

Supplemental Data

American Journal of Human Genetics, Volume 88

**Adaptor Protein Complex 4 Deficiency Causes Severe
Autosomal-Recessive Intellectual Disability, Progressive
Spastic Paraplegia, Shy Character, and Short Stature**

**Rami Abou Jamra, Orianne Philippe, Annick Raas-Rothschild, Sebastian H.
Eck, Elisabeth Graf, Rebecca Buchert, Arif Ekici, Felix F. Brockschmidt, Markus
M. Nöthen, Arnold Munnich, Tim M. Strom, Andre Reis, and Laurence Colleaux**

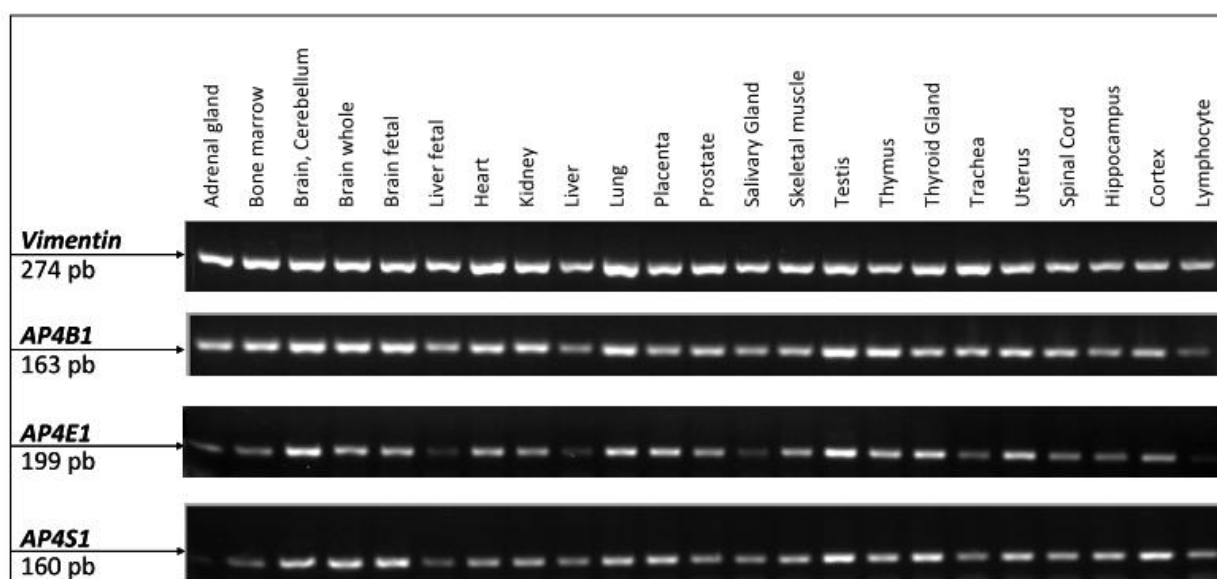


Figure S1. Expression Profile of the AP4 Genes

Expression analysis of *AP4B1*, *AP4E1*, *AP4S1* and Vimentin. Expression was assayed by RT-PCR on adult and fetal total RNA isolated from various tissues (Clontech). Amplicon lengths are indicated in bp.

Table S1. Summary of the Linkage Results

	candidate region	start SNP	end SNP	start bp	end bp	LOD score	length mb	#RefSeq genes*	identified mutation
ID01	1p13.2q21.2	rs692982	rs1868992	112,380,534	149,908,108	2.66	37.5	150	<i>AP4B1</i> ;NM_006594.2:c.487_488insTAT;p.Glu163_Ser739delinsVal
MR061	14q11-q12	rs10132585	rs1278951	22,399,285	31,493,380	3.85	9.1	94	<i>AP4S1</i> ;NM_007077.3:c.124C>T, NP_009008.2:p.Arg42*
MR071	15q21.1-q25.1	rs28464106	rs1867315	44,082,523	77,056,789	3.32	33.0	290	<i>AP4E1</i> ;NM_007347.3:c.542+1_542+4delGTAA, r.543_702del

* according to UCSC