

G. References.

- Banfi, S., Chung, M.Y., Kwiatkowski, T.J. Jr , Ranum, L.P., McCall, A.E., Chinault, A.C., Orr, H.T., Zoghbi, H.Y. (1993).** Mapping and cloning of the critical region for the spinocerebellar ataxia type 1 gene (SCA1) in a yeast artificial chromosome contig spanning 1.2 Mb. *Genomics* Dec; 18 (3): 627-35.
- Banfi, S., Servadio, A., Chung, M.Y., Kwiatkowski, T.J. Jr, McCall, A.E., Duvick, L.A., Shen, Y., Roth, E.J., Orr, H.T., Zoghbi, H.Y. (1993)** Identification and characterization of the gene causing type 1 spinocerebellar ataxia. *Nature Genet.* 7, 513-520.
- Barbeau, A., Roy, M., Cunha, L., de Vincente, A. N., Rosenberg, R. N., Nyhan, W. L., MacLeod, P. L., Chazot, G., Langston, L. B., Dawson, D., Coutinho, P. (1984).** The natural history of Machado-Joseph disease: An analysis of 138 personally examined cases. *Canad. J. Neurol. Sci.* 11: 510-525.
- Birnboim, H.C., Doly, J.A. (1979).** Rapid alkaline extraction procedure for screening recombinant plasmid DNA. *Nucleic Acids Res.* Nov 24; 7 (6): 1513-23.
- Boutell, J.M., Wood, J.D., Harper, P.S., Jones, A.L. (1998).** Huntingtin interacts with cystathionine beta-synthase. *Hum. Mol. Genet.* Mar, 7 (3): 371-8.
- Burke, J.R., Enghild, J.J., Martin, M.E., Jou, Y.S., Myers, R.M., Roses, A.D., Vance, J.M., Strittmatter, W.J. (1996).** Huntingtin and DRPLA proteins selectively interact with the enzyme GAPDH. *Nature Med.* Mar, 2 (3): 347-50.
- Büssow, K., Cahill, D., Nietfeld, W., Bancroft, D., Scherzinger, E., Lehrach, H., Walter, G., (1998)** A method for global protein expression and antibody screening on high-density filters of an arrayed cDNA library. *Nucleic Acids Res.*, 26, 5007-5008.

Cancel, G., Abbas, N., Stevanin, G., Dürr, A., Chneiweiss, H., Neri, C., Duyckaerts, C., Penet, C., Cann, H. M., Agid, Y., Brice, A. (1995). Marked phenotypic heterogeneity associated with expansion of a CAG repeat sequence at the spinocerebellar ataxia3/Machado-Joseph disease locus. *Am. J. Hum. Genet.* 57: 809-816.

Celis, J.E. (1994) Cell Biology. A laboratory handbook. Academic Press.

Chai, Y., Koppenhafer, S.L., Shoesmith, S.J., Perez, M.K., Paulson, H.L. (1999). Evidence for proteasome involvement in polyglutamine disease: Localization to nuclear inclusions in SCA3/MJD and suppression of polyglutamine aggregation in vitro. *Hum. Mol. Genet.* Apr; 8(4): 673-82.

Conner, K.E. and Rosenberg, R. N. (1993). The hereditary ataxias. In: " The Molecular and Genetic Basis of Neurological Disease " (R. N. Rosenberg, S., Prusiner, S., Di Mauro, R. L., Barchi, and L. M. Kunkel, Eds.), pp. 697-737. Butterworth-Heinemann, Boston.

Coutinho, P., Andrade, C. (1978). Autosomal dominant system degeneration in Portuguese families of the Azores Islands: A new genetic disorder involving cerebellar, pyramidal, extrapyramidal and spinal cord motor functions. *Neurology* 28: 703-709, 1978.

DiFiglia, M., Sapp, E., Chase, K.O., Davies, S.W., Bates, G.P., Vonsattel, J.P. and Aronin, N. (1997). Aggregation of Huntingtin in neuronal intranuclear inclusions and dystrophic neurites in brain. *Science* 277, 1990-1993.

Duyao, M., Ambrose, C., Myers, R., Novelletto, A., Persichetti, F., Frontali, M., Folstein, S., Ross, C., Franz, M., Abbott, M., Gray, J., Conneally, P., Young, A., Penney, J., Hollingsworth, Z., Shoulson, I., Lazzarini, A., Falek, A., Koroshetz, W., Sax, D., Bird, E., Vonsattel, J., Bonilla, E., Alvir, J., Conde, J., Cha, J.H., Dure, L., Gomez, F., Ramos, M., Sanchez-Ramos, J., Snodgrass, S., de Young, M., Wexler, N., Moscovitz, C., Penchaszadeh, G.,

- MacFarlane, H., Anderson, M., Jenkins, B., Srinidhi, J., Barnes, G., Gusella, J., and MacDonald, M. (1993).** Trinucleotide repeat length instability and age of onset in Huntington`s disease. *Nature Genet.* 4, 387-392.
- Dürr, A., Stevanin, G., Cancel, G., Duyckaerts, C., Abbas, N., Didierjean, O., Chneiweiss, H., Benomar, A., Lyon-Caen, O., Julien, J., Serdaru, Penet, C., Agid, Y., Brice, A. (1995).** Spinocerebellar ataxia 3 and Machado-Joseph disease: Clinical, molecular, and neuropathological features. *Ann. Neurol.* Apr; 39 (4): 490-9.
- Evert, B.O., Wullner, U., Schulz, J.B., Weller, M., Groscurth, P., Trottier, Y., Brice, A., Klockgether, T. (1999).** High level expression of expanded full-length ataxin-3 in vitro causes cell death and formation of intranuclear inclusions in neuronal cells. *Hum. Mol. Genet.* Jul; 8(7): 1169-76.
- Fey, E.G., Wan, K.M. and Penman, S. (1984).** Epithelial cytoskeletal framework and nuclear matrix intermediate filaments scaffold: Three dimensional organization and protein composition. *J. Cell. Biol.* 98, 1973-1984.
- Giunti, P., Sweeney, M. G., Harding, A. E. (1995).** Detection of the Machado-Joseph disease/spinocerebellar ataxia three trinucleotide repeat expansion in families with autosomal dominant motor disorders, including the Drew family of Walworth. *Brain* 118: 1077-1085.
- Goldberg, Y.P., Nicholson, D.W., Rasper, D.M., Kalchman, M.A., Koide, H.B., Graham, R.K., Bromm, M., Kazemi-Esfarjani, P., Thornberry, N.A., Vaillancourt, J.P., Hayden, M.R. (1996).** Cleavage of huntingtin by apopain, a proapoptotic cysteine protease, is modulated by the polyglutamine tract. *Nature Genet.* Aug; 13 (4): 42-9.

- Goldberg-Stern, H., D'jarldetti, R., Melamed, E., Gadoth, N. (1994).** Machado-Joseph (Azorean) disease in a Yemenite Jewish family in Israel. *Neurology* 44: 1298-1301.
- Görlich, D. (1997).** Nuclear protein import. *Current Opinion in Cell Biology* 9: 412-419.
- Gorman, C.M., Lane, D.M. and Rigby, P.W. (1984).** High efficiency gene transfer into mammalian cells. *Philos. Trans. R. Soc. Lond. Biol.*, 307, 343-346.
- Goto, J., Watanabe, M., Ichikawa, Y., Yee, SB., Ihara, N., Endo, K., Igarashi, S., Takiyama, Y., Gaspar, C., Maciel, P., Tsuji S., Rouleau, G.A., Kanazawa, I. (1997).** Machado-Joseph disease gene products carrying different carboxyl termini. *Neuroscience Research* 28 373-377.
- Green, H. (1993).** Human genetic diseases due to codon reiteration: Relationship to an evolutionary mechanism. *Cell* 74: 955-6.
- Gu, J., Stephenson, C.G., Iadarola, M.J. (1994)** Recombinant proteins attached to a nickel-NTA column: Use in affinity purification of antibodies. *BioTech.* Vol. 17, No 2, 257-262.
- Healton, E. B., Brust, J. C. M., Kerr, D. L., Resor, S., Penn, A. (1980).** Presumably Azorean disease in a presumably non-Portuguese family. *Neurology* 30: 1084-1089.
- Hilbich, C., Kisters-Woike, B., Reed, J., Masters, C.L., Beyreuther, K. (1991).** Aggregation and secondary structure of synthetic amyloid beta A4 peptides of Alzheimer's disease. *J. Mol. Biol.* Mar 5; 218 (1): 149-63
- The Huntington's Disease Collaborative Research Group. (1993).** A novel gene containing a trinucleotide repeat that is expanded and unstable on Huntington's disease chromosomes. *Cell* Mar 26; 72 (6): 971-83.

Igarashi, S., Koide, R., Shimohata, T., Yamada, M., Hayashi, Y., Takano, H., Date, H., Oyake, M., Sato, T., Sato, A., Egawa, S., Ikeuchi, T., Tanaka, H., Nakano, R. (1998) Suppression of aggregate formation and apoptosis by transglutaminase inhibitors in cells expressing truncated DRPLA protein with an expanded polyglutamine stretch. *Nature Genet.* Feb; 18 (2): 111-7.

Ikeda, H., Yamaguchi, M., Sugai, S., Aze, Y., Narumiya, S., Kakizuka, A. (1996). Expanded polyglutamine in the Machado-Joseph disease protein induces cell death in vitro and in vivo. *Nature Genet.* Jun; 20 (6): 649-655.

Ikeuchi, T., Igarashi, S., Takiyama, Y., Onodera, O., Oyake, M., Takano, H., Koide, R., Tanaka, H., Tsuji, S. (1996). Non-mendelian transmission in dentatorubral-pallidoluysian atrophy and Machado-Joseph disease: The mutant allele is preferentially transmitted in male meiosis. *Am. J. Hum. Genet.* 58: 730-733.

Ishino, H., Sata, M., Mii, T., Terao, A., Hayahara, T., Otsuki, S., Hoaki, T. (1971). An autopsy case of Marie's hereditary ataxia. *Psychiat. Neurol. Jpn.* 73: 747-757.

Jarrett, J.T., Lansbury P.T. (1993). Seeding "one-dimensional crystallization" of amyloid: A pathogenic mechanism in Alzheimer's disease and scrapie? *Cell* Jun 18; 73 (6): 1055-1058.

Jodice, C., Malaspina, P., Persichetti, F., Novelletto, A., Spadaro, M., Giunti, P., Morocutti, C., Terrenato, L., Harding, A.E., Frontali, M. (1994). Effect of trinucleotide repeat length and parental sex on phenotypic variation in spinocerebellar ataxia I. *Am. J. Hum. Genet.* Jun; 54 (6): 959-965.

Kahlem, P., Green, H., Djian, P. (1998). Transglutaminase action imitates Huntington's disease: Selective polymerization of Huntingtin containing expanded polyglutamine. *Mol. Cell* Mar; 1 (4): 595-601.

Kakizuka, A., (1998) Protein precipitation: A common etiology in neurodegenerative disorders? *Trends in Genetics* Vol. 14, No 10, 396-402.

Kalchman, M.A., Koide, H.B., McCutcheon, K., Graham, R.K., Nichol, K., Nishiyama, K., Kazemi-Esfarjani, P., Lynn, F.C., Wellington, C., Metzler, M., Goldberg, Y.P., Kanazawa, I., Gietz, R.D., Hayden, M.R. (1997). HIP1, a human homologue of *S. cerevisiae* Sla2p, interacts with membrane-associated huntingtin in the brain. *Nature Genet.* May; 16 (1): 44-53.

Karpuj, M.V., Garren, H., Slunt, H., Price, D.L., Gusella, J., Becher, M.W., Steinman, L. (1999). Transglutaminase aggregates huntingtin into nonamyloidogenic polymers, and its enzymatic activity increases in Huntington's disease brain nuclei. *Proc. Natl. Acad. Sci. U S A* Jun 22; 96 (13): 7388-93.

Kawaguchi, Y., Okamoto, T., Taniwaki, M., Aizawa, M., Inoue, M., Katayama, S., Kawakami, H., Nakamura, S., Nishimura, M., Akiguchi, I., Kimura, J., Narumiya, S., Kakizuka, A. (1994). CAG expansions in a novel gene for Machado-Joseph disease at chromosome 14q32.1. *Nature Genet.* 8: 221-228.

Kawakami, H., Maruyama, H., Nakamura, S., Kawaguchi, Y., Kakizuka, A., Doyu, M., Sobue, G. (1995). Unique features of the CAG repeats in Machado-Joseph disease. (Letter) *Nature Genet.* 9: 344-345.

Kaytor, M.D., Duvick, L.A., Skinner, P.J., Koob, M.D., Ranum, L.P., Orr, H.T. (1999). Nuclear localization of the spinocerebellar ataxia type 7 protein, ataxin-7. *Hum. Mol. Genet.* Sep; 8 (9): 1657-1664.

Koide, R., Ikeuchi, T., Onodera, O., Tanaka, H., Igarashi, S., Endo, K., Takahashi, H., Kondo, R., Ishikawa, A., Hayashi, T., Saito, M., Tomoda, A., Miike, T., Naito, H., Ikuta, F., and Tsuji, S. (1994). Unstable expansion of

- CAG repeat in hereditary dentatorubral-pallidoluysian atrophy (DRPLA). *Nature Genet.* 6, 14-18.
- Koshy, B., Matilla, T., Burright, E.N., Merry, D.E., Fischbeck, K.H., Orr, H.T., Zoghbi, H.Y. (1996).** Spinocerebellar ataxia type-1 and spinobulbar muscular atrophy gene products interact with glyceraldehyde-3-phosphate dehydrogenase. *Hum. Mol. Genet.* Sep; 5 (9): 1311-8.
- Kunkel, T.A. (1985).** Rapid and efficient site-specific mutagenesis without phenotypic selection. *Proc. Natl. Acad. Sci. U S A* Jan; 82 (2): 488-92.
- La Spada, A.R., Roling, D.B., Harding, A.E., Warner, C.L., Spiegel, R., Hausmanowa-Petrusewicz, I., Yee, W.C., Fischbeck, K.H. (1992).** Meiotic stability and genotype-phenotype correlation of the trinucleotide repeat in X-linked spinal and bulbar muscular atrophy. *Nature Genet.* Dec; 2 (4): 301-4.
- Lee, K.A.W., Bindereif, A., Green, M.R. (1988)** A small-scale procedure for preparation of nuclear extracts that support efficient transcription and pre-mRNA splicing. *Gene Anal. Techn.* 5: 22-31.
- Le Grice, S.F.J. and Grueninger-Leitch, F. (1990).** Rapid purification of homodimer HIV-1 reverse transcriptase by metal chelate affinity chromatography. *Eur. J. Biochem.* 187, 307-314.
- Li, X.J., Li, S.H., Sharp, A.H., Nucifora, F.C. Jr, Schilling, G., Lanahan, A., Worley, P., Snyder, S.H., Ross, C.A. (1995).** A huntingtin-associated protein enriched in brain with implications for pathology. *Nature* Nov 23; 378 (6555): 398-402.
- Linhartova, I., Repitz, M., Draber, P., Nemec, M., Wiche, G., Propst, F. (1999).** Conserved domains and lack of evidence for polyglutamine length polymorphism in the chicken homolog of the Machado-Joseph disease gene product ataxin-3. *Biochim. Biophys. Acta* Feb 16; 1444(2): 299-305.

Lopes-Cendes, I., Maciel, P., Kish, S., Gaspar, C., Robitaille, Y., Clark, H.B., Koeppen, A.H., Nance, M., Schut, L., Silveira, I., Coutinho, P., Sequeiros, J., Rouleau, G.A.(1996). Somatic mosaicism in the central nervous system in spinocerebellar ataxia type 1 and Machado-Joseph disease. *Ann. Neurol.* Aug; 40 (2): 199-206.

Lunkes, A., Mandel, J.L. (1997). Polyglutamines, nuclear inclusions and neurodegeneration. *Nature Med.* 11, 1201-1202.

Maniatis, T., Fritsch, E.F. and Sambrook, J. (1989). Molecular Cloning: A Laboratory Manual. 2nd edn. *Cold Spring Harbor Laboratory Press*, Cold Spring Harbor, NY.

Maraldi, N.M., Zini, N., Santi, S., Bavelloni, A., Valmori, A., Marmioli, S. and Ognibene, A. (1993). Phosphoinositidase C isozymes in SaOS-2 cells: Immunocytochemical detection in nuclear and cytoplasmic compartments. *Biol. Cell.* 79, 243-250.

Martindale, D., Hackam, A., Wieczorek, A., Ellerby, L., Wellington, C., McCutcheon, K., Singaraja, R., Kazemi-Esfarjani, P., Devon, R., Kim, S.U., Bredesen, D.E., Tufaro, F., Hayden, M.R. (1998). Length of huntingtin and its polyglutamine tract influences localization and frequency of intracellular aggregates. *Nature Genet.* Feb;18 (2): 150-4.

Matilla, A., Koshy, B.T., Cummings, C.J., Isobe, T., Orr, H.T., Zoghbi, H.Y. (1997). The cerebellar leucine-rich acidic nuclear protein interacts with . *Nature* Oct 30; 389 (6654): 974-8.

Nagafuchi, S., Yanagisawa, H., Sato, K., Shirayama, T., Ohsaki, E., Bundo, M., Takeda, T., Tadokoro, K., Kondo, I., Murayama, N., Tanaka, Y., Kikushima, H., Umino, K., Kurosawa, H., Furukawa, T., Nihei, K., Inoue, T., Sano, A., Komure, O., Takahashi, M., Yoshizawa, T., Kanazawa, I., and

- Yamada, M. (1994).** Dentatorubral and pallidoluysian atrophy expansion of an unstable CAG trinucleotide on chromosome 12p. *Nature Genet.* Jan; 6 (1): 14-8.
- Nakai, K. (1991).** Predicting various targeting signals in amino acid sequences. *Bull. Inst. Chem. Res.*, 69, 269-291.
- Nakai, K. and Kanehisa, M. (1992).** A knowledge base for predicting protein localization sites in eukaryotic cells. *Genomics* 14, 897-911.
- Nakamoto, M., Ikeda, H., Kakizuka, A. (1998)** Genetic and molecular studies of Machado-Joseph disease. Genetic instabilities and and hereditary neurological diseases. *Academic Press* 283-297.
- Nakano, K. K., Dawson, D. M., Spence, A. (1972).** Machado disease: A hereditary ataxia in Portuguese emigrants to Massachusetts. *Neurology* 22: 49-55.
- Onodera, O., Burke, J.R., Miller, S.E., Hester, S., Tsuji, S., Roses, A.D., Strittmatter, W.J. (1997).** Oligomerization of expanded-polyglutamine domain fluorescent fusion proteins in cultured mammalian cells. *Biochem. Biophys. Res. Commun.* Sep 18; 238 (2): 599-605.
- Orr, H.T., Chung, M.Y., Banfi, S., Kwiatkowski, T.J. Jr, Servadio, A., Beaudet, A.L., McCall, A.E., Duvick, L.A., Ranum, L.P., Zoghbi, H.Y. (1993).** Expansion of an unstable trinucleotide CAG repeat in spinocerebellar ataxia type 1. *Nature Genet.* Jul; 4 (3): 221-6.
- Paulson, H.L., Das, S.S., Crino, P.B., Perez, M.K., Patel, S.C., Gotsdiner, D., Fischbeck, K.H. and Pittman, R.N. (1997).** Machado-Joseph disease gene product is a cytoplasmic protein widely expressed in brain. *Ann. Neurol.* 41, 453-462.

Paulson, H.L., Perez, M.K., Trotter, Y., Trojanowski, J.Q., Subramony, S.H., Das, S.S., Vig P., Mandel J.L., Fischbeck, K.H. and Pittman R.N. (1997). Intranuclear inclusions of expanded polyglutamine protein in spinocerebellar ataxia type 3. *Neuron* 19, 333-344.

Perez, M.K., Paulson, H.L., Pendse, S.J., Saionz, S.J., Bonini, N.M., Pittman, R.N. (1998). Recruitment and the role of nuclear localization in polyglutamine-mediated aggregation. *J. Cell Biol.* Vol 143, No 6 1457-1470.

Perutz, M.F., Johnston, T., Suzuki, M., and Finch, J.T. (1994). Glutamine repeats as polar zippers: Their possible role in neurodegenerative diseases. *Proc. Natl. Acad. Sci. USA* 91, 5355-5358.

Perutz, M.F. (1996). Glutamine repeats and inherited neurodegenerative diseases: Molecular aspects. *Curr. Opin. Struct. Biol.* Dec; 6 (6): 848-58.

Prusiner, S.B.(1998). Prions. *Proc. Natl. Acad. Sci. U S A* Nov 10; 95 (23): 13363-83.

Ranum, L. P. W., Lundgren, J. K., Schut, L. J., Ahrens, M. J., Perlman, S., Aita, J., Bird, T. D., Gomez, C., Orr, H. T. (1995). Spinocerebellar ataxia type 1 and Machado-Joseph disease: Incidence of CAG expansions among adult-onset ataxia patients from families with dominant, recessive, or sporadic ataxia. *Am. J. Hum. Genet.* 57: 603-608.

Rihs, H.P., Jans, D.A., Fan, H. and Peters, R. (1991). The rate of nuclear cytoplasmic protein transport is determined by the casein kinase II site flanking the nuclear localization sequence of the SV40 T-antigen. *EMBO J.* 10, 633-639.

Romanul, F. C. A., Fowler, H. L., Radvany, J., Feldman, R. G., Feingold, M. (1977). Azorean disease of the nervous system. *New Eng. J. Med.* 296: 1505-1508.

Rosenberg, R. N. (1977). Azorean disease of the nervous system.(Letter) *New Eng. J. Med.* 297: 729.

Rosenberg, R. N. (1983). Dominant ataxias. In: Kety, S. S., Rowland, L. P., Sidman, R. L., Matthysse, S. W.: *Genetics of Neurological and Psychiatric Disorders.* New York: Raven Press (pub.).

Rosenberg, R.N. (1990). Autosomal dominant cerebellar phenotypes: The genotype will settle the issue. *Neurology* 40, 1329-1331.

Rosenberg, R.N., and Grossman, A. (1989). Hereditary ataxia In " Neurologic Clinics " (W.G. Johnson, Ed.), Vol. 1, pp. 25-36, Saunders, Philadelphia.

Rosenberg, R. N., Nyhan, W. L., Bay, C., Shore, P. (1976). Autosomal dominant striato-nigral degeneration: A clinical, pathologic and biochemical study of a new genetic disorder. *Neurology* 26 (8): 703-14.

Sakai, T., Ohta, M., Ishino, H. (1983). Joseph disease in a non-Portuguese family. *Neurology* 33: 74-80.

Scherzinger, E., Lurz, R., Turmaine, M., Mangiarini, L., Hollenbach, B., Hasenbank, R., Bates, G.P., Davies, S.W., Lehrach, H. and Wanker, E.E. (1997). Huntingtin-encoded polyglutamine expansions form amyloid-like protein aggregates in vitro and in vivo. *Cell* 90, 549-558.

Scherzinger, E., Sittler, A., Schweiger, K., Heiser, V., Lurz, R., Hasenbank, R., Bates, G.P., Lehrach, H., Wanker, E.E. (1999) Self-assembly of polyglutamine-containing huntingtin fragments into amyloid-like fibrils: Implications for Huntington's disease pathology. *Proc. Natl. Acad. Sci. U S A* 13;96(8):4604-9.

Schmitt, I., Brattig, T., Gossen, M., Riess, O. (1997). Characterization of the rat spinocerebellar ataxia type 3 gene. *Neurogenetics* 1:103-112.

Schmidt, T., Landwehrmeyer, G.B., Schmitt, I., Trottier, Y., Auburger, G., Laccone, F., Klockgether, T., Volpel, M., Epplen, J.T., Schols, L., Riess, O. (1998). An isoform of ataxin-3 accumulates in the nucleus of neuronal cells in affected brain regions of SCA3 patients. *Brain Pathol.* Oct; 8 (4): 669-79.

Sittler, A., Devys, D., Weber, C. and Mandel, J.L. (1996). Alternative splicing of exon 14 determines nuclear or cytoplasmic localisation of fmr1 protein isoforms. *Hum. Mol. Genet.* 5, 95-102.

Sittler, A., Walter, S., Wedemeyer, N., Hasenbank, R., Scherzinger, E., Eickhoff, H., Bates, G.P., Lehrach, H., Wanker, E.E. (1998). SH3GL3 associates with the Huntingtin exon 1 protein and promotes the formation of polyglutamine-containing protein aggregates. *Mol. Cell* Oct; 2 (4): 427-36.

Skinner, P.J., Koshy, B.T., Cummings, C.J., Klement, I.A., Helin, K., Servadio, A., Zoghbi, H.Y. and Orr, H.T. (1997). Ataxin-1 with an expanded glutamine tract alters nuclear matrix-associated structure. *Nature* 389, 971-974.

Smith, D.B., and Johnson, K.S. (1988). Single-step purification of peptides expressed in *Escherichia coli* as fusions with glutathione S-transferase. *Gene* 67, 31-40.

Snell, R.G., MacMillan, J.C., Cheadle, J.P., Fenton, I., Lazarou, L.P., Davies, P., MacDonald, M.E., Gusella, J.F., Harper, P.S., Shaw, D.J. (1993). Relationship between trinucleotide repeat expansion and phenotypic variation in Huntington's disease. *Nature Genet.* Aug; 4 (4): 393-7.

Sobue, G., Doyu, M., Nakao, N., Shimada, N., Mitsuma, T., Maruyama, H., Kawakami, H., and Nakamura, S. (1996). Homozygosity for Machado-

Joseph disease gene enhances phenotypic severity. *J. Neurol. Neurosurg. Psychiatry*. Mar; 60 (3): 354-6.

Stott, K., Blackburn, J.M., Butler, P.J., Perutz, M. (1995). Incorporation of glutamine repeats makes protein oligomerize: Implication for neurodegenerative diseases. *Proc. Natl. Acad. Sci. USA* 76: 4350-54.

Tait, D., Riccio, M., Sittler, A., Scherzinger, E., Santi, S., Ognibene, A., Maraldi, N.M., Lehrach, H., Wanker, E.E. (1998). Ataxin-3 is transported into the nucleus and associates with the nuclear matrix. *Hum. Mol. Genet.* Jun; 7 (6): 991-7.

Takiyama, Y., Igarashi, S., Rogaeva, E. A., Endo, K., Rogaev, E. I., Tanaka, H., Sherrington, R., Sanpei, K., Liang, Y., Saito, M., Tsuda, T.; Takano, H., Ikeda, M., Lin, C., Chi, H., Kennedy, J. L., Lang, A. E., Wherrett, J. R., Segawa, M., Nomura, Y., Yuasa, T., Weissenbach, J., Yoshida, Nishizawa, M., Kidd, K. K., Tsuji, S., St George-Hyslop, P. H. (1995). Evidence for inter-generational instability in the CAG repeat in the MJD1 gene and for conserved haplotypes at flanking markers amongst Japanese and Caucasian subjects with Machado-Joseph disease. *Hum. Mol. Genet.* 4: 1137-1146.

Takiyama, Y., Oyanagi, S., Kawashima, S., Sakamoto, H., Saito, K., Yoshida, M., Tsuji, S., Mizuno, Y., Nishizawa, M. (1994). A clinical and pathologic study of a large Japanese family with Machado-Joseph disease tightly linked to the DNA markers on chromosome 14q. *Neurology* 44: 1302-1308.

Trottier, Y., Cancel, G., An-Gourfinkel, I., Lutz, Y., Weber, C., Brice, A., Hirsch, E., Mandel, J.L. (1998). Heterogeneous intracellular localization and expression of ataxin-3. *Neurobiol. Dis.* Nov; 5 (5): 335-47.

Trottier, Y., Lutz, Y., Stevanin, G., Imbert, G., Devys, D., Cancel, G., Saudou, F., Weber, C., David, G., Tora, L. (1995). Polyglutamine expansion as a

pathological epitope in Huntington's disease and four dominant cerebellar ataxias. *Nature* Nov 23; 378(6555): 403-6.

Wanker, E.E., Rovira, C., Scherzinger, E., Hasenbank, E., Walter, S., Tait, D., Colicelli, J. and Lehrach, H. (1997). HIP-1: A huntingtin interacting protein isolated by the two hybrid system. *Hum. Mol. Genet.* 6, 487-495.

Warner, T.T., Williams, L.D., Walker, R.W., Flinter, F., Robb, S.A., Bunday, S.E., Honavar, M., Harding A.E. (1995). A clinical and molecular genetic study of dentatorubropallidoluysian atrophy in four European families. *Ann. Neurol.* Apr; 37 (4): 452-9.

Warrick, J. M., Paulson, H. L., Gray-Board, G. L., Bui, Q. T., Fischbeck, K. H., Pittman, R. N., Bonini, N. M. (1998). Expanded polyglutamine protein forms nuclear inclusions and causes neural degeneration in *Drosophila*. *Cell* 93: 939-949.

Wellington, C.L., Ellerby, L.M., Hackam, A.S., Margolis, R.L., Trifiro, M.A., Singaraja, R., McCutcheon, K., Salvesen, G.S., Propp, S.S., Bromm, M., Rowland, K.J., Zhang, T., Rasper, D., Roy, S., Thornberry, N., Pinsky, L., Kakizuka, A., Ross, C.A., Nicholson, D.W., Bredesen, D.E., Hayden, M.R. (1998). Caspase cleavage of gene products associated with triplet expansion disorders generates truncated fragments containing the polyglutamine tract. *J. Biol. Chem.* Apr 10; 273 (15): 9158-67.

Wells, R.D. (1996). Molecular basis of genetic instability of triplet repeats. *J. Biol. Chem.* 271, No 6, 2875-2878.

Woods, B. T., Schaumburg, H. H. (1972). Nigro-spino-dentatal degeneration with nuclear ophthalmoplegia: A unique and partially treatable clinico-pathological entity. *J. Neurol. Sci.* 17: 149-166.

Yvert, G., Mandel, J.L. (1999). Variation on a trinucleotide theme.

Nature Med. Apr;5(4): 383-4.

Zuber. M., Heyden. T.S., Lajous-Petter. A.M. (1995). A human autoantibody recognizing nuclear matrix-associated nuclear protein localized in dot structures. *Biol. Cell* 85(1): 77-86.