000168712.1:p.Pro195Ala	0.000195	gnomAD_S	1/1	76	1/1	141
ENST00000333003.9:c.198G>A	ENSP00000333275.4:p.Pro66%3D	0.003092	gnomAD_N	1/1	197	1/1	249
ENST00000616721.5:c.3603+33T>A		0.005882	gnomAD_C	1/1	31	1/1	37
ENST00000616721.5:c.3603+30G>A		0.005373	gnomAD_C	1/1	36	1/1	41

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ENST00000616721.5:c.107A>G	ENSP00000481056.2:p.Glu36%3D	0.000154	gnomAD_A	1/1	11	1/1	12
ENST00000616721.5:c.103G>A	ENSP00000481056.2:p.Arg35His	0.007639	gnomAD_N	1/1	10	1/1	11
ENST00000616721.5:c.95G>A	ENSP00000481056.2:p.Ser32%3D	0.000501	gnomAD_E	1/1	9	1/1	9
ENST00000616721.5:c.82A>G	ENSP00000481056.2:p.Glu28Gly	9.88E-05	gnomAD_E	1/1	36	1/1	46
ENST00000441802.6:c.4380G>A	ENSP00000406229.2:p.Lys1460%3D	0.001	SAS	1/1	273	1/1	279

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