A main study has been aimed for epidemiological studies on the late effects of atomic bomb (A-bomb) survivors and the actual conditions from socio-medical viewpoints. A purpose as this master does not change, but it passes after being bombed in 70, and A-bomb survivors have gone old aged, and a lifestyle-realted disease and a problem in geriatrics become important as a healthy problem of A-bomb survivors, too. Therefore, as well as a cohort study based on a database, we take in the new study method how we used molecular markers and genetic polymorphic markers for geriatric and neurological diseases to increase with senior people.

1. A-bomb survivors cohort study.

Kawakami H, Morino H, Satou Y*, Hara N* (* International Radiation Information Center)

Purpose: From November 1965, we followed it in “Hiroshima atom bomb Survivors cohort” resident in Hiroshima from biological influence and points of view such as the later living environment of survivors. There is it for the purpose of making basics document contributing to health care / the welfare of A-bomb survivors. As for the examination problem, next is put up.

1) Analysis by A-bomb dose based on ABS93D
2) Analysis of exposure status
3) Analysis by the distance from the hypocenter
4) Analysis of the effects by environment factors
5) Study of disease development by family analysis

2. Study of the disease causative genes in the hereditary neurological diseases.

Morino H, Kawakami H

Purpose & Results: There are many neurological diseases with unknown genes. We have tried to discover these causative genes with 500K SNP chip technology in view of whole genome approach. Recently, we identified that MRE11 mutation leads to progressive myoclonic ataxia and that Perrault syndrome, an autosomal recessive disease exhibiting hearing loss and primary amenorrhea, is caused by compound heterozygous mutations of C10orf2.

3. Study of the mechanisms of amyotrophic lateral sclerosis (ALS).


We detected new mutations of optineurin in amyotrophic lateral sclerosis. We have tried to clarify the mechanisms of ALS with optineurin. So far, we performed behavioral and pathological evaluation using OPTN knockout mice. As a result, significant loss of motor neuron in the spinal cord was observed.
A. Original papers


5. Ohsawa R, Seol J-H*1, Tyler JK*1 (*1 The University of Texas MD Anderson Cancer Center) : At the intersection of non-coding transcription, DNA repair, chromatin structure, and cellular senescence. *Front. Genet.* 2013 4:136. (I)


7. Nakamura S*1, Wate R*1, Kaneko S*1, Ito H*2, Oki M*1, Tsuge A*1, Nagashima M*1, Asayama S*1, Fujita K*1, Nakamura M*1, Maruyama H, Kawakami H, Kusaka H*1 (*1 Kansai Medical University, *2 Wakayama Medical University) : An autopsy case of sporadic amyotrophic lateral sclerosis associated with the I113T SOD1 mutation. *Neuropathology.* 2013 Jun 17. (I)

8. Kamada M*1, Izumi Y*1, Ayaki T*1, Nakamura M*1, Kagawa S*1, Kudo E*1, Sakai W*1, Maruyama H, Nishida Y*1, Kawakami H, Ito H*4,6, Kaji R*1 (*1 Tokushima University, *2 Itsuki Hospital, *3 Kagawa University, *4 Kyoto University, *5 Kansai Medical University, *6 Wakayama Medical University) : Clinicopathologic features of autosomal recessive amyotrophic lateral sclerosis associated with optineurin mutation. *Neuropathology.* 2013 Jul 29. (I)


10. Miyamoto R*1,2, Koizumi H*1,3, Morino H, Kawarai T*1, Maruyama H, Mukai Y*2,3, Miyashiro A*1,2, Sakai W*4, Izumi Y*1, Kawakami H, Kaji R*1,2 (*1 Tokyo Metropolitan Neurological Hospital, *2 Takeda Hospital, *3 National Center of Neurology and Psychiatry, *4 The Feinstein Institute for Medical Research) : DYT6 in Japan–genetic screening and clinical characteristics of the patients. *Mov Disord.* 2013 Nov 13. (I)

11. Homma T*1,2, Nagaoka U*1, Kawata A*1, Mochizuki Y*1,3, Kawakami H, Maruyama H, Matsubara S*1, Komori T*1 (*1 Tokushima University, *2 Saitama Medical University, *3 Tokyo Metropolitan North Rehabilitation Medical Center) : Authors’ Reply to Drs van Blitterswijk M, Rademakers R and van den Berg LH. *Neuropathol Appl Neurobiol.* 2013 Dec 5. (I)

12. Kamon M*1,2, Katano M*1, Hiraki-Kamon K*1,2, Hishida T*1, Nakachi Y*1, Mizuno Y*1, Okazaki Y*1, Suzuki A*1, Hirasaki M*1, Ueda A*1, Nishimoto M*1, Kato H*1, Okuda A*1,2 (*1 Saitama Medical University, *2CREST) : Identification of Ccr4-Not Complex Components as Regulators of Transition from Partial to Genuine Induced Pluripotent Stem Cells. *Stem Cells Dev.* 2013 Dec 9. (I)

13. Hirasaki M*1, Hiraki-Kamon K*1, Katano M*1, Suzuki A*1, Okuda A*1,2 (*1 Saitama Medical University, *2CREST) : Striking similarity in the gene expression levels of individual Myc module members among ESCs, EpiSCs, and partial iPSCs. *PLoS One.* 2013 Dec 26;8(12):e83769. (I)

14. Morino H, Miyamoto R, Ohnishi S*1, Maruyama H, Kawakami H (*1 Rapport Tanabe Neuro Hospital) :

**B. Meeting Presentations**

1. Kawanami A*1, Nagai M*1, Hayakawa H*1, Nihira Y*1, Mizuno Y*2, Maruyama H, Kawakami H, Nishiyama K*1 (*1 Kitazato University): Establishment of ALS model mice expressing mutant OPTN using AAV1. 54th Annual Meeting of the Japanese Society of Neurology, Tokyo, 2013/5/30


3. Izumi Y*1, Miyamoto R, Morino H, Yoshizawa A, Nishinaka K*2, Udaka F*2, Kameyama M*2, Maruyama H, Kawakami H (*1 Tokushima University, *2 Sumitomo Hospital): Clinical manifestation of Cerebellar ataxia with SYNE1 mutation diagnosed by next-generation sequencer. 54th Annual Meeting of the Japanese Society of Neurology, Tokyo, 2013/5/31

4. Kamata M*1, Ikeda K*1, Kume K*1, Urai Y*1, Deguchi K*1, Toge T*1, Miyamoto R, Sugihara K, Morino H, Maruyama H, Kawakami H (*1 Kagawa University): Clinical features of MND+IBM patient with VCP mutation and the frequency of VCP mutation in familial ALS. 54th Annual Meeting of the Japanese Society of Neurology, Tokyo, 2013/6/1

5. Sugihara K, Maruyama H, Miyamoto R, Morino H, Ueno H*1, Matsumoto M*1, Kitaguchi H*2, Yukitake M*2, Higashi Y*1, Nishinaka K*2, Oda M*3, Izumi Y*1, Kawakami H (*1 Neurology, *2 Kurashiki Central Hospital, *3 Saga University, *4 Himeji Central Hospital, *5 Sumitomo Hospital, *6 Vihara Hananosato Hospital, *7 Tokushima University): The clinical characteristics of spinocerebellar ataxia 36: A study of 2,121 Japanese ataxia patients. 54th Annual Meeting of the Japanese Society of Neurology, Tokyo, 2013/6/1


9. Hiraki K, Funayama S*1, Moriyama Y*1,2, Eitoku K*3, Thierry Forné*4, Araki T*5, Okazaki K*1, Kiyosawa H*1, Kawakami H, Okuda A*1, Kato H*1 (*1 Saitama Medical University, *2 Fujita Health University, *3 Kochi University, *4 Institut de Génétique Moléculaire de Montpellier, *5 National Center of Neurology and Psychiatry): Tet1 is essential for efficient differentiation of human iPS cells. 13th Congress of the Japanese Society for Regenerative Medicine, Kyoto, 2014/3/4

(I) indicates reports printed in the scientific journals listed in Current Contents.