Ⅲ．研究成果の刊行に関する一覧表

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◎は、本研究によることが明記されている論文
○は、本研究に関連する論文

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Ⅳ．研究成果の刊行物・別冊

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LETTER TO THE EDITOR

ALDH2 polymorphism in patients with Diamond-Blackfan anemia in Japan

Fumika Ikeda[1] · Tsutomu Toki[1] · Rika Kanezaki[1] · Kiminori Terui[1] · Kenichi Yoshida[2] · Hitoshi Kanno[3] · Shouichi Ohga[4] · Akira Ohara[5] · Seiji Kojima[6] · Seishi Ogawa[2] · Etsuro Ito[1]

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Alcohol aldehyde dehydrogenase 2 (ALDH2) is one of several enzymes that catalyzes the dehydrogenization of aldehydes. ALDH2 deficiency resulting from a Glu504Lys substitution (rs671, c.1510G>A) is prevalent in the Japanese population. ALDH2 is strongly suppressed in the presence of the dominant-negative A allele (Lys504).

Recently, Hira et al. showed that the A allele causes accelerated progression of bone marrow failure and malformations in Japanese Fanconi anemia (FA) patients [1].

However, the pathological significance of *ALDH2* polymorphisms on other inherited bone marrow failure syndromes (IBMFS) remains unknown.

Diamond-Blackfan anemia (DBA) is an inherited bone marrow failure syndrome (IBMFS) characterized by red blood cell aplasia, generally presenting in infancy, sometimes with various malformations [2, 3]. More than 50 % of DBA patients possess heterozygous mutations in 1 of 15 genes encoding ribosomal proteins (RP), and rare X-linked DBA families have shown to have *GATA1* mutations. Corticosteroids remain the primary treatment, and these are beneficial in 80 % of the patients. It is clear that DBA phenotypes can vary widely in severity, even within the same family, ranging from the classical syndrome described above to an absence of symptoms. Therefore, considerable influence from modifying genes has been suggested. However, the primary modifying gene has not yet been identified. In this report, we investigated the effects of *ALDH2* polymorphism on phenotypes in Japanese DBA patients.

All clinical samples were obtained with informed consent from pediatric and/or hematology departments in Japan. The *ALDH2* genotyping of rs671 was performed by TaqMan genotyping assay according to the manufacturer's protocol. The Ethics Committee of Hirosaki University Graduate School of Medicine approved this study.

We performed *ALDH2* genotyping on 113 DBA patients and statistically assessed the effect of *ALDH2* polymorphism on their clinical states. In 113 DBA cases, 75 (66.4 %) possessed heterozygous mutations or deletions in RP genes. The number of patients with *ALDH2* alleles, GG and GA/AA, was 69 and 44, respectively (Table 1). The allele frequencies appeared to be not significantly different from those previously reported for the Japanese population. Age at onset, the time of detection of anemia, in our DBA patients was not affected by *ALDH2* polymorphism. Among the 113 DBA patients, 72 had one or more physical anomalies, including short stature. We categorized the physical anomalies characteristic of DBA into organ/system groups [2]. There were no significant differences in the expression or frequency of anomalies between the GG and the GA/AA groups within any of the organ/system categories. Of the 84 children who received corticosteroid therapy, 40 of 56 (71.4 %) in the GG group and 19 of 28 (67.9 %) in the GA/AA group responded to initial corticosteroid

✉ Etsuro Ito
eturou@hirosaki-u.ac.jp

1 Department of Pediatrics, Hirosaki University Graduate School of Medicine, 5 Zaifu-cho, Hirosaki, Aomori 036-8563, Japan

2 Department of Pathology and Tumor Biology, Graduate School of Medicine, Kyoto University, Kyoto, Japan

3 Department of Transfusion Medicine and Cell Processing, Tokyo Women's Medical University Tokyo, Tokyo, Japan

4 Department of Pediatrics, Yamaguchi University Graduate School of Medicine, Ube, Japan

5 Department of Transfusion, Omori Hospital, Toho University, Tokyo, Japan

6 Department of Pediatrics, Nagoya University Graduate School of Medicine, Nagoya, Japan

Table 1 *ALDH2* polymorphism and clinical states of DBA patients

	ALDH2 polymorphism		Total	GG vs. GA/AA
	GG	GA/AA		*P* value
Patients, *n*	69	44 (37/7)	113	
Onset, months, median (range)	3.6 (0–48)	6.5 (0–72)	4.8 (0–72)	0.45*
Anomaly				
Organ/system, *n* ± SD	1.3 ± 0.15	1.4 ± 0.23	1.3 ± 0.13	0.99*
Frequency				
Craniofacial, *n* (%)	18 (26)	10 (23)	28 (25)	0.69
Ophthalmological, *n* (%)	2 (3)	1 (2)	3 (3)	0.67†
Neck, *n* (%)	2 (3)	2 (5)	4 (4)	0.51†
Thumbs, *n* (%)	12 (17)	6 (14)	18 (16)	0.6
Urogenital, *n* (%)	3 (4)	5 (11)	8 (7)	0.15
Cardiac, *n* (%)	10 (15)	9 (21)	19 (17)	0.41
Other musculoskeletal[a], *n* (%)	31 (45)	22 (50)	53 (47)	0.6
Neuromotor, *n* (%)	9 (13)	5 (11)	14 (12)	0.8
Corticosteroid therapy				
Patients, *n*	56	28	84	
Steroid responder, *n* (%)	40 (71)	19 (68)	59 (70)	0.74

We defined the number of patients for whom clinical data was available as (*n*). *P* values estimated by Pearson's chi-square test, * Mann–Whitney *U* test, and † Fisher's exact test

SD standard deviations

[a] Including short stature (height below −2 SD)

therapy. The reactivity to initial corticosteroid therapy was not affected by *ALDH2* polymorphism. As previous reports had suggested that AA genotype could present noticeable phenotypes in patients with Fanconi anemia [1] and idiopathic aplastic anemia [4], we also compared the clinical characteristics among the GG, GA and AA groups. However, there were no significant differences in age at onset, malformations, or reactivity to corticosteroid among the GG, GA and AA groups (data not shown).

Additionally, to control for differences in effect due to difference of causative gene, we categorized DBA patients into subgroups according to the causative mutations (RP genes, undetected in known mutations, *RPS19*, *RPL5*, and large deletion in RP genes), and performed the same statistical examination on each subgroup. We detected no significant differences in any subgroup in age at onset, malformations, or reactivity to corticosteroid between the GG and the GA/AA groups (data not shown).

The difference in the effects of *ALDH2* polymorphism on FA and DBA is thought to result from differences in pathophysiology. ALDH2 deficiency leads to the accumulation of aldehydes, which may create genotoxic DNA adducts, including DNA interstrand crosslinks (ICLs) and DNA–protein crosslinks. FA cells are defective in a common DNA repair network and are hypersensitive to ICLs. The pathophysiology of DBA remains unclear. However, a widely accepted hypothesis is that ribosomal haploinsufficiency leads to the accumulation of free ribosomal proteins, which bind to MDM2, causing p53 activation and resulting in apoptosis and cell cycle arrest. Furthermore, a recent study showed that selective defects in translation of GATA1 due to haploinsufficiency for RP genes could result in DBA [5]. Neither of these DBA mechanisms involves the disruption of DNA repair pathways, and, as a result, the DBA phenotype would not be expected to be affected by *ALDH2* polymorphism.

Our current data indicate that aldehydes do not have a significant effect on DBA pathophysiology, and support the notion that the accumulation of aldehydes plays an important role specifically in FA. We will continue to search for genetic modifiers of DBA.

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Mutations in the Gene Encoding the E2 Conjugating Enzyme UBE2T Cause Fanconi Anemia

Asuka Hira,[1] Kenichi Yoshida,[2] Koichi Sato,[3] Yusuke Okuno,[4] Yuichi Shiraishi,[5] Kenichi Chiba,[5] Hiroko Tanaka,[6] Satoru Miyano,[5,6] Akira Shimamoto,[7] Hidetoshi Tahara,[7] Etsuro Ito,[8] Seiji Kojima,[4] Hitoshi Kurumizaka,[3] Seishi Ogawa,[2] Minoru Takata,[1,*] Hiromasa Yabe,[9] and Miharu Yabe[9]

Fanconi anemia (FA) is a rare genetic disorder characterized by genome instability, increased cancer susceptibility, progressive bone marrow failure (BMF), and various developmental abnormalities resulting from the defective FA pathway. FA is caused by mutations in genes that mediate repair processes of interstrand crosslinks and/or DNA adducts generated by endogenous aldehydes. The UBE2T E2 ubiquitin conjugating enzyme acts in FANCD2/FANCI monoubiquitination, a critical event in the pathway. Here we identified two unrelated FA-affected individuals, each harboring biallelic mutations in *UBE2T*. They both produced a defective UBE2T protein with the same missense alteration (p.Gln2Glu) that abolished FANCD2 monoubiquitination and interaction with FANCL. We suggest this FA complementation group be named FA-T.

Fanconi anemia (FA) is a rare genetic disease characterized by genome instability, cancer predisposition, progressive bone marrow failure (BMF), and various developmental abnormalities that often include radial ray anomalies, short stature, and visceral malformations.[1] FA cells are hypersensitive to DNA interstrand crosslink damage (ICL) and various types of damage due to endogenous aldehydes.[2–5] FA is caused by mutations in any one of 16 genes that together comprise the FA pathway. These genes include *FANCA* (MIM: 617139), *FANCB* (MIM: 300515), *FANCC* (MIM: 613899), *FANCD1* (*BRCA2*) (MIM: 600185), *FANCD2* (MIM: 613984), *FANCE* (MIM: 613976), *FANCF* (MIM: 603467), *FANCG* (*XRCC9*) (MIM: 600901), *FANCI* (MIM: 611360), *FANCJ* (*BRIP1*) (MIM: 614082), *FANCL* (*PHF9*) (MIM: 614083), *FANCN* (*PALB2*) (MIM: 610832), *FANCO* (*RAD51C*) (MIM: 613390), *FANCP* (*SLX4*) (MIM: 613951), *FANCQ* (*XPF*) (MIM: 615272), and *FANCS* (*BRCA1*) (MIM: 113705). A recent study indicated that biallelic mutations in FA-related *FANCM* (MIM: 609644) do not cause an FA phenotype in humans,[6] raising a concern whether this nomenclature is appropriate or not. In the upstream part of the pathway, the FA core E3 ligase complex consisting of eight gene products and other associated proteins monoubiquitinates FANCD2 and FANCI, resulting in chromatin accumulation/focus formation of FANCD2 that probably recognizes stalled replication forks upon ICL or aldehyde damage. This is the critical event that regulates recruitment of structure-specific nucleases and subsequent incision/unhooking of fork-blocking lesions, mobilizing the downstream repair pathway components.[2,3] *UBE2T* (MIM: 610538) encodes an E2 ubiquitin conjugating enzyme (EC: 6.3.2.19) which has been implicated in this monoubiquitination reaction both in vivo[7–9] and in vitro.[10–13]

We previously analyzed the *ALDH2* genotypes in 64 Japanese FA-affected individuals with the approval of the Research Ethics Committee of the Tokai University Hospital and Kyoto University and obtained informed consent from the families of all subjects involved.[14] Our report included two case subjects in which mutations in the genes previously associated with FA were excluded by whole exome sequencing (WES) (listed as numbers 60 and 61 in Table S1 in Hira et al.[14]) (Figure S1). Serendipitously, *UBE2T* mutations were found in both of them (Figures 1A–1C). The two persons are hereafter designated PNGS-252 (family 1-II-1 in Figure 1D) and PNGS-255 (family 2-II-1 in Figure 1D) (Table 1). They were from unrelated families (Figure 1D) living in different geographic locations in Japan. Both individuals displayed typical FA phenotypes, with malformations and hematological abnormalities that necessitated hematopoietic stem cell transplantation (Table 1; see Supplemental Data). Chromosome fragility in lymphocytes (described in Table S2 in Hira et al.[14]) was consistent with the diagnosis of FA.

WES and validation by Sanger sequencing in PNGS-252 revealed an apparent homozygous c.4C>G missense alteration (GenBank: NM_014176.3), resulting in the amino acid substitution p.Gln2Glu (Figure 1A). This mutation must be very rare, because this is not listed in the NHLBI Exome Sequencing Project or the Human Genetic

[1]Laboratory of DNA Damage Signaling, Department of Late Effects Studies, Radiation Biology Center, Kyoto University, Kyoto 606-8501, Japan; [2]Department of Pathology and Tumor Biology, Graduate School of Medicine, Kyoto University, Kyoto 606-8501, Japan; [3]Laboratory of Structural Biology, Graduate School of Advanced Science and Engineering, Waseda University, Tokyo 162-8480, Japan; [4]Department of Pediatrics, Nagoya University Graduate School of Medicine, Nagoya 466-8550, Japan; [5]Laboratory of DNA Information Analysis, Human Genome Center, Institute of Medical Science, The University of Tokyo, Tokyo 108-8639, Japan; [6]Laboratory of Sequence Analysis, Human Genome Center, Institute of Medical Science, The University of Tokyo, Tokyo 108-8639, Japan; [7]Department of Cellular and Molecular Biology, Graduate School of Biomedical & Health Sciences, Hiroshima University, Minami-ku, Hiroshima 734-8551, Japan; [8]Department of Pediatrics, Hirosaki University Graduate School of Medicine, Hirosaki 036-8562, Japan; [9]Department of Cell Transplantation and Regenerative Medicine, Tokai University School of Medicine, Isehara 259-1193, Japan
*Correspondence: mtakata@house.rbc.kyoto-u.ac.jp
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