

Bibliographie

- Ahn, V.E., Faull, K.F., Whitelegge, J.P., Fluharty, A.L. und Prive, G.G.** (2003) Crystal structure of saposin B reveals a dimeric shell for lipid binding. *Proc. Natl. Acad. Sci. U. S. A.*, **100**, 38-43.
- Anderson, D.H., Sawaya, M.R., Cascio, D., Ernst, W., Modlin, R., Krensky, A. und Eisenberg, D.** (2003) Granulysin crystal structure and a structure-derived lytic mechanism. *J. Mol. Biol.*, **325**, 355-365.
- Arndt, U.W. und Wonnacott, A.J.** (1977) *The Rotation Method in Crystallography*. North Holland, Amsterdam.
- Banerjee, P., Boyers, M.J., Berry-Kravis, E. und Dawson, G.** (1994) Preferential beta-hexosaminidase (Hex) A (alpha beta) formation in the absence of beta-Hex B (beta beta) due to heterozygous point mutations present in beta-Hex beta-chain alleles of a motor neuron disease patient. *J. Biol. Chem.*, **269**, 4819-4826.
- Banerjee, P., Siciliano, L., Oliveri, D., McCabe, N.R., Boyers, M.J., Horwitz, A.L., Li, S.C. und Dawson, G.** (1991) Molecular basis of an adult form of beta-hexosaminidase B deficiency with motor neuron disease. *Biochem. Biophys. Res. Commun.*, **181**, 108-115.
- Banner, D.W., Bloomer, A.C., Petsko, G.A., Phillips, D.C., Pogson, C.I., Wilson, I.A., Corran, P.H., Furth, A.J., Milman, J.D., Offord, R.E., Priddle, J.D. und Waley, S.G.** (1975) Structure of chicken muscle triose phosphate isomerase determined crystallographically at 2.5 angstrom resolution using amino acid sequence data. *Nature*, **255**, 609-614.
- Baric, I., Fumic, K. und Hoffmann, G.F.** (2001) Inborn errors of metabolism at the turn of the millennium. *Croat. Med. J.*, **42**, 379-383.
- Barton, G.J.** (1993) ALSCRIPT: a tool to format multiple sequence alignments. *Protein Eng.*, **6**, 37-40.
- Bierfreund, U., Lemm, T., Hoffmann, A., Uhlhorn-Dierks, G., Childs, R.A., Yuen, C.T., Feizi, T. und Sandhoff, K.** (1999) Recombinant GM2-activator protein stimulates in vivo degradation of GA2 in GM2 gangliosidosis AB variant fibroblasts but exhibits no detectable binding of GA2 in an in vitro assay. *Neurochem. Res.*, **24**, 295-300.
- Bolhuis, P.A., Ponne, N.J., Bikker, H., Baas, F. und Vianney de Jong, J.M.** (1993) Molecular basis of an adult form of Sandhoff disease: substitution of glutamine for arginine at position 505 of the beta-chain of beta-hexosaminidase results in a labile enzyme. *Biochim. Biophys. Acta*, **1182**, 142-146.
- Breiter, D.R., Kanost, M.R., Benning, M.M., Wesenberg, G., Law, J.H., Wells, M.A., Rayment, I. und Holden, H.M.** (1991) Molecular structure of an apolipoprotein determined at 2.5-A resolution. *Biochemistry (Mosc.)*, **30**, 603-608.

- Brunger, A.T., Adams, P.D., Clore, G.M., DeLano, W.L., Gros, P., Grosse-Kunstleve, R.W., Jiang, J.S., Kuszewski, J., Nilges, M., Pannu, N.S., Read, R.J., Rice, L.M., Simonson, T. und Warren, G.L.** (1998) Crystallography & NMR system: A new software suite for macromolecular structure determination. *Acta Crystallogr. D.*, **54**, 905-921.
- Cashman, N.R., Antel, J.P., Hancock, L.W., Dawson, G., Horwitz, A.L., Johnson, W.G., Huttenlocher, P.R. und Wollmann, R.L.** (1986) N-acetyl-beta-hexosaminidase beta locus defect and juvenile motor neuron disease: a case study. *Ann. Neurol.*, **19**, 568-572.
- CCP4.** (1994) The CCP4 Suite: Programs for Protein Crystallography. *Acta Crystallogr. D.*, **50**, 760 -763.
- Ciaffoni, F., Salvioli, R., Tatti, M., Arancia, G., Crateri, P. und Vaccaro, A.M.** (2001) Saposin D solubilizes anionic phospholipid-containing membranes. *J. Biol. Chem.*, **276**, 31583-31589.
- Ciaffoni, F., Tatti, M., Salvioli, R. und Vaccaro, A.M.** (2003) Interaction of saposin D with membranes: effect of anionic phospholipids and sphingolipids. *Biochem. J., Pt.*
- Clayton, P.T.** (2001) Applications of mass spectrometry in the study of inborn errors of metabolism. *J. Inherit. Metab. Dis.*, **24**, 139-150.
- Cowtan, K.** (1994) dm: An automated procedure for phase improvement by density modification. *Joint CCP4 and ESF-EACBM Newsletter on Protein Crystallography*, **31**, 34-38.
- Cowtan, K.** (1998) Modified phased translation functions and their application to molecular-fragment location. *Acta Crystallogr. D.*, **54 (Pt 5)**, 750-756.
- Dauter, Z., Dauter, M. und Rajashankar, K.R.** (2000) Novel approach to phasing proteins: derivatization by short cryo-soaking with halides. *Acta Crystallogr. D.*, **56**, 232-237.
- d'Azzo, A., Proia, R.L., Kolodny, E.H., Kaback, M.M. und Neufeld, E.F.** (1984) Faulty association of alpha- and beta-subunits in some forms of beta-hexosaminidase A deficiency. *J. Biol. Chem.*, **259**, 11070-11074.
- Diederichs, K. und Karplus, P.A.** (1997) Improved R-factors for diffraction data analysis in macromolecular crystallography. *Nat. Struct. Biol.*, **4**, 269-275.
- Ducruix, A. und Giegé, R.** (1992) *Crystallization of Nucleic acids and Proteins*. Oxford University Press.
- Evans, G. und Bricogne, G.** (2002) Triiodide derivatization and combinatorial counter-ion replacement: two methods for enhancing phasing signal using laboratory Cu Kalpha X-ray equipment. *Acta Crystallogr. D.*, **58**, 976-991.
- Fujibayashi, S. und Wenger, D.A.** (1986a) Biosynthesis of the sulfatide/GM1 activator protein (SAP-1) in control and mutant cultured skin fibroblasts. *Biochim. Biophys. Acta*, **875**, 554-562.
- Fujibayashi, S. und Wenger, D.A.** (1986b) Synthesis and processing of sphingolipid activator protein-2 (SAP-2) in cultured human fibroblasts. *J. Biol. Chem.*, **261**, 15339-15343.

- Fujimaru, M., Tanaka, A., Choeh, K., Wakamatsu, N., Sakuraba, H. und Isshiki, G.** (1998) Two mutations remote from an exon/intron junction in the beta-hexosaminidase beta-subunit gene affect 3'-splice site selection and cause Sandhoff disease. *Hum. Genet.*, **103**, 462-469.
- Fürst, W. und Sandhoff, K.** (1992) Activator proteins and topology of lysosomal sphingolipid catabolism. *Biochim. Biophys. Acta*, **1126**, 1-16.
- Gomez-Lira, M., Sangalli, A., Mottes, M., Perusi, C., Pignatti, P.F., Rizzuto, N. und Salviati, A.** (1995) A common beta hexosaminidase gene mutation in adult Sandhoff disease patients. *Hum. Genet.*, **96**, 417-422.
- Gravel, A., Kaback, M., Proia, R.L., Sandhoff, K., Suzuki, K. und Suzuki, K.** (2001) The GM2 gangliosidoses. In Scriver, C.R., Beaudet, A.L., Sly, W.S. and Valle, D. (eds.), *The Metabolic and Molecular Bases of Inherited Diseases*. McGraw-Hill, New York, Vol. 3, pp. 3827-3877.
- Hakomori, S. und Kannagi, R.** (1983) Glycosphingolipids as tumor-associated and differentiation markers. *J. Natl. Cancer Inst.*, **71**, 231-251.
- Hammel, M., Kriechbaum, M., Gries, A., Kostner, G.M., Laggner, P. und Prassl, R.** (2002) Solution structure of human and bovine beta(2)-glycoprotein I revealed by small-angle X-ray scattering. *J. Mol. Biol.*, **321**, 85-97.
- Hayward, S. und Berendsen, H.J.** (1998) Systematic analysis of domain motions in proteins from conformational change: new results on citrate synthase and T4 lysozyme. *Proteins*, **30**, 144-154.
- Henseler, M., Klein, A., Glombitza, G.J., Suzuki, K. und Sandhoff, K.** (1996) Expression of the three alternative forms of the sphingolipid activator protein precursor in baby hamster kidney cells and functional assays in a cell culture system. *J. Biol. Chem.*, **271**, 8416-8423.
- Hepbildikler, S.T., Sandhoff, R., Kolzer, M., Proia, R.L. und Sandhoff, K.** (2002) Physiological substrates for human lysosomal beta -hexosaminidase S. *J. Biol. Chem.*, **277**, 2562-2572.
- Hiraiwa, M., Soeda, S., Martin, B.M., Fluharty, A.L., Hirabayashi, Y., O'Brien, J.S. und Kishimoto, Y.** (1993) The effect of carbohydrate removal on stability and activity of saposin B. *Arch. Biochem. Biophys.*, **303**, 326-331.
- Holmgren, J., Lonnroth, I., Mansson, J. und Svennerholm, L.** (1975) Interaction of cholera toxin and membrane GM1 ganglioside of small intestine. *Proc. Natl. Acad. Sci. U. S. A.*, **72**, 2520-2524.
- Hooft, R.W., Vriend, G., Sander, C. und Abola, E.E.** (1996) Errors in protein structures. *Nature*, **381**, 272.
- Hou, Y., McInnes, B., Hinek, A., Karpati, G. und Mahuran, D.** (1998) A Pro504 --> Ser substitution in the beta-subunit of beta-hexosaminidase A inhibits alpha-subunit hydrolysis of GM2 ganglioside, resulting in chronic Sandhoff disease. *J. Biol. Chem.*, **273**, 21386-21392.

- Ichikawa, S., Nakajo, N., Sakiyama, H. und Hirabayashi, Y.** (1994) A mouse B16 melanoma mutant deficient in glycolipids. *Proc. Natl. Acad. Sci. U. S. A.*, **91**, 2703-2707.
- Ito, K., Takahashi, N., Takahashi, A., Shimada, I., Arata, Y., O'Brien, J.S. und Kishimoto, Y.** (1993) Structural study of the oligosaccharide moieties of sphingolipid activator proteins, saposins A, C and D obtained from the spleen of a Gaucher patient. *Eur. J. Biochem.*, **215**, 171-179.
- Jones, S. und Thornton, J.M.** (1996) Principles of protein-protein interactions. *Proc. Natl. Acad. Sci. U. S. A.*, **93**, 13-20.
- Jones, T.A.** (1978) A graphics model building and refinement system for macromolecules. *J. Appl. Cryst.*, **11**, 268-272.
- Kabsch, W.** (1976) Solution for best rotation to relate 2 sets of vectors. *Acta Crystallogr. A.*, **32**, 922-923.
- Kabsch, W.** (1993) Automatic Processing of Rotation Diffraction Data from Crystals of Initially Unknown Symmetry and Cell Constants. *J. Appl. Cryst.*, **26**, 795-800.
- Karlsson, K.A.** (1989) Animal glycosphingolipids as membrane attachment sites for bacteria. *Annu. Rev. Biochem.*, **58**, 309-350.
- Katchalski-Katzir, E., Shariv, I., Eisenstein, M., Friesem, A.A., Aflalo, C. und Vakser, I.A.** (1992) Molecular surface recognition: determination of geometric fit between proteins and their ligands by correlation techniques. *Proc. Natl. Acad. Sci. U. S. A.*, **89**, 2195-2199.
- Kleywegt, G.J.** (1995) Dictionaries for Heteros. *Joint CCP4 and ESF-EACBM Newsletter on Protein Crystallography*, **32**, 45-50.
- Konarev, P.V., Petoukhov, M.V. und Svergun, D.I.** (2001) MASSHA - a graphics system for rigid-body modelling of macromolecular complexes against solution scattering data. *J. Appl. Cryst.*, **34**, 527-532.
- Konarev, P.V., Volkov, V.V., Koch, M.H.J. und Svergun, D.I.** wintel-based menu-driven program packages for small-angle scattering data analysis. *submitted*.
- Kozin, M.B. und Svergun, D.I.** (2001) A software system for rigid body modelling of solution scattering data. *J. Appl. Cryst.*, **34**, 33-41.
- Kozin, M.B., Volkov, V.V. und Svergun, D.I.** (1997) ASSA - a program for three-dimensional rendering in solution scattering from biopolymers. *J. Appl. Cryst.*, **30**, 811-815.
- Kraulis, P.J.** (1991) MOLSCRIPT: A Program to Produce Both Detailed and Schematic Plots of Protein Structures. *J. Appl. Cryst.*, **24**, 946-950.
- Krebs, W.G. und Gerstein, M.** (2000) The morph server: a standardized system for analyzing and visualizing macromolecular motions in a database framework. *Nucleic Acids Res.*, **28**, 1665-1675.
- Kumar, M.S., Carson, M., Hussain, M.M. und Murthy, H.M.** (2002) Structures of apolipoprotein A-II and a lipid-surrogate complex provide insights into apolipoprotein-lipid interactions. *Biochemistry (Mosc)*. **41**, 11681-11691.

- Kuroki, Y., Itoh, K., Nadaoka, Y., Tanaka, T. und Sakuraba, H.** (1995) A novel missense mutation (C522Y) is present in the beta-hexosaminidase beta-subunit gene of a Japanese patient with infantile Sandhoff disease. *Biochem. Biophys. Res. Commun.*, **212**, 564-571.
- Kytzia, H.J. und Sandhoff, K.** (1985) Evidence for two different active sites on human beta-hexosaminidase A. Interaction of GM2 activator protein with beta-hexosaminidase A. *J. Biol. Chem.*, **260**, 7568-7572.
- Laskowski, R.A., MacArthur, M.W., Moss, D.S. und Thornton, J.M.** (1993) PROCHECK: a program to check the stereochemical quality of protein structures. *J. Appl. Cryst.*, **26**, 283-291.
- Lawrence, M.C. und Bourke, P.** (2000) CONSCRIPT: a program for generating electron density isosurfaces for presentation in protein crystallography. *J. Appl. Cryst.*, **33**, 990-991.
- Legler, G., Lullau, E., Kappes, E. und Kastenholz, F.** (1991) Bovine N-acetyl-beta-D-glucosaminidase: affinity purification and characterization of its active site with nitrogen containing analogs of N-acetylglucosamine. *Biochim. Biophys. Acta*, **1080**, 89-95.
- Leippe, M., Ebel, S., Schoenberger, O.L., Horstmann, R.D. und Muller-Eberhard, H.J.** (1991) Pore-forming peptide of pathogenic Entamoeba histolytica. *Proc. Natl. Acad. Sci. U. S. A.*, **88**, 7659-7663.
- Liepinsh, E., Andersson, M., Ruysschaert, J.M. und Otting, G.** (1997) Saposin fold revealed by the NMR structure of NK-lysin. *Nat. Struct. Biol.*, **4**, 793-795.
- Liessem, B., Glombitza, G.J., Knoll, F., Lehmann, J., Kellermann, J., Lottspeich, F. und Sandhoff, K.** (1995) Photoaffinity labeling of human lysosomal beta-hexosaminidase B. Identification of Glu-355 at the substrate binding site. *J. Biol. Chem.*, **270**, 23693-23699.
- Liu, J., Shikhman, A.R., Lotz, M.K. und Wong, C.H.** (2001) Hexosaminidase inhibitors as new drug candidates for the therapy of osteoarthritis. *Chem. Biol.*, **8**, 701-711.
- Lodge, J.A., Maier, T., Liebl, W., Hoffmann, V. und Strater, N.** (2003) Crystal structure of Thermotoga maritima alpha-glucosidase AgI A defines a new clan of NAD⁺-dependent glycosidases. *J. Biol. Chem.*, **278**, 19151-19158.
- Lovell, S.C., Word, J.M., Richardson, J.S. und Richardson, D.C.** (2000) The penultimate rotamer library. *Proteins*, **40**, 389-408.
- Lukatela, G., Krauss, N., Theis, K., Selmer, T., Gieselmann, V., von Figura, K. und Saenger, W.** (1998) Crystal structure of human arylsulfatase A: the aldehyde function and the metal ion at the active site suggest a novel mechanism for sulfate ester hydrolysis. *Biochemistry (Mosc)*, **37**, 3654-3664.
- Makita, A. und Yamakawa, R.** (1963) The glycolipids of the brain of Tay-sachs disease. Chemical structure of globoside and main ganglioside. *Jpn. J. Exp. Med.*, **33**, 361.
- Mark, B.L., Vocadlo, D.J., Knapp, S., Triggs-Raine, B.L., Withers, S.G. und James, M.N.** (2001) Crystallographic evidence for substrate-assisted catalysis in a bacterial beta-hexosaminidase. *J. Biol. Chem.*, **276**, 10330-10337.

- Markwell, M.A., Svennerholm, L. und Paulson, J.C.** (1981) Specific gangliosides function as host cell receptors for Sendai virus. *Proc. Natl. Acad. Sci. U. S. A.*, **78**, 5406-5410.
- Martins, A.M.** (1999) Inborn errors of metabolism: a clinical overview. *Sao Paulo Med. J.*, **117**, 251-265.
- Marti-Renom, M.A., Stuart, A.C., Fiser, A., Sanchez, R., Melo, F. und Sali, A.** (2000) Comparative protein structure modeling of genes and genomes. *Annu. Rev. Biophys. Biomol. Struct.*, **29**, 291-325.
- McDonald, I.K. und Thornton, J.M.** (1994) Satisfying hydrogen bonding potential in proteins. *J. Mol. Biol.*, **238**, 777-793.
- McInnes, B., Potier, M., Wakamatsu, N., Melancon, S.B., Klavins, M.H., Tsuji, S. und Mahuran, D.J.** (1992) An unusual splicing mutation in the HEXB gene is associated with dramatically different phenotypes in patients from different racial backgrounds. *J. Clin. Invest.*, **90**, 306-314.
- Meier, E.M., Schwarzmann, G., Furst, W. und Sandhoff, K.** (1991) The human GM2 activator protein. A substrate specific cofactor of beta-hexosaminidase A. *J. Biol. Chem.*, **266**, 1879-1887.
- Merritt, E.A. und Bacon, D.J.** (1997) Raster3D: Photorealistic Molecular Graphics. *Methods Enzymol.*, **277**, 505-524.
- Morimoto, S., Martin, B.M., Yamamoto, Y., Kretz, K.A., O'Brien, J.S. und Kishimoto, Y.** (1989) Saposin A: second cerebrosidase activator protein. *Proc. Natl. Acad. Sci. U. S. A.*, **86**, 3389-3393.
- Munford, R.S., Sheppard, P.O. und O'Hara, P.J.** (1995) Saposin-like proteins (SAPLIP) carry out diverse functions on a common backbone structure. *J. Lipid Res.*, **36**, 1653-1663.
- Murshudov, G.N., Vagin, A.A. und Dodson, E.J.** (1997) Refinement of macromolecular structures by the maximum-likelihood method. *Acta Crystallogr. D.*, **53**, 240-255.
- Narkis, G., Adam, A., Jaber, L., Pennybacker, M., Proia, R.L. und Navon, R.** (1997) Molecular basis of heat labile hexosaminidase B among Jews and Arabs. *Hum. Mutat.*, **10**, 424-429.
- Navaza, J.** (1994) Amore - an Automated Package for Molecular Replacement. *Acta Crystallogr. A.*, **50**, 157-163.
- Navon, R. und Adam, A.** (1990) Thermolabile hexosaminidase (Hex) B: diverse frequencies among Jewish communities and implication for screening of sera for Hex A deficiencies. *Hum. Hered.*, **40**, 99-104.
- Niere, M., Dettloff, M., Maier, T., Ziegler, M. und Wiesner, A.** (2001) Insect immune activation by apolipoporphin III is correlated with the lipid-binding properties of this protein. *Biochemistry (Mosc.)*, **40**, 11502-11508.
- Ohkuma, S. und Poole, B.** (1978) Fluorescence probe measurement of the intralysosomal pH in living cells and the perturbation of pH by various agents. *Proc. Natl. Acad. Sci. U. S. A.*, **75**, 3327-3331.

- Otwinowski, Z. und Minor, W.** (1997) Processing of X-Ray Diffraction Data Collected in Oscillation Mode. In Carter C.W.Jr, S.R.M. (ed.), *Macromolecular Crystallography Part A*. Academic Press, Vol. 276, pp. 307-326.
- Pennybacker, M., Schuette, C.G., Liessem, B., Hepbildikler, S.T., Kopetka, J.A., Ellis, M.R., Myerowitz, R., Sandhoff, K. und Proia, R.L.** (1997) Evidence for the involvement of Glu-355 in the catalytic action of human beta-hexosaminidase B. *J. Biol. Chem.*, **272**, 8002-8006.
- Perrakis, A., Harkiolaki, M., Wilson, K.S. und Lamzin, V.S.** (2001) ARP/wARP and molecular replacement. *Acta Crystallogr. D.*, **57**, 1445-1450.
- Prag, G., Papanikolau, Y., Tavlas, G., Vorgias, C.E., Petratos, K. und Oppenheim, A.B.** (2000) Structures of chitobiase mutants complexed with the substrate Di-N-acetyl-d-glucosamine: the catalytic role of the conserved acidic pair, aspartate 539 and glutamate 540. *J. Mol. Biol.*, **300**, 611-617.
- Prevelige, P. und Fasman, G.D.** (1989) Chou-Fasman Prediction of Secondary Structure. In Fasman, G.D. (ed.), *Prediction of Protein Structure and the Principles of Protein Conformation*. Plenum, New York, pp. 1-91.
- Proia, R.L.** (1988) Gene encoding the human beta-hexosaminidase beta chain: extensive homology of intron placement in the alpha- and beta-chain genes. *Proc. Natl. Acad. Sci. U. S. A.*, **85**, 1883-1887.
- Proia, R.L., d'Azzo, A. und Neufeld, E.F.** (1984) Association of alpha- and beta-subunits during the biosynthesis of beta-hexosaminidase in cultured human fibroblasts. *J. Biol. Chem.*, **259**, 3350-3354.
- Provost, K. und Robert, M.C.** (1991) Application of gel growth to hanging drop technique. *J. Cryst. Growth*, **110**, 258-264.
- Qi, X. und Grabowski, G.A.** (2001) Differential membrane interactions of saposins A and C: implications for the functional specificity. *J. Biol. Chem.*, **276**, 27010-27017.
- Qi, X., Leonova, T. und Grabowski, G.A.** (1994) Functional human saposins expressed in Escherichia coli. Evidence for binding and activation properties of saposins C with acid beta-glucosidase. *J. Biol. Chem.*, **269**, 16746-16753.
- Reiner, O., Dagan, O. und Horowitz, M.** (1989) Human sphingolipid activator protein-1 and sphingolipid activator protein-2 are encoded by the same gene. *J. Mol. Neurosci.*, **1**, 225-233.
- Sachs, B.** (1887) On arrested cerebral development with special reference to its cortical pathology. *J. Nerv. Ment. Dis.*, **14**, 541.
- Sali, A. und Blundell, T.L.** (1993) Comparative protein modelling by satisfaction of spatial restraints. *J. Mol. Biol.*, **234**, 779-815.
- Sanner, M.F., Olson, A.J. und Spehner, J.C.** (1996) Reduced surface: an efficient way to compute molecular surfaces. *Biopolymers*, **38**, 305-320.

- Sano, A. und Radin, N.S.** (1988) The carbohydrate moiety of the activator protein for glucosylceramide beta-glucosidase. *Biochem. Biophys. Res. Commun.*, **154**, 1197-1203.
- Saudubray, J.M. und Charpentier, C.** (1995) Clinical Phenotypes:Diagnosis/Algorithms. In Scriver, C.R. (ed.), *The metabolic and molecular bases of inherited disease*. McGraw-Hill.
- Schuette, C.G., Weisgerber, J. und Sandhoff, K.** (2001) Complete analysis of the glycosylation and disulfide bond pattern of human beta-hexosaminidase B by MALDI-MS. *Glycobiology*, **11**, 549-556.
- Shindyalov, I.N. und Bourne, P.E.** (1998) Protein structure alignment by incremental combinatorial extension (CE) of the optimal path. *Protein Eng.*, **11**, 739-747.
- Sonderfeld-Fresco, S. und Proia, R.L.** (1988) Synthesis and assembly of a catalytically active lysosomal enzyme, beta-hexosaminidase B, in a cell-free system. *J. Biol. Chem.*, **263**, 13463-13469.
- Soulages, J.L. und Arrese, E.L.** (2000) Dynamics and hydration of the alpha-helices of apolipoporphin III. *J. Biol. Chem.*, **275**, 17501-17509.
- Stowell, M.H.B., Soltis, S.M., Kisker, C., Peters, J.W., Schindelin, H., Rees, D.C., Cascio, D., Beamer, L., Hart, P.J., Wiener, M.C. und Whitby, F.G.** (1996) A simple device for studying macromolecular crystals under moderate gas pressures (0.1-10 MPa). *J. Appl. Cryst.*, **29**, 608-613.
- Svennerholm, L.** (1962) The chemical structure of normal human brain and Tay-Sachs gangliosides. *Biochem. Biophys. Res. Commun.*, **9**, 436.
- Svergun, D., Barberato, C. und Koch, M.H.J.** (1995) CRYSTOL - a Program to Evaluate X-ray Solution Scattering of Biological Macromolecules from Atomic Coordinates. *J. Appl. Cryst.*, **28**, 768-773.
- Svergun, D.I., Petoukhov, M.V. und Koch, M.H.J.** (2001) Determination of domain structure of proteins from X-ray solution scattering. *Biophys. J.*, **80**, 2946-2953.
- Svergun, D.I., Volkov, V.V., Kozin, M.B. und Stuhrmann, H.B.** (1996) New Developments in Direct Shape determination from Small-Angle Scattering. 2. Uniqueness. *Acta Crystallogr. A.*, **52**, 419-426.
- Tatti, M., Salvioli, R., Ciaffoni, F., Pucci, P., Andolfo, A., Amoresano, A. und Vaccaro, A.M.** (1999) Structural and membrane-binding properties of saposin D. *Eur. J. Biochem.*, **263**, 486-494.
- Tay, W.** (1881) Symmetrical changes in the reion of the yellow spot in each eye of an infant. *Trans. Ophthalmol. Soc. UK*, **1**, 155.
- Terwilliger, T.C. und Berendzen, J.** (1999) Automated MAD and MIR structure solution. *Acta Crystallogr. D.*, **55**, 849-861.
- Tews, I., Perrakis, A., Oppenheim, A., Dauter, Z., Wilson, K.S. und Vorgias, C.E.** (1996) Bacterial chitobiase structure provides insight into catalytic mechanism and the basis of Tay-Sachs disease. *Nat. Struct. Biol.*, **3**, 638-648.

- Thompson, J.D., Higgins, D.G. und Gibson, T.J.** (1994) CLUSTAL W: improving the sensitivity of progressive multiple sequence alignment through sequence weighting, position-specific gap penalties and weight matrix choice. *Nucleic Acids Res.*, **22**, 4673-4680.
- Tse, R., Wu, Y.J., Vavouglis, G., Hou, Y., Hinek, A. und Mahuran, D.J.** (1996) Identification of functional domains within the alpha and beta subunits of beta-hexosaminidase A through the expression of alpha-beta fusion proteins. *Biochemistry (Mosc)*. **35**, 10894-10903.
- Vaccaro, A.M., Salvioli, R., Barca, A., Tatti, M., Ciaffoni, F., Maras, B., Siciliano, R., Zappacosta, F., Amoresano, A. und Pucci, P.** (1995) Structural analysis of saposin C and B. Complete localization of disulfide bridges. *J. Biol. Chem.*, **270**, 9953-9960.
- Vagin, A. und Teplyakov, A.** (1997) MOLREP: an automated program for molecular replacement. *J. Appl. Cryst.*, **30**, 1022-1025.
- Vagin, A. und Teplyakov, A.** (2000) An approach to multi-copy search in molecular replacement. *Acta Crystallogr. D.*, **56**, 1622-1624.
- Volkov, V.V. und Svergun, D.** (2003) Uniqueness of ab initio shape determination in small angle scattering. *J. Appl. Cryst.*, **36**, 860-864.
- von Bulow, R., Schmidt, B., Dierks, T., Schwabauer, N., Schilling, K., Weber, E., Uson, I. und von Figura, K.** (2002) Defective oligomerization of arylsulfatase a as a cause of its instability in lysosomes and metachromatic leukodystrophy. *J. Biol. Chem.*, **277**, 9455-9461.
- Waber, L.** (1990) Inborn errors of metabolism. *Pediatr. Ann.*, **19**, 105-109, 112-103, 117-108.
- Wakamatsu, N., Kobayashi, H., Miyatake, T. und Tsuji, S.** (1992) A novel exon mutation in the human beta-hexosaminidase beta subunit gene affects 3' splice site selection. *J. Biol. Chem.*, **267**, 2406-2413.
- Wallace, A.C., Laskowski, R.A. und Thornton, J.M.** (1995) LIGPLOT: a program to generate schematic diagrams of protein-ligand interactions. *Protein Eng.*, **8**, 127-134.
- Weik, M., Ravelli, R.B., Kryger, G., McSweeney, S., Raves, M.L., Harel, M., Gros, P., Silman, I., Kroon, J. und Sussman, J.L.** (2000) Specific chemical and structural damage to proteins produced by synchrotron radiation. *Proc. Natl. Acad. Sci. U. S. A.*, **97**, 623-628.
- Weiss, M.S.** (2001) Global indicators of X-ray data quality. *J. Appl. Cryst.*, **34**, 130-135.
- Weitz, G. und Proia, R.L.** (1992) Analysis of the glycosylation and phosphorylation of the alpha-subunit of the lysosomal enzyme, beta-hexosaminidase A, by site-directed mutagenesis. *J. Biol. Chem.*, **267**, 10039-10044.
- Westhead, D.R., Slidel, T.W., Flores, T.P. und Thornton, J.M.** (1999) Protein structural topology: Automated analysis and diagrammatic representation. *Protein Sci.*, **8**, 897-904.

- Wright, C.S., Li, S.C. und Rastinejad, F.** (2000) Crystal structure of human GM2-activator protein with a novel beta-cup topology. *J. Mol. Biol.*, **304**, 411-422.
- Yadao, F., Hechtman, P. und Kaplan, F.** (1997) Formation of a ternary complex between GM2 activator protein, GM2 ganglioside and hexosaminidase A. *Biochim. Biophys. Acta*, **1340**, 45-52.
- Yamashita, K., Inui, K., Totani, K., Kochibe, N., Furukawa, M. und Okada, S.** (1990) Characteristics of asparagine-linked sugar chains of sphingolipid activator protein 1 purified from normal human liver and GM1 gangliosidosis (type 1) liver. *Biochemistry (Mosc)*, **29**, 3030-3039.
- Yamashita, T., Wada, R., Sasaki, T., Deng, C., Bierfreund, U., Sandhoff, K. und Proia, R.L.** (1999) A vital role for glycosphingolipid synthesis during development and differentiation. *Proc. Natl. Acad. Sci. U. S. A.*, **96**, 9142-9147.
- You, H.X., Yu, L. und Qi, X.** (2001) Phospholipid membrane restructuring induced by saposin C: a topographic study using atomic force microscopy. *FEBS Lett.*, **503**, 97-102.
- Zaltash, S., Palmblad, M., Curstedt, T., Johansson, J. und Persson, B.** (2000) Pulmonary surfactant protein B: a structural model and a functional analogue. *Biochim. Biophys. Acta*, **1466**, 179-186.
- Zeller, C.B. und Marchase, R.B.** (1992) Gangliosides as modulators of cell function. *Am. J. Physiol.*, **262**, C1341-1355.
- Zhang, Z.X., Wakamatsu, N., Akerman, B.R., Mules, E.H., Thomas, G.H. und Gravel, R.A.** (1995) A second, large deletion in the HEXB gene in a patient with infantile Sandhoff disease. *Hum. Mol. Genet.*, **4**, 777-780.