

## 遺伝性網膜疾患のDNA診断と標準化

著者	三木 直正
著者別表示	Miki Naomasa
雑誌名	平成1(1989)年度 科学研究費補助金 試験研究 研究成果報告書概要
巻	1987 1989
ページ	3p.
発行年	1993-03-25
URL	<a href="http://doi.org/10.24517/00067608">http://doi.org/10.24517/00067608</a>



# 1989 Fiscal Year Final Research Report Summary

## DNA-diagnosis and standardization of hereditary retial diseases.

Research Project

### Project/Area Number

62870071

### Research Category

Grant-in-Aid for Developmental Scientific Research

### Allocation Type

Single-year Grants

### Research Field

Ophthalmology

### Research Institution

Osaka University Medical School (1989)  
Kanazawa University (1987-1988)

### Principal Investigator

**MIKI Naomasa** Osaka Univ.Medical School, Prof., 医学部, 教授 (40094445)

### Co-Investigator(Kenkyū-buntansha)

HONDA Yoshihito Kyoto Univ.Medical School, Prof., 医学部, 教授 (90026930)  
NAKAZIMA Akira Juntendo Univ.Medical School, Prof., 医学部, 名誉教授 (90052927)  
TAMAI Makoto Tohoku Univ.Medical School, Prof., 医学部, 教授 (90004720)  
KAWASAKI Kazuo Kanazawa Univ.Medical School, Prof., 医学部, 教授 (20019920)  
OHBA Norio Kagoshima Univ.Medical School, Prof., 医学部, 教授 (50010070)

### Project Period (FY)

1987 - 1989

### Keywords

Retina / Hereditary diseases / Diagnosis / Molecular biology / Photo receptor cells / Pigment epithelium

### Research Abstract

The retina has many hereditary diseases compared to other tissues. Recently the locus genes or the genes close to the locus have been isolated such as L1.28 for retinitis pigmentosa, red, blue and green genes for color blindness, ornithine aminotransferase for Gray's atrophy, mitochondrial DNA for Leber's disease, Opsin for night blindness, Rb gene for retinoblastoma. Diagnosis by restriction fragment polymorphism(RFP) was applied to retinitis pigmentosa and Leber's disease using L1.28 and mitochondrial DNA probes, respectively. The distribution of ornithine aminotransferase which is a locus gene of Gray' atrophy

was studied immunohistochemically in the retinas. The structures and functions of two retina-specific cDNAs (MEKA and visinin) were studied and it was found that MEKA protein is associated with beta- and gamma- subunits of transducin, and visinin is a calcium-binding protein in the cone cells. Pigment epithelium-specific cDNA clone (MM115) was also isolated and it had a protease inhibitory activity. It is suggested that a clinician should perform the DNA-diagnosis for himself and use the locus DNA for a probe, not a probe which needs the linkage analysis.

## Research Products (25 results)

	All	Other
	All	Publications (25 results)
[Publications] 三木直正、林要喜知、谷浦秀夫: "突起伸展因子 of 構造と機能" 薬物精神行動. 8. 471-473 (1989)		▼
[Publications] 三木直正、谷浦秀夫、林要喜知: "筋肉より抽出した神経伸展因子" 神経研究の進歩. 33. 1018-1023 (1989)		▼
[Publications] 谷浦秀夫、林要喜知、A.F.Horwits、三木直正: "シナプス形成と細胞外マトリックス" 蛋白質核酸酵素「神経化学特集号」. 35. 357-364 (1990)		▼
[Publications] 三木直正: "神経突起伸展因子とその受容体" 医学のあゆみ. (1990)		▼
[Publications] H.Taniura,C-H.Kuo,Y.Hayashi and N.Miki: "Purification and Characterization of a receptor for neurite outgrowth factor(NOF)" J.Biol.Chem.(1990)		▼
[Publications] H.Taniura,Y.Hayashi and N.Miki: "Possible involvement of neurite outgrowth factor(NOF)-receptor in axonal outgrowth from developing chick retina" Neurosci.Res. (1990)		▼
[Publications] 三木直正、谷浦秀夫、林要喜知: "分子神経生物(分担、ニユ-ロンの突起形成)" 分子生物学会編、丸善出版, 16 (1989)		▼
[Publications] 三木直正、谷浦秀夫、山形要人、林要喜知: "内因性神経活性物質(分担、神経細胞の生存および突起伸展に働く因子)" 吉田博編、中外出版, 11 (1989)		▼
[Publications] K. Yamagata, K. Goto, C-H. Kuo, H. Kondo and N. Miki: "Visinin; A novel calcium-binding protein expressed in retinal cone cells." Neuron. (1990).		▼
[Publications] A. Nakajima, K. Fujiki and Y. Hotta: "Advances in ophthalmic molecular genetics." Asia Pacific Acad. Ophthal. 12 (1990).		▼
[Publications] K. Goto, N. Miki and H. Kondo: "An immunohistochemical study of pinealocytes of chicks and some other lower vertebrates by means of visinin (retinal cone-specific protein)-immunoreactivity." Arch. Histol. Cytol. 52, 451-458 (1989).		▼
[Publications] Y. Hotta, M. Hayakawa, K. Saito, A. Kanai, A. Nakajima and K. Fujiki: "Diagnosis of Leber's optic neuropathy by means of polymerase chain reaction amplification." Amer. J. Ophthal. 108, 601-602 (1989).		▼
[Publications] T. Mito, T. Shiono, S. Ishiguro, M. Tamai, K. Mizuno and T. Ohura: "Immunocytochemical localization of ornithine aminotransferase in human ocular tissues." Arch. Ophthalmol. 107, 1372-1374 (1989).		▼
[Publications] K. Nakao, N. Ohba, M. Isashiki, Y. Isashiki, K. Unoki and M. Osame: "Pigmentary retinal degeneration in patients with HTLV-I-associated myelopathy." Jpn. J. Ophthalmol. 33, 383-391 (1989).		▼
[Publications] C-H. Kuo and N. Miki: "Translocation of MEKA protein in photoreceptor cells by light." Neurosci. Lett. 103, 8-10 (1989).		▼
[Publications] C-H. Kuo, H. Taniura, Y. Watanabe, Y. Fukada, T. Yoshizawa and N. Miki: "Identification of a retina-specific MEKA protein as a 33kDa protein." Biochem. Biophys. Res. Comm. 162, 1063-1068 (1989).		▼
[Publications] C-H. Kuo, Y. Watanabe, K. Yamagata and N. Miki: "Developmental changes of MEKA protein and opsin in normal and rd mice." Dev. Brain Res. 50, 139-141 (1989).		▼
[Publications] C-H. Kuo, M. Akiyama and N. Miki: "Isolation of a novel retina-specific clone (MEKA cDNA) encoding a photoreceptor soluble protein." Molec. Brain Res. 6, 1-10 (1989).		▼
[Publications] F. Uehara, M. Sameshima, K. Takumi, K. Unoki, T. Muramatsu and N. Ohba: "Sialic acid in the retina and its significance in the retinal degeneration." Kagoshima Internatl. Symp. Glycoconjugates in Medicine. 310-315 (1988).		▼

[Publications] S. Hirashima and N. Ohba: "A pedigree of Leber's congenital amaurosis." *Ophthalmic Paediatr. Genet.* 9, 29-36 (1988). ▼

[Publications] K. Unoki, F. Uehara, M. Sameshima, K. Nakano and N. Ohba: "Specific binding of peanut agglutinin to foveal and peripheral cone photoreceptors of monkey retina." *Ophthalmic Res.* 20, 112-126 (1988). ▼

[Publications] C-H. Kuo, M. Akiyama and N. Miki: "Isolation of a novel retina-specific clone (MEKA cDNA) encoding a photoreceptor soluble protein." *Molec. Brain Res.* 6, 1-10 (1988). ▼

[Publications] C-H. Kuo, M. Akiyama and N. Miki: "Cloning and characterization of retina specific cDNAs, MEKA and transducin gamma (Tr)." *Molecular physiology of retinal proteins* (ed. T. Hara). Yamada Conference 21, 9-14 (1988). ▼

[Publications] S. Iseki, H. Kondo, C-H. Kuo and N. Miki: "Longitudinal study on the expression of opsin in the degenerating retina of C3H/He mice." *Arch. Histol. Cytol.* 52, 197-200 (1989). ▼

[Publications] M. Mochii, K. Agata, H. Kobayashi, T. Yamamoto and G. Eguchi: "Expression of a gene coding a melanosomal matrix protein transcriptionally regulated in the transdifferentiation of chick pigmented cells." *Cell Differ.* 24, 67-74 (1988). ▼

**URL:**

Published: 1993-03-25