

Fig. S1

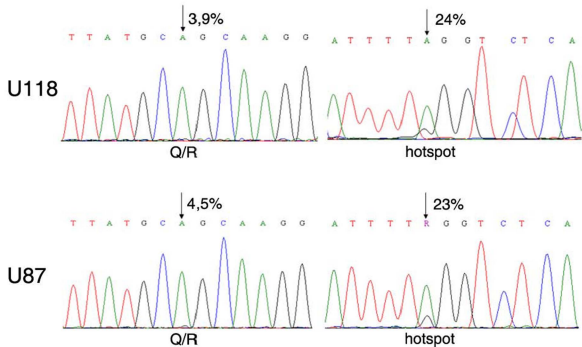


Fig. S2

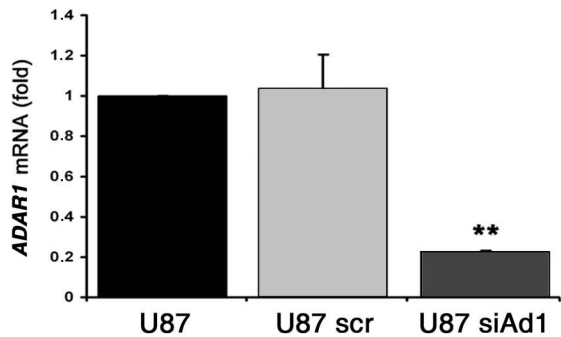
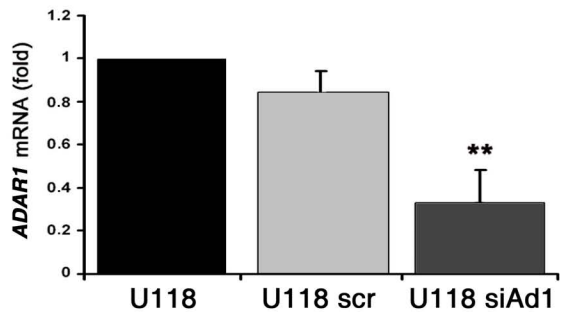
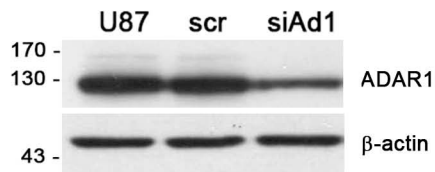
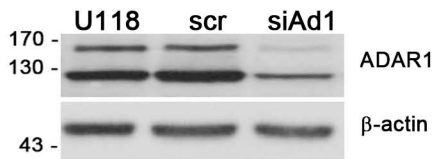
**A****B**

Fig. S3

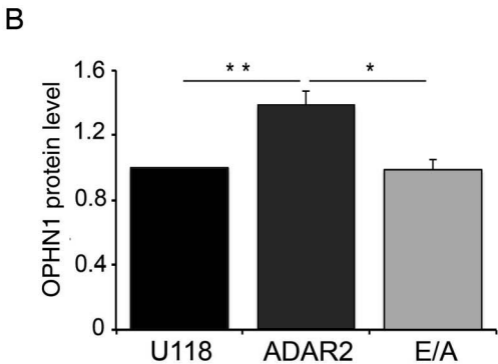
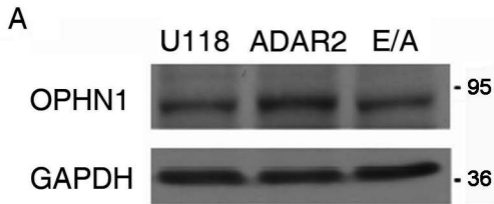


Fig. S4

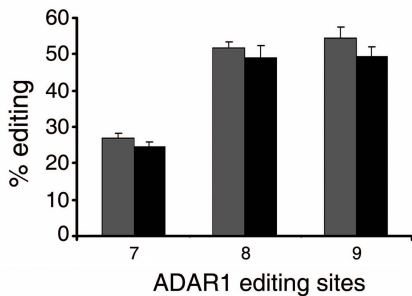
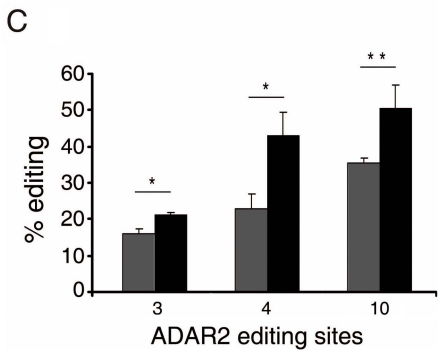
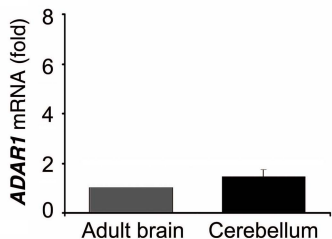
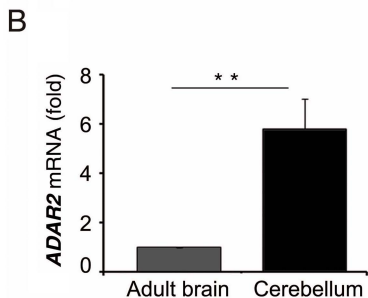
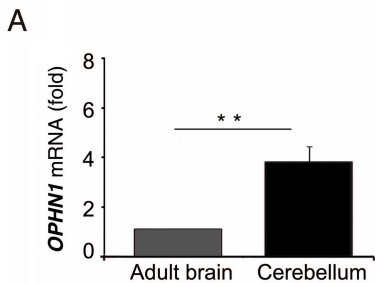


Fig. S5

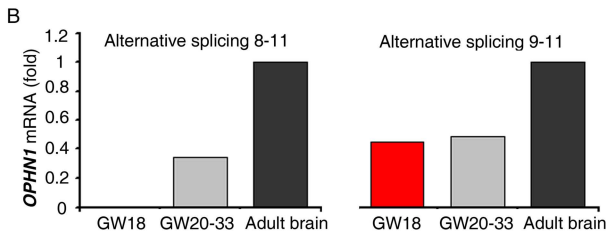
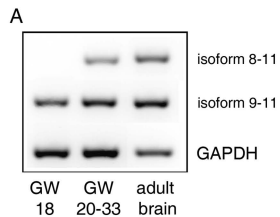


Fig. S6

**Table S1**

<b>OPHN1 - gDNA</b>	<b>Forward</b>	<b>Reverse</b>
Ex2 gDNA	5'-gtgcattgatgetctcgg-3'	5'-atttgcctcagagatcggg-3'
Ex3 gDNA	5'-cctgctcagagcacttacc-3'	5'-ccaagtgctctgtatccaca-3'
Ex4 gDNA	5'-gtctgtcaagcaccatgatta-3'	5'-gcaccaagaagctatctgac-3'
Ex5 gDNA	5'-aggaatcttgtatctgaac-3'	5'-gtcatgttagctgaggctac-3'
Ex6 gDNA	5'-gacagagttacaactacagac-3'	5'-ctgtttctaaggcatag-3'
Ex7 gDNA	5'-tggaccagaaactggaatcc-3'	5'-ttccactgattatccttcacg-3'
Ex8 gDNA	5'-agggattacaggcataagcc-3'	5'-ttggataggagttagtaacc-3'
Ex9 gDNA	5'-tgcacatgctgtaagcaggg-3'	5'-aattcactgccgacaagtgg-3'
Ex10 gDNA	5'-gactctgaagagcaatgtgta-3'	5'-atggcctcacgtaacagtga-3'
Ex11 gDNA	5'-gacactgaagacactgtagg-3'	5'-ggacattgccaatttcacagc-3'
Ex12 gDNA	5'-tctgtaagcctatgctgttc-3'	5'-actcatacactggcatatgct-3'
Ex13 gDNA	5'-cgctggcctgttcaagta-3'	5'-cccatattcagacttgcgttag-3'
Ex14 gDNA	5'-ttaataggtgacacctgttc-3'	5'-gcttgcataccactagtttag-3'
Ex15 gDNA	5'-atttgagagatgctgtagg-3'	5'-cacatttagcagtgtctcc-3'
Ex16 gDNA	5'-cagaccagaatgcagtcag-3'	5'-cacgttaactccgctcaaca-3'
Ex17 gDNA	5'-agaatccaataccagctgc-3'	5'-cttgagttagacagcaagcaa-3'
Ex18 gDNA	5'-gctcaatccagttgccag-3'	5'-ttcctcacagagcattcagc-3'
Ex19 gDNA	5'-ttgtaatcttgcttccaatac-3'	5'-ggaagacaggtagttagaata-3'
Ex20 gDNA	5'-ctagatataaggccaacaag-3'	5'-tggcatgccaccgcttct-3'
Ex21 gDNA	5'-tgccacagtacataactg-3'	5'-ctcagaaggatctcaagg-3'
Ex22 gDNA	5'-agccagagttgttagtcacc-3'	5'-gcctagtacaggacttcag-3'
Ex23 gDNA	5'-attggttctcacgtgtatacc-3'	5'-tgtatgctaaggatgcagttat-3'
Ex24 gDNA	5'-tagcactggcctagaagtgt-3'	5'-cgtaatgaaggcttatgtgga-3'

<b>OPHN1 - cDNA</b>	
cDNA-1Fw	5'-actgcgatagttagttagctctcc-3'
cDNA-1Rev	5'-ccgctccttgggtgaag-3'
cDNA-2Fw	5'-ttgctgattaaaccttggaaaa-3'
cDNA-2Rev	5'-acatctctgggcatttagg-3'
cDNA-3Fw	5'-agaacctatctaccacagcc-3'
cDNA-3Rev	5'-agtttcccactcctctgaa-3'
cDNA-4Fw	5'-ccctggatgaaagcgaagat-3'
cDNA-4Rev	5'-cttcagaacctggaaaggg-3'
AluJo Fw	5'-acataaggcactaaggctc-3'
AluJo Rev	5'-gcctgagcaacatagtgaga-3'
AluSZ Fw	5'-aagtgatgggagagatagac-3'
AluSZ Rv	5'-cacaacctgcttactca-3'
AluJo 2Fw	5'-cccaactacaagtgtcagg-3'
AluJo 2 Rev	5'-aacttagccaagtgtagtgg-3'

<b>OPHN1 - splicing</b>	
Splice exons 8-11 Fw	5'-cagtttacagaatgggcct-3'
Splice exons 9-11 Fw	5'-aagagaaatgggccttggga-3'
Splice 12-13 Rev	5'-ggctgtgtagatggttct-3'
Exon 8 Fw	5'-agcctgttggccttct-3'
Exon 11 Rev	5'-ggttctggccttctcattag-3'