Figure S1: Schematic of Genotyping Results across Mouse Chromosome 6 in Affected Mice

Black squares are indicative of a C57BL/6J homozygous genotype; light grey squares, a 129x1/SvJ homozygous genotype; grey squares, a C57BL/6J 129x1/SvJ heterozygous genotype; white squares, undetermined genotype. The black box bounds a region of homozygous 129x1/SvJ genotypes that segregate with disease; thus, the critical region was determined to be between markers D6Mit37 and 44.MMHAP85FLG5.

Figure S2: Sequence of Exon 36 of Itpr1 from Four Mice

A wild-type homozygous C57BL/6J mouse (A), a wild-type homozygous 129x1/SvJ mouse (B), an affected 129x1B6 mice homozygous for the 18-bp deletion mutation (C), and an unaffected mouse heterozygous for the 18-bp deletion mutation (D). The deleted nucleotides are bounded by a green box.

Figure S3: Additional Families Harboring Deletion at the SCA15 Locus

(A) Family H33; (B) family H27. Upper panel shows log R ratio and B allele frequency metrics generated from Infinium HumanHap550 arrays for an affected family member from each family. Log R ratio is the ratio of normalized, observed R to expected R for each SNP (each SNP is a blue dot) and thus serves as a surrogate of copy number at each locus. B allele frequency is a measure of the number of times the A or B alleles are detected at each locus (each SNP is denoted by a blue dot). Thus, SNPs with a B allele frequency of one are apparent B/B homozygotes, SNPs with a B allele frequency of 0.5 are apparent A/B heterozygotes, and those with a B allele frequency of zero are apparent A/A homozygotes. These plots show a contiguous region ~310 kb long (family H33) and ~350 kb long (family H27) with decreased copy number and apparent homozygosity indicative of a genomic deletion (shaded grey). The pedigrees below show the available family members assayed for these deletions, all of whom were affected and all of whom carried a deletion at this locus.

Figure S4: Deleted Regions Identified in Families AUS1 and H27

(Top) Family AUS1; sequence from the PCR product generated using primers T3f and C11r from genomic DNA from an affected family member. Red arrowhead denotes the deletion breakpoint; the deletion is 201,510 bp in length.

(Bottom) Family H27; sequence flanking deleted region. Green font indicates nucleotides telomeric to the deletion; blue font indicates nucleotides centromeric to the deletion. The deletion is 344,408 bp in length. Basepair positions are based on NCBI genome build 36 reference assembly.