

(付表)

## 日本語版Life Experiences Survey

下記の項目は、生活に大きな変化をもたらす出来事や、社会的な再適応が必要とされる出来事です。この1年間に、あなたが経験した下記の出来事は、

どのくらい前の時期であったのか（0から6か月前、もしくは7か月から1年前）、当てはまる欄に○印をつけてください。

その際、○印が、横に並んだ項目と一致した列についているよう確認してください。

また、あなたの生活が、その出来事が起きたことによって肯定的な影響を受けたか、否定的な影響を受けたかについてお答えください。

すなわち、出来事の種類の強さについて、当てはまる数字に○印をつけてください。

“-3”の評点は「非常に否定的な影響」、 “0”の評点は「肯定的でも否定的でもない」、 “+3”の評点は「非常に肯定的な影響」を表します。

## セクションI

	0から 6か月前	7か月から 1年前	非常に 否定的	否定的	やや 否定的	影響 なし	やや 肯定的	肯定的	非常に 肯定的
例 結婚		○	-3	-2	-1	0	+1	+2	+3
1 結婚			-3	-2	-1	0	+1	+2	+3
2 刑務所もしくは同等の施設への拘留			-3	-2	-1	0	+1	+2	+3
3 配偶者の死			-3	-2	-1	0	+1	+2	+3
4 睡眠習慣の大きな変化 (眠り過ぎ、もしくは不眠)			-3	-2	-1	0	+1	+2	+3
5 親しい家族の死			-3	-2	-1	0	+1	+2	+3
a. 母親			-3	-2	-1	0	+1	+2	+3
b. 父親			-3	-2	-1	0	+1	+2	+3
c. 兄・弟			-3	-2	-1	0	+1	+2	+3
d. 姉・妹			-3	-2	-1	0	+1	+2	+3
e. 祖母			-3	-2	-1	0	+1	+2	+3
f. 祖父			-3	-2	-1	0	+1	+2	+3
g. その他 ( )			-3	-2	-1	0	+1	+2	+3
6 食習慣の大きな変化 (食べ過ぎ、 もしくはほとんど食べない)			-3	-2	-1	0	+1	+2	+3
7 抵当権の損失、借金			-3	-2	-1	0	+1	+2	+3
8 親しい友人の死			-3	-2	-1	0	+1	+2	+3
9 個人の優れた業績			-3	-2	-1	0	+1	+2	+3
10 小さな法律違反 (交通違反、治安妨害など)			-3	-2	-1	0	+1	+2	+3
11 男性：妻/恋人の妊娠			-3	-2	-1	0	+1	+2	+3
12 女性：妊娠			-3	-2	-1	0	+1	+2	+3
13 勤務状況の変化 (異なる仕事上の責務、勤務条件 の大きな変化、勤務時間など)			-3	-2	-1	0	+1	+2	+3
14 新しい仕事			-3	-2	-1	0	+1	+2	+3

	0から 6か月前	7か月から 1年前	非常に 否定的	否定的	やや 否定的	影響 なし	やや 肯定的	肯定的	非常に 肯定的
15 親しい家族メンバーの深刻な病気 や怪我			-3	-2	-1	0	+1	+2	+3
a. 父親			-3	-2	-1	0	+1	+2	+3
b. 母親			-3	-2	-1	0	+1	+2	+3
c. 姉・妹			-3	-2	-1	0	+1	+2	+3
d. 兄・弟			-3	-2	-1	0	+1	+2	+3
e. 祖父			-3	-2	-1	0	+1	+2	+3
f. 祖母			-3	-2	-1	0	+1	+2	+3
g. 配偶者			-3	-2	-1	0	+1	+2	+3
h. その他 ( )			-3	-2	-1	0	+1	+2	+3
16 性的な問題			-3	-2	-1	0	+1	+2	+3
17 雇用主とのトラブル (失業の危機、停職、降格など)			-3	-2	-1	0	+1	+2	+3
18 法律上のトラブル			-3	-2	-1	0	+1	+2	+3
19 財務状況の大きな変化(良くなっ た、もしくは悪くなった)			-3	-2	-1	0	+1	+2	+3
20 家族の親密さに関する大きな変化 (親密さの増加、もしくは減少)			-3	-2	-1	0	+1	+2	+3
21 新しい家族が増えた(出産、養子、 家族が移してくるなど)			-3	-2	-1	0	+1	+2	+3
22 引越し			-3	-2	-1	0	+1	+2	+3
23 配偶者との別居 (高齢などの理由で)			-3	-2	-1	0	+1	+2	+3
24 宗教活動の大きな変化 (活動参加の増加、もしくは減少)			-3	-2	-1	0	+1	+2	+3
25 夫婦間の和解			-3	-2	-1	0	+1	+2	+3
26 配偶者との口論の回数大きな変 化(より多い、もしくはより少な い口論)			-3	-2	-1	0	+1	+2	+3
27 既婚男性: 妻の外での仕事の変 化(仕事を始める、辞める、転職な ど)			-3	-2	-1	0	+1	+2	+3
28 既婚女性: 夫の仕事の変 化(失業、新しい仕事を始める、退職 など)			-3	-2	-1	0	+1	+2	+3
29 普段の娯楽の種類とそれに費やす 金額の大きな変化			-3	-2	-1	0	+1	+2	+3
30 100万円以上の借金 (家の購入、事業など)			-3	-2	-1	0	+1	+2	+3
31 100万円以下の借金 (車やテレビの購入、奨学金など)			-3	-2	-1	0	+1	+2	+3
32 仕事を解雇される			-3	-2	-1	0	+1	+2	+3
33 男性: 妻/恋人の妊娠中絶			-3	-2	-1	0	+1	+2	+3
34 女性: 妊娠中絶			-3	-2	-1	0	+1	+2	+3
35 自身の深刻な病気や怪我			-3	-2	-1	0	+1	+2	+3

	0 から 6 か月前	7 か月から 1 年前	非常に 否定的	否定的	やや 否定的	影響 なし	やや 肯定的	肯定的	非常に 肯定的
36 社会活動の大きな変化 例：飲み会、映画鑑賞、訪問（参加の増加もしくは減少）			-3	-2	-1	0	+1	+2	+3
37 家族の居住状況の大きな変化 （家の新築、リフォーム、家や近隣環境の悪化など）			-3	-2	-1	0	+1	+2	+3
38 離婚			-3	-2	-1	0	+1	+2	+3
39 親しい友人の深刻な病気や怪我			-3	-2	-1	0	+1	+2	+3
40 退職			-3	-2	-1	0	+1	+2	+3
41 息子、娘が家を出る （結婚、大学などの理由で）			-3	-2	-1	0	+1	+2	+3
42 義務教育の終了			-3	-2	-1	0	+1	+2	+3
43 配偶者と離れる （仕事や旅行などの理由で）			-3	-2	-1	0	+1	+2	+3
44 婚約			-3	-2	-1	0	+1	+2	+3
45 恋人と別れる			-3	-2	-1	0	+1	+2	+3
46 初めて実家を出る			-3	-2	-1	0	+1	+2	+3
47 恋人との和解/仲直り			-3	-2	-1	0	+1	+2	+3

その他の最近、あなたの生活に影響を与えた経験を下記の欄に記入し、評価してください。

	0 から 6 か月前	7 か月から 1 年前	非常に 否定的	否定的	やや 否定的	影響 なし	やや 肯定的	肯定的	非常に 肯定的
48			-3	-2	-1	0	+1	+2	+3
49			-3	-2	-1	0	+1	+2	+3
50			-3	-2	-1	0	+1	+2	+3

セクション II

	0 から 6 か月前	7 か月から 1 年前	非常に 否定的	否定的	やや 否定的	影響 なし	やや 肯定的	肯定的	非常に 肯定的
51 大学、大学院、専門学校等の学術レベルの高い学校生活の始まり			-3	-2	-1	0	+1	+2	+3
52 学術レベルの同じ、別の大学/大学院などへの変更			-3	-2	-1	0	+1	+2	+3
53 大学の試験			-3	-2	-1	0	+1	+2	+3
54 大学寮や他の居住からの退去			-3	-2	-1	0	+1	+2	+3
55 重要な試験に失敗する			-3	-2	-1	0	+1	+2	+3
56 専攻科目の変更			-3	-2	-1	0	+1	+2	+3
57 単位取得に失敗する			-3	-2	-1	0	+1	+2	+3
58 単位取得をあきらめる			-3	-2	-1	0	+1	+2	+3
59 大学のサークルに参加する			-3	-2	-1	0	+1	+2	+3
60 大学に関する財政上の問題 （大学を続けるための十分なお金がない危険）			-3	-2	-1	0	+1	+2	+3

# 生理指標を用いた精神疾患に対する偏見の研究 (2)

## サーモグラフィによるアプローチ

Investigation of Prejudice Against Mental Illness Using Physiological Indices: Method by Thermography

(キーワード: 偏見, 生理指標, 体温変化)

(KEYWORDS: Prejudice, Physiological Index, Body Temperature Change)

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### 1. はじめに

精神疾患に対する偏見は, その低減が社会政策上重要な問題である。偏見の測定方法は質問紙による測定が主であるが, これには社会的望ましさの影響による回答の歪曲が考えられる。そのために, 社会的望ましさの影響が少ない潜在的認知測定が必要である。

本発表では, 精神疾患への偏見に対する, 生理指標を用いた潜在的測定手法の提案を行う。精神疾患に対する偏見には, 精神疾患患者の振舞や, 精神疾患に関連した事物に対する恐怖や驚愕といった, 非意図的な反応が関与していると考えられる。そこで, 精神疾患に関連した事物を呈示した際の体表温度や皮膚コンダクタンス反応の変化による, 被験者の偏見の測定手法の提案を行う。

### 2. 提案する偏見の測定手法

#### 2.1. 提案内容

本発表で提案する手法は, 偏見を受けている疾患と, 偏見を受けていない疾患とで, 疾患に関連した事物を呈示した際の体温変化と皮膚コンダクタンス反応の変化を比較するものである。偏見を受けている疾患に対する驚愕や恐怖といった非意図的な反応が, より急激な生理指標の変化に反映されると考えられる。

#### 2.2. 実施例の手続き

統合失調症に対する偏見を測定する実施例の説明を行

う。統合失調症と比較する疾患としては, 糖尿病を用いた。疾患に関連する事物としては当該疾患患者が描いたとされる絵を用い, 「統合失調症患者の描いた絵」であるとの教示とともに画像刺激を呈示することで, 精神疾患に対する非意図的な反応の生起を図った。

「生理指標を用いた精神疾患に対する偏見の研究(1)」で用いた画像刺激を, ランダムに統合失調症患者が描いた絵, もしくは糖尿病患者が描いた絵として割り当て, ブロックデザインで呈示を行った。各ブロックの開始前と終了後に体温測定を行い, 皮膚コンダクタンス反応は実験開始から終了まで連続して測定を行った。実施状況を Figure1 に, 手続きの流れの模式図を Figure2 に示す。なお, 発表当日には, 実施例のデータと分析結果についても報告を行う予定である。

#### 2.3. 装置

体温測定用赤外線サーモグラフィ装置として Thermo Shot F30 (NEC Avio 赤外線テクノロジー株式会社) を使用した。皮膚コンダクタンス反応測定のためには Polymate II (株式会社デジテックス研究所) を使用した。

#### 謝辞

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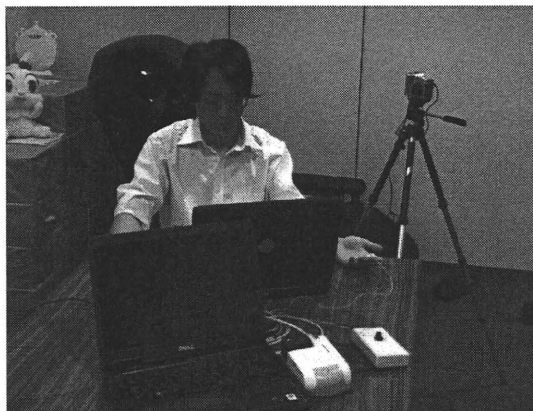


Figure 1. 実施状況

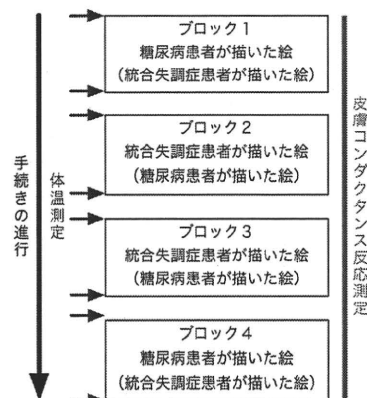


Figure 2. 手続きの流れ

## Perceptions of the Concept of Mutation among Family Members of Patients Receiving Outpatient Genetic Services and University Students

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**Abstract** Our objectives were to investigate: (1) relationships between perceptions of various terms regarding mutation and the depth of knowledge regarding mutation among family members of patients receiving genetic outpatient services, and (2) differences in perceptions of the term “gene mutation” for family members versus university students. Fifty-eight family members and 178 university students responded to two questionnaires: *Impressions regarding the term*, and *Knowledge about the concept of mutation*. Factor analyses were conducted to determine the factor structure of ratings of the terms, and two-way analyses of variance [(1)Term, (2)Group × Knowledge] were conducted to examine differences in perceptions of the terms as measured by scores for each

extracted factor. Family members had a significantly more negative perception of the term “gene mutation” than “gene change” and a less negative perception of the term “gene mutation” than “gene lesion”; they had significantly more negative perceptions of the term “gene mutation” than did university students.

**Keywords** Genetic counseling · Mutation · Knowledge · Health communication

### Introduction

The advent of medical genetics has produced a significant body of knowledge pertaining to the relationships between diseases and genes. As discoveries about genetic factors for disease continue to be made, it is expected that genetic-based medicine will become more routine in clinical settings (Collins and McKusick 2001). Drawing upon developments in medical genetics, geneticists and genetic counselors provide genetic information to their clients to help them “understand and adapt to the medical, psychological and familial implications of genetic contributions to disease” (Resta et al. 2006, p.79), often using technical terms with which clients are unfamiliar.

It has been shown that there are disparities between how lay people perceive medical and biological terms, and the actual definitions of these terms (Blake et al. 2004; Chapman et al. 2003; Cooke et al. 2000; Gittelman et al. 2004; Lucero et al. 2007). The same is true for genetic terms and concepts (Lanie et al. 2004). The term “mutation” is used frequently in genetic counseling, but is a complicated concept. In addition, the term “mutation” has become increasingly negative in its connotations because of an association with mutation in human genes as a result of

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nuclear radiation (Condit et al. 2002). A study of preference regarding four terms (mutation, variation, alteration, change) revealed that most participants had strongly negative responses to the term “mutation,” perceiving it as unintended, while they perceived the term “alteration” as intended (Condit et al. 2004).

In Japanese clinical settings, healthcare professionals typically use three terms: gene mutation, gene change, and gene lesion, to explain the concept of mutation to patients and family members. The term “gene mutation” is the technical term and is already used widely. The term “gene change” has a relatively neutral connotation, and “lesion” (“kizu” in Japanese) is thought to enable lay people to better understand mutation, as “lesion” is a familiar term to them. In a previous study, we showed that Japanese university students had different perceptions of commonly used terms in genetic consultations (“gene mutation,” “gene change,” and “gene lesion”), despite their definitions being virtually the same (Ando et al. 2008). Specifically, 175 university students and auditing students responded to two questionnaires: “Impressions regarding the term” which assessed respondent perceptions of the three Japanese terms (gene mutation, gene change, and gene lesion), and “Knowledge about the concept of mutation.” A factor analysis of respondents’ perceptions of the three terms yielded three factors: Value, Change Rate, and Intention. Participants were divided into three groups depending on their knowledge, and a two-way analysis of variance (Term  $\times$  Knowledge Group) was conducted on the score for each factor. Results showed that students had a more negative perception of the term “gene mutation” than the term “gene change” regardless of their knowledge about mutation, and knowledge regarding mutation affected perceptions of the extent to which “mutation” was intentional vs. unintentional. These results suggest the term “mutation” is perceived negatively by lay people, and healthcare professionals should be aware of this perception when using the term with patients and family members.

In our previous study (Ando et al. 2008), we assessed university students’ perceptions. However, genetic counseling clients are often individuals with a genetic disease and their family members. Their perceptions of the term “mutation” might differ from those of university students and therefore could provide important insight for genetic counselors and geneticists. In fact, very little research has been conducted concerning how affected individuals perceive the terminology used to discuss their conditions. Wakefield et al. (2007) compared preferences for terms describing a cancer-related mutation among three groups: the general community, clinicians, and patients who are at high-risk for cancer. The term “faulty gene” was preferred by the community sample. However, females, those affected by cancer, and those who felt cancer had a large

impact on their lives, preferred the terms “gene change” or “altered gene.” Clinicians showed equal preference for the terms “faulty gene” and “gene change.” The results revealed that situation-specific differences seem to affect term preferences.

Alternatives to the term “mutation” have been proposed because of these possible negative perceptions. However, because “mutation” is a familiar technical term for genetics service providers, many are likely to use it to explain genetic conditions to their clients in a clinical setting. Therefore, perceptions of the term “mutation” should be investigated further.

#### Purpose of the Present Study

There were two specific objectives of the current study. First, we investigated the relationship between perceptions of three terms: gene mutation, gene change, and gene lesion (all having similar meanings in Japanese), and the depth of knowledge regarding mutation among people with a family member(s) affected with or at risk for a genetic condition. In particular, we focused on a sample of family members of patients visiting pediatric genetic outpatient services. Second, we compared perceptions of the term “gene mutation,” as well as the depth of knowledge regarding mutation, between a sample of university students and the family members.

## Methods

### Participants

#### *Family Members of Patients*

Family members of patients using outpatient genetic services in Pediatrics at Kitasato University Hospital were provided with an explanation of the study. Seventy-two individuals from whom consent was obtained were given questionnaires and asked either to complete them immediately after their genetic appointment or to return them by mail. Responses were obtained from 63 individuals. We excluded five participants who returned incomplete questionnaires. Analyses included the remaining 58 individuals (mean age = 36.2 years; SD = 6.5 years; Range: 27 to 56 years.).

#### *University Students*

A sample of 178 undergraduate and auditing students were included in this study (Males,  $n=63$ ; Females,  $n=115$ ; age, mean = 19.8 years, SD = 3.0 years., Range: 18 to 54 years.). Data for 175 of the university students were obtained from

the previous study (Ando et al. 2008), and data for the remaining three students that were excluded from the previous study because of missing values were added anew. All participants were volunteers, and they were recruited from the pool of university students in the Faculty of Arts and Social Psychology.

## Questionnaires

### *Impression Regarding the Terms*

A 5-point, semantic differential scale containing 14 adjective pairs was used to assess participants' impressions of the terms "gene mutation," "gene change," and "gene lesion." Development of this questionnaire is described in detail in our previous study (Ando et al. 2008), and the items are contained in the Appendix.

### *Knowledge About the Concept of Mutation*

Thirteen questions were used to evaluate depth of knowledge regarding the concept of mutation. Participants were asked to read a passage and were then given a choice of "Correct," "Incorrect," or "Do not know" in regard to the passage. This questionnaire contained items such as "Changes in genes can lead to disease," and each represented item contained accurate information. A value of 1 was given for each correct answer and 0 for each incorrect answer. Details about this questionnaire are also described in our previous study (Ando et al. 2008).

### *Demographics and Medical Information*

The family members of patients were asked their gender, age, and education level, as well as the gender and age of the patient, their relationship to the patient, and the patient's diagnosis. The university students were asked about their gender and age.

## Procedures

The family members of patients who had visited genetic outpatient services in Pediatrics at Kitasato University Hospital between April 2006 and March 2007 were asked to participate in the study anonymously. Those individuals from whom informed consent was obtained were given the questionnaires and asked to rate their perceptions of three terms and to complete the questionnaires concerning their knowledge about the basic concept of mutation and their demographic characteristics. The university students also were asked to complete the questionnaires anonymously. Both groups of participants were told the purpose of this study was to investigate perceptions of the presented terms

used in clinical settings in Japan and knowledge about the concept of mutation. The study was conducted entirely in Japanese, and the results were translated into English. The institutional review board of Kitasato University School of Medicine and Hospital approved the study.

## Data Analyses

All data analyses were performed using SPSS statistical software version 16.0. Prior to the analyses of perceptions of the terms (see below), the family members and the university students were placed into two groups based upon the number of correct responses to the knowledge of the basic concept of mutation questionnaire. Using the median number of correct responses (family members'  $mdn = 8$ ; students'  $mdn = 9$ ), the participants who answered 3 to 8 questions correctly were placed in a "Low-knowledge group" (family members:  $n=35$ , students:  $n=77$ ), and those who answered 9 to 12 questions correctly were placed in a "High-knowledge group" (family member:  $n=23$ , students:  $n=101$ ). These two groups were created based on their relative level of knowledge, and not on absolute characteristics. Two-way analyses of variance (ANOVA) were conducted to analyze possible differences in age between the two groups (family members, students), and possible differences in the number of correct answers as a function of either group (family members, students) or age.

### *Analysis 1*

A factor analysis (varimax rotation) unconstrained for number of factors was used to identify the factor structure for ratings of the three terms (gene mutation, gene change and gene lesion) by family members of patients. The Guttman rule (an eigenvalue  $>1$ ) was used to determine the number of factors to retain for rotation (Guttman 1954). When identifying the factor structure, a factor score for each extracted factor was calculated, and then each extracted factor received a name. Next, a two-way ANOVA was conducted on each factor score using the two levels of knowledge (Low, High) and three terms (gene mutation, gene change, gene lesion), in order to identify the impact of knowledge level on perceptions of the three terms.

### *Analysis 2*

To reveal the factor structure of the term "gene mutation" for all participants (family members and students), we performed a factor analysis (varimax rotation) unconstrained for number of factors on perceptions of the term "gene mutation." The Guttman rule was used to determine the number of factors to retain for rotation (Guttman 1954). A factor score for each extracted factor was calculated and

each extracted factor received a name. Next, to determine the effects of knowledge level and group differences on perceptions of the term “gene mutation,” a two-way ANOVA with two levels of knowledge (Low, High) and two groups (family members, students) was conducted on each factor score.

## Results

### Family Members’ Demographic Characteristics and Patients’ Demographic and Medical Characteristics

Table 1 contains a summary of the demographic characteristics of family members and the demographic and medical characteristics of patients. In 20 of 59 patient cases, a definite diagnosis either was not found ( $n=7$ ), or the participants did not provide this information ( $n=13$ ). In most undiagnosed/no response cases, patients were affected with undefined multiple congenital anomalies and/or mental retardation. In no response cases, most participants left blank the column because they knew the patients were not definitely-diagnosed.

Table 2 shows age and numbers of correct answers on the knowledge questionnaire for the two levels of knowledge (Low, High) and the two groups (family members, students). There was a significant main effect due to group for age,  $F(1,232)=664.0$ ,  $p<.01$ , with family members of patients being older than students. There were no significant effects due to age on level of knowledge. Whether participants were family members of patients or university students did not significantly affect their number of correct answers,  $F(1,232)=3.585$ ,  $p=.06$ .

### Differences in Family Members’ Perceptions of the Three Terms (Results of Analysis 1)

We extracted four factors for ratings of the three terms, with a cumulative contribution rate of 49.3% (Table 3). We interpreted the factors as follows: Factor 1 (Desirability) is comprised of adjective pairs representing the concept of what is generally “desirable and undesirable,” Factor 2 (Suddenness of Change) is comprised of adjective pairs representing “sudden and not sudden” and “changing and unchanging,” Factor 3 (Growth) represents “development in response to nature,” and Factor 4 (Slow Intentionality) is comprised of adjective pairs representing the concepts of “intended and unintended” and “fast and slow.” Cronbach’s alpha coefficients for each factor were 0.89, 0.68, 0.65, and 0.29, respectively.

We calculated scores for each of the extracted factors and then applied two-way ANOVAs for each factor using as independent variables the knowledge level (Low,

**Table 1** Demographics of Family Members and Demographics and Medical Information of Patients

	<i>n</i>	Mean (SD)	Range
Family members of patients ( $N=58$ )			
Gender			
Male	11		
Female	47		
Age		36.2 (6.5)	27–56
Education level			
High school	19		
College	21		
University	14		
Graduate School	2		
No response	2		
Relationship to the patient			
Parent	57		
No response	1		
Patients ( $N=59^a$ )			
Gender			
Male	24		
Female	35		
Age		4.6 (6.0)	0–22
Diagnosis			
Down syndrome (including mosaic type)	14		
Sotos syndrome	6		
Prader-Willi Syndrome	5		
Unbalanced translocation	3		
Osteogenesis imperfecta syndrome	2		
Neurofibromatosis type I	2		
Achondroplasia	2		
Unbalanced translocation, Pierre Robin sequence	1		
Jeune syndrome	1		
Williams syndrome	1		
Cleid Cranial Dysplasia	1		
Noonan syndrome	1		
Undiagnosed	7		
No response	13		

<sup>a</sup> including siblings

High) and term (gene mutation, gene change and gene lesion). The results showed a significant main effect due to term,  $F(2,112)=11.962$ ,  $p<.01$ , partial  $\eta^2=0.176$ , for the Desirability factor (Fig. 1a). A post-hoc analysis (Bonferroni method) conducted on the main effect of the terms indicated that the Desirability factor score for “gene change” was significantly higher than for the other two terms ( $p<.05$ ). We also observed a main effect due to term for the Suddenness of Change factor,  $F(2,112)=5.181$ ,  $p<.01$ , partial  $\eta^2=0.085$ , because this factor score was higher for “gene mutation”



**Table 2** Distribution of Age and Numbers of Correct Answers on the Questionnaire in Two Levels of Knowledge (Low-Knowledge, High-Knowledge) and in Two Groups (Family Members of Patients, University Students)

	Family members of patients		University students	
	Low-knowledge group	High-knowledge group	Low-knowledge group	High-knowledge group
<i>n</i>	35	23	77	101
Age (Mean ± SD)	35.9±6.04	36.6±7.14	20.2±4.28	19.6±1.22
Number of correct answer (Mean ± SD)	6.4±1.56	9.8±0.98	6.7±1.31	10.4±1.23

than “gene lesion” (Fig. 1b). We also found a main effect due to term for the Growth factor,  $F(2,112)=10.032, p<.01$ , partial  $\eta^2=0.152$ , such that the Growth factor score for “gene lesion” was significantly lower than that for the other two terms (Fig. 1c). We found no significant differences between the groups for the Slow Intentionality factor (Fig. 1d). There were no significant main effects due to knowledge level for scores on any of the four factors.

Differences in Perceptions of “Gene Mutation” for Family Members versus Students (Result of Analysis 2)

We extracted four factors for the term “gene mutation,” with a cumulative contribution rate of 48.7% (Table 4). We interpreted Factor 1 as having positive–negative connotations and designated it as the Favorability factor. We interpreted Factor 2 as having developmental connotations and designated it as the Development factor. We interpreted Factor 3 as relating to the rate of change and designated it as the Change Rate factor. We interpreted Factor 4 as

having planned–unplanned connotations and designated it as the Intention factor. Cronbach’s alpha coefficients for each factor were 0.84, 0.68, 0.58, and 0.56, respectively.

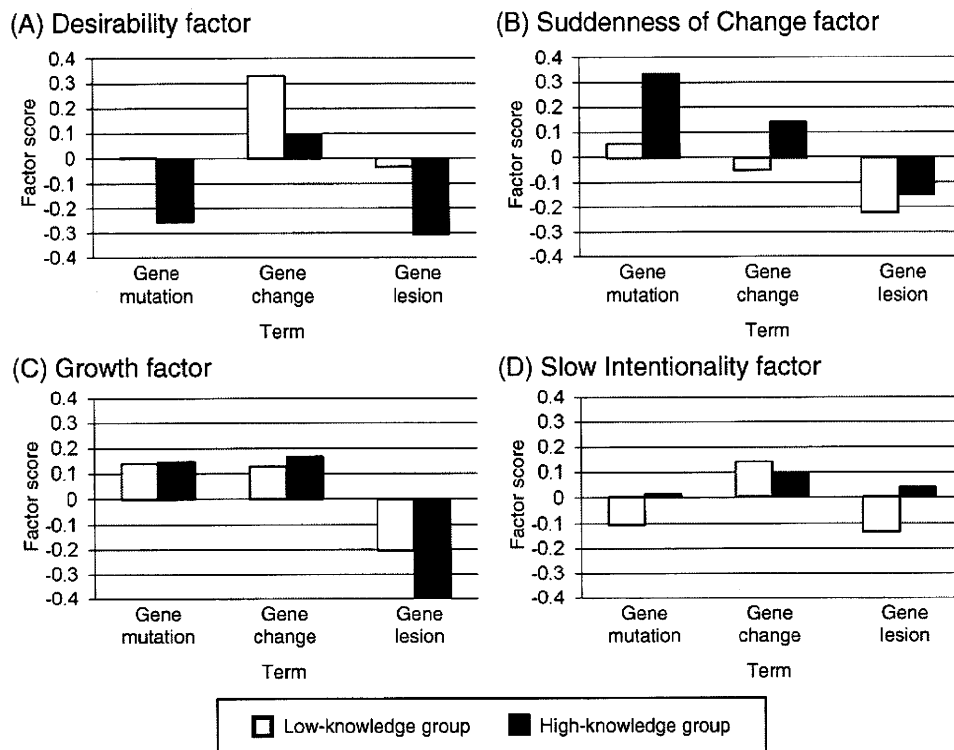
To assess possible effects of knowledge about mutation and group (family members, students) on perceptions of the term “gene mutation,” we conducted a two-way ANOVA [Knowledge level (Low, High) × group (family members, students)] on the scores for each extracted factor. We observed a significant main effect due to group on the Favorability factor,  $F(1,232)=9.915, p<.01$ , partial  $\eta^2=0.041$ , where the factor score for family members was lower than that for students (Fig. 2a). We also observed a significant main effect for group on the Development factor,  $F(1,232)=4.112, p<.05$ , partial  $\eta^2=0.017$ , where the factor score for family members was again lower than that for students (Fig. 2b). We found significant main effects for both knowledge level and group on the Change Rate factor,  $F(1,232)=5.863, p<.05$ , partial  $\eta^2=0.025$ , and  $F(1,232)=6.482, p<.05$ , partial  $\eta^2=0.027$ , respectively (Fig. 2c). Specifically, the factor score for the high-

**Table 3** Varimax Rotated Factor Loading of ‘Gene Mutation,’ ‘Gene Change,’ and ‘Gene Lesion’ in Impressions of the Family Members of Patients

	Factor 1 Desirability	Factor 2 Suddenness of Change	Factor 3 Growth	Factor 4 Slow Intentionality	Communality
Scary / Not scary	<b>-0.790</b>	0.260	-0.124	-0.060	0.710
Normal / Not normal	<b>0.763</b>	-0.344	0.103	0.166	0.739
Desirable / Undesirable	<b>0.698</b>	0.006	0.187	0.370	0.659
Avoidable / Unavoidable	<b>0.683</b>	-0.205	0.076	-0.027	0.515
Healthy / Unhealthy	<b>0.574</b>	-0.085	0.285	0.304	0.510
Good / Bad	<b>0.494</b>	0.021	0.255	0.350	0.432
Sudden / Gradual	-0.210	<b>0.772</b>	-0.022	-0.170	0.669
Changing / Unchanging	-0.155	<b>0.634</b>	-0.032	0.072	0.433
Able to develop / Unable to develop	0.371	-0.029	<b>0.644</b>	-0.324	0.659
Natural / Unnatural	0.224	-0.188	<b>0.522</b>	0.157	0.382
Evolved / Degenerate	-0.074	0.231	<b>0.473</b>	0.261	0.351
Adapted / Maladapted	0.267	-0.122	<b>0.455</b>	0.382	0.439
Intended / Unintended	0.131	0.109	0.095	<b>0.416</b>	0.211
Fast / Slow	-0.078	0.157	-0.019	<b>-0.400</b>	0.191
Cumulative contribution rate (%)	22.063	31.880	41.465	49.287	

The values in bold indicate the items loaded on each Factor.

**Fig. 1** Means for Family Member Factor Scores for Each Term: ‘Gene Mutation’, ‘Gene Change’ and ‘Gene Lesion.’ Means are Shown for Each Knowledge Group.



knowledge group was higher than that for the low-knowledge group, and the factor score for family members was lower than that for students. Lastly, we observed a main effect due to group for the Intention factor,  $F(1,232)=6.707, p<.01$ , partial  $\eta^2=0.028$ , such that the factor score for family members was lower than that for students (Fig. 2d).

**Discussion**

Family Members’ Perceptions of Terms Regarding Mutation

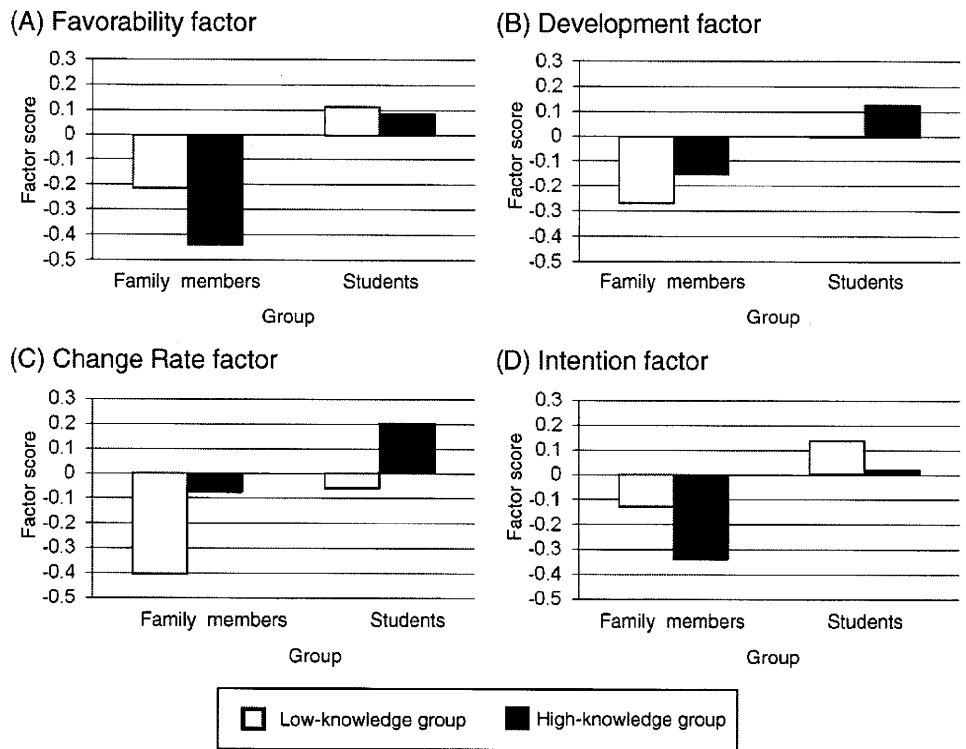
Factor analysis of the family members’ ratings of gene change, gene mutation, and gene lesion yielded four

**Table 4** Varimax Rotated Factor Loading of ‘Gene Mutation’ in Impressions of the Family Members of Patients and University Students

	Factor 1 Favorability	Factor 2 Development	Factor 3 Change Rate	Factor 4 Intention	Communality
Scary / Not scary	<b>-0.705</b>	-0.005	0.275	-0.145	0.594
Good / Bad	<b>0.702</b>	0.349	0.030	0.122	0.631
Healthy / Unhealthy	<b>0.697</b>	0.231	-0.167	0.047	0.569
Desirable / Undesirable	<b>0.660</b>	0.319	-0.038	0.305	0.632
Natural / Unnatural	<b>0.573</b>	0.207	-0.259	-0.237	0.494
Normal / Not normal	<b>0.540</b>	0.084	-0.397	0.134	0.475
Evolved / Degenerate	0.107	<b>0.857</b>	0.070	0.013	0.750
Adapted / Maladapted	0.352	<b>0.528</b>	-0.058	0.041	0.408
Able to develop / Unable to develop	0.180	<b>0.489</b>	0.015	0.088	0.279
Sudden / Gradual	-0.182	-0.030	<b>0.718</b>	-0.105	0.561
Changing / Unchanging	0.002	0.258	<b>0.564</b>	-0.123	0.400
Fast / Slow	-0.210	-0.087	<b>0.402</b>	-0.013	0.214
Avoidable / Unavoidable	0.105	-0.029	-0.121	<b>0.681</b>	0.490
Intended / Unintended	0.059	0.127	-0.071	<b>0.545</b>	0.321
Cumulative contribution rate (%)	19.932	31.868	41.544	48.695	

The values in bold indicate the items loaded on each Factor.

**Fig. 2** Means for Family Member and University Student Factor Scores for ‘Gene Mutation.’ Means are Shown for Each Knowledge Group.



factors. The Desirability factor contained the general “desirable–undesirable” concept; the Suddenness of Change factor contained the adjective pairs “sudden and not sudden” and “changing and unchanging”; the Growth factor represented development in response to nature; and the Slow Intentionality factor contained the adjective pairs “intended and unintended” and “fast and slow.” Comparison of these results to a factor analysis of ratings by university students in a previous study (Ando et al. 2008), indicates a different factor structure. Specifically, family members’ perceptions were best represented by the four factor model mentioned above, while students’ perceptions were best represented by a three factor model: Value, Change Rate, and Intention. It is particularly noteworthy that family members separated perceptions of growth from desirability, while university students’ perceptions of growth and desirability loaded onto the Value factor. In other words, the present sample of parents of children using outpatient genetic services viewed desirability as being different from growth. These differences between family members and students might be explained in part by the family members’ first-hand experiences as parents of a child with a genetic condition. For example, regarding the concept of mutation, perceptions of desirability might be based on parents’ theoretical views about the concept of mutation, while growth might be based on their first hand experiences with their children’s development. According to previous studies, experience with a disease may be related to perceptions of terms used to refer to “mutation.” For instance, Wakefield et al. (2007) found

language preferences were affected by having a personal history of cancer, and a similar trend was found in people with a strong family history of cancer. In a qualitative study of how parents first learned their child has Down’s syndrome and/or congenital heart disease, Garwick et al. (1995) found that family caregivers (who were mainly parents) clearly distinguished their personal, emotional reactions to the diagnosis from their understanding of how health care professionals informed them about their child’s condition. A comparison of these studies to the present investigation suggests that the theoretical and emotional views for disease might represent the Desirability factor, and first hand experiences of disease with their children’s development might reflect the Growth factor.

These findings indicate patients and family members that have personal experience with a genetic condition may have a more differentiated and nuanced set of perceptions regarding genetic mutation. Therefore, in a clinical setting, psychological counseling and education about the disease should involve assessments of individuals’ emotional reactions and their personal understanding of the genetic disease in question. Healthcare professionals should be aware that the Desirability factor and Growth factor separately contribute to perceptions of the concept of mutation among family members. Their word choices should support individuals’ cognitive appraisal of the genetic disease.

The results of the two-way ANOVA of scores on the Desirability factor and the Growth factor for “gene change”

revealed that they were higher than those for the other two terms, and they were lower for “gene lesion” than for the other two terms. However, the score for the Desirability factor for “gene mutation” was similar to that for “gene lesion,” while the score for the Growth factor for “gene mutation” was similar to that for “gene change.” Consistent with a previous report on a sample of university students (Ando et al. 2008), we conclude that “gene lesion” (lesion means “kizu” in Japanese) has a more negative connotation than the other two terms, regardless of an individual’s knowledge level. Use of a negative term might lead to psychological distress in genetic counseling clients that present with anxiety and stress (Chapple et al. 1997). We suggest healthcare professionals avoid using the term “gene lesion” (“idenshi-no-kizu” in Japanese) when introducing patients and families to the fact that they or their offspring carry a disease-associated allele.

#### Perceptions of “Gene Mutation”

Factor analysis for dependent groups’ (Analysis 2) ratings of “gene mutation” yielded four factors. The Favorability factor contained the positive–negative concept, the Development factor contained the developmental concept, the Change Rate factor contained the rate of change concept, and the Intention factor contained the planned–unplanned concept. The ANOVAs for each factor demonstrated that family members of patients have more negative perceptions of the term “gene mutation” than do university students, based on their lower scores for the Favorability and Development factors. Similarly, previous studies have demonstrated that people report strong negative reactions to the term “mutation” (Condit et al. 2004), they sometimes consider the terms “mutation” and “mutant gene” offensive (Hodgson et al. 2005), and they prefer the term “faulty gene” to the term “mutation” (Wakefield et al. 2007). The present findings and those of prior research indicate the need for a higher level of sensitivity when using the term “mutation” with family members of patients as well as with lay people.

A comparison of perceptions of the term “gene mutation” between family members of patients and university students revealed family members perceived the term as less intentional than university students, based on the students’ higher score for the Intention factor. Condit et al. (2000) found that both lay people and experts perceived the term “mutation” as unplanned and unintentional, but knowledge affected the perception of “mutation” as intentional in lay people (Ando et al. 2008). In the present study, however, we did not find a significant effect for knowledge for either group (family members, students) on intentionality factor. Inconsistency in the results for students between our previous study (Ando et al. 2008) and this study might be induced by a difference in analysis

strategy. In other words, students were divided into three groups based on their depth of knowledge in the previous study, and Low-knowledge group perceived mutation as being more intentional compared with Middle- and High-knowledge groups. In this study, however, constructions of groups were different from our previous study, so this difference might obscure a possible effect of knowledge. Further investigation of this matter should be required.

In a qualitative study of understanding of illness causality among British Pakistani (Shaw and Hurst 2008), some mothers worried that what they had done or what had happened to them during pregnancy might have caused their child’s problem. Such cases are common in clinical settings, and healthcare professionals typically explain that gene mutations can happen to anyone; it is not their “fault.” Such communication by clinical practitioners might partly explain the present finding that family members were less likely than university students to perceive the term “gene mutation” as intentional.

#### Implications of the Concept of Mutation

Factor analyses yielded four factors (Desirability, Suddenness of Change, Growth, Slow Intentionality) for the three terms (“gene mutation”, “gene change”, and “gene lesion”) among family members of patients in this study; three factors (Value, Change Rate, Intention) for the three terms among the university students in the previous study (Ando et al. 2008); and four factors (Favorability, Development, Change Rate, Intention) for the term “gene mutation” among the family members of patients and university students in this study. These factor structures resemble each other to some degree, but there are distinct differences. These differences might indicate the perception of “gene mutation” varies from the perceptions of other terms and, thus, the implications of the terms themselves are different.

Condit et al. (2004) demonstrated differences in the factor structure of four terms: mutation, alteration, variation, and change. “Mutation” had the same structure as “variation,” while “alteration” had the same structure as “change.” However, the two former terms had a different structure from the latter two terms. In a contextualized study of public discourse regarding mutation, Condit et al. (2002) suggested that “variation” or “alteration” might fail to convey a true sense of the important concept of “mutation,” although replacement terms have some merit. They concluded that care should be taken to clarify the concept of mutation and to enable people to understand it, even when using alternate terms, as terms other than mutation might obscure the meaning of the concept. Cotton (2002) similarly expressed concern with balancing sensitive language and scientific accuracy when suggesting use of the term “faulty gene” as an alternative expression.

Previous studies of “mutation” have identified it as a potentially stigmatizing expression likely to promote discrimination because many people have a strongly negative response to the term (Condit et al. 2004). Some researchers have shown that the term “mutant gene” is offensive and suggested careful word-choice might reduce the risk of labeling and stigmatization, as well as prevent unnecessary anxiety (Hodgson et al. 2005). In addition, the general public as well as clinicians have been found to be less likely to prefer the term “mutation” over “faulty gene” (Wakefield et al. 2007). Condit et al. (2004) previously suggested healthcare professionals avoid using the term “mutation” when disclosing to patients and families that they or their offspring carry a disease-associated allele. The present results strongly support these suggestions because family members of patients, more so than university students, demonstrated more negative perceptions regarding the term “mutation.” However, since “mutation” is the familiar technical term for a change in genetic information, genetic counseling clients are likely to hear the term “mutation” in contexts other than clinical settings with geneticists and genetic counselors. We suggest that whether or not healthcare professionals engaging in genetic counseling use the term “mutation” to explain genetic conditions to their clients at the appropriate time, they carefully explain the term and assess their clients’ understanding of their explanation.

We demonstrated in this study and a previous study (Ando et al. 2008) that knowledge level appears to affect some aspects of perceptions of the term “mutation.” To communicate effectively with lay people, experts should have a reasonably accurate idea of what they do and do not know about the topic (Nickerson 1999). They also should be sensitive to the fact that patients and their families may be experiencing a variety of emotions such as guilt, anger, worry, grief, and anxiety concerning these genetic conditions (McAllister et al. 2007).

In a study of parents’ responses to a child’s diagnosis of Neurofibromatosis (NF) 1, which has an unpredictable prognosis, Ablon (2000) compared the results to her previous study of dwarfism and suggested that the unpredictability of NF1 greatly contributed to parental fear and anxiety. Moreover, she suggested that parents were more likely to be influenced by a physician’s every literal word, compared to parents of children whose conditions were more benign or predictable. The quality of information about diseases is of great importance to the parents of patients, particularly with respect to perceptions of growth, and high quality information might mitigate parents’ confusion and anxieties. Healthcare professionals who provide genetic counseling to clients and who work in heredity clinics should be cautious with their words and disseminate accurate information based on an assessment of what clients know about the topic.

#### Typical Characteristics of “Mutation” in Japanese Language

Among family members, a two-way ANOVA revealed that ratings of the Suddenness of Change factor for “gene mutation” was higher than that for “gene lesion.” Moreover, among both family members and university students, the High-knowledge group perceived “gene mutation” as being more sudden and changing compared to the Low-knowledge group. One possible explanation of these results is that the technical term “mutation” in Japanese language includes the term “sudden.” That is, “*Totsuzen-Hen’i*” (Japanese technical term for “mutation”) includes the term “*totsuzen*” which means sudden. Japanese high school textbooks teach about the concept of mutation by using *Kankyo-Hen’i* (environmental variation) and *Totsuzen-Hen’i* (gene mutation, chromosomal mutation). The term “mutation” (presented to participants in the questionnaire as “*Hen’i*”) might have promoted a perception of “sudden.” Furthermore, the term “mutation” might have given the High-knowledge group a more distinct perception of “sudden,” compared to the Low-knowledge group, because “*Totsuzen-Hen’i*” is a technical term.

#### Study Limitations

Some limitations should be noted when interpreting the findings of this study. First, family members of patients with various types of genetic conditions were included in the sample. The breadth and genetic heterogeneity of genetic conditions among patients might affect individuals’ perceptions of the term “mutation.” However, possible differences due to individual diagnoses and severity of disease could not be determined statistically because of the small number of participants. In particular, a definite diagnosis was either not found or not reported for about one-third of patients, and most patients were affected with undefined multiple congenital anomalies and/or mental retardation. Thus, their prognoses are not clear, and their family members might be concerned about their children as their prognoses are unpredictable. The Growth factor in analysis 1 and the Development factor in analysis 2, in particular, might be influenced by diagnosis and severity of disease.

Second, the number of participants was small for statistical analyses, especially for family members of patients. The stability and reliability of the present results may be questionable, and therefore results should be interpreted with caution. In particular, the coefficient for the Slow Intentionality factor was very small and might represent a spurious result. Replications with more diverse and larger clinical populations would help to validate the findings obtained in this study. Additional limitations include age differences between family members of patients and

university students were which not controlled, and use of a survey with only Japanese terms. Connotations of the term “mutation” might be different between Japanese and other languages, and they might also differ across cultural groups.

**Implications for Practice and Future Research Recommendations**

The findings of the present study have important implications for geneticists and genetic counselors, and they may help to improve the care of patients and their families using genetic services. They suggest that healthcare professionals should be aware of the potential effects of their words and pay particular attention to word choice when explaining “mutation” to patients and their family members. Additionally, healthcare professionals should disseminate accurate information based on an assessment of what patients know about the topic and introduce the term “mutation” to explain genetic conditions at the appropriate time and with proper communication skills (e.g., being sensitive to and addressing any negative perceptions).

Given the limitations of the present study, further research on how to use the term “mutation” is needed. Perceptions of the term “mutation” tend to be negative, but other terms used to describe the concept of mutation exist, some of which have even more negative connotations. We

found that in the Japanese language, the term “lesion” has a more negative perception than the term “mutation”. In fact, there are other less perjorative terms which could be used to explain the concept of mutation. The present findings and those of previous studies (Condit et al. 2004; Ando et al. 2008) suggest that the term “change” is suitable. However, alternate terminology might create different perceptions (and possibly misperceptions) compared to the familiar technical term “mutation.” Therefore, researchers should investigate perceptions of the use of alternate terms in clinical settings in Japan, paying particular attention to how they are perceived and how accurately they represent the concept of “mutation.” Finally, future investigations should evaluate how patients with genetic conditions perceive the concept of “mutation.” Studies of actual patients will generate findings that are applicable for people who receive genetic counseling and help in developing communication skills of healthcare professionals that provide genetic counseling.

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**Conflicts of interest notification** The authors have no commercial association or conflicts of interest.

**Appendix: Questionnaire ‘Impression Regarding the Terms’**

What is your impression of the term, “gene \_\_\_\_\_ \* .”  
Please check the box which most represents your impression in each adjective set.

	Strongly agree	Agree	Neutral	Agree	Strongly agree	
1. Good	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	Bad
2. Sudden	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	Not sudden
3. Changing	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	Unchanging
4. Evolved	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	Degenerate
5. Abnormal	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	Normal
6. Natural	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	Unnatural
7. Avoidable	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	Unavoidable
8. Healthy	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	Unhealthy
9. Intended	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	Unintended
10. Adapted	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	Maladapted
11. Scary	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	Not scary
12. Desirable	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	Undesirable
13. Slow	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	Fast
14. Unable to develop	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	Able to develop

\* : One of three terms (mutation, change, lesion) is randomly selected and inserted.



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## Short Report

# Knowledge and impressions regarding the concept of mutation among Japanese university students

Ando N, Saito Y, Takemura K, Takada F, Iwamitsu Y. Knowledge and impressions regarding the concept of mutation among Japanese university students.

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Although the term *mutation* is frequently used in genetic counseling, it may carry negative connotations and create misunderstanding. Our objective was to investigate the relationship between the impressions regarding three Japanese terms *mutation of gene*, *change of gene*, and *lesion of gene* as well as to investigate the depth of understanding regarding mutation. A total of 175 university students and auditing students were included and responded to two questionnaires that were *Impressions regarding the term* in the semantic differential method and *Knowledge about the concept of mutation*. In factor analysis, three factors (Value, Change Rate, and Intention) were extracted. Participants were divided into three groups depending on their knowledge, and a two-way analysis of variance (Term × Knowledge Group) was conducted on the factor score for each. Results showed that the main effect of the 'Term' was significant for the Value Factor and that interaction was significant for the Change Rate Factor, and that the main effect of Knowledge Group was significant for the Intention Factor. The findings suggest that healthcare professionals should demonstrate an awareness of varying impressions of the different terms used to refer to the identical concepts of mutation. This is of particular importance when communicating with patients and their families.

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Key words: education – genetic counseling – health communication – knowledge – mutation – public understanding of science

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Significant new knowledge regarding the biological basis of heredity and the genetics of human traits has accumulated during the past 100 years. As discoveries continue to be made about genetic factors in diseases, it is anticipated that genetic-based medicine will become more routine in future clinical settings (1). With the development of medical genetics, geneticists and genetic counselors are increasingly providing genetic information to clients and helping them to adapt to the medical, psychological and familial implications of genetic contribution to disease (2) using unfamiliar technical terms.

It has been shown that medical and biological terms used in medical examinations are widely

misunderstood by many lay people (3–6) and that this is also similar with genetic terms and concepts (7). The term *mutation* is frequently used in genetic counseling, but mutation is a complicated concept that may carry negative connotations. The primary meaning of mutation is 'the action or process of changing (8)'. This definition is neutral and simply means changing biologically and physically. However, through time, the term *mutation* has become increasingly negative in its connotations, particularly because of the general fear of nuclear radiation in the public mind and the direct association between this fear and genetic mutation (9). A study on the bias against the four terms *mutation*, *variation*,

*alteration*, and *change* has revealed that most people had a strongly negative bias against the term *mutation* that was perceived as being an unintended change, whereas the term *alteration* was perceived as being an intended change (10). The authors suggested that healthcare professionals avoided using the term *mutation* when informing patients and families that they, or their offspring, carried a disease-associated allele. In an Australian study, some participants found the term *mutant gene* offensive and suggested that the careful choice of words might reduce the risk of labeling and stigmatization as well as prevent unnecessary anxiety (11). These results have highlighted that the term *mutation* has a negative connotation and have suggested that healthcare professionals should demonstrate awareness when using the term *mutation*.

Our objective was to investigate the relationship regarding the impression of three Japanese terms *mutation of gene*, *change of gene*, and *lesion of gene* (all having a similar meaning in Japanese) as well as to investigate the depth of understanding regarding mutation.

## Materials and methods

### Participants

Participants were 193 students in the Faculty of Arts at Kitasato University and Social Psychology at Waseda University who participated voluntarily. Of these, 188 responded to our questionnaire. We excluded four participants who had healthcare professional experiences and nine who filled out the questionnaires incompletely. Therefore, a total of 175 undergraduate and auditing students were included in this study (male:  $n = 62$ , female:  $n = 113$ , age: mean = 19.8 years, SD = 3.0 years, and range: 18–54 years).

### Questionnaires

#### *Impressions regarding the terms*

The three Japanese terms (*mutation of gene*, *change of gene*, and *lesion of gene*) were rated on 14 dimensions using a semantic differential scale with adjective sets (5-point scale). Before preparing the questionnaire, 18 lay people and 7 healthcare professionals (doctors, nurses, clinical technologists and speech therapists) were asked about their impressions regarding six Japanese terms (*mutation*, *change*, *lesion*, *mutation of gene*, *change of gene*, and *lesion of gene*; the last three terms have a similar meaning in Japanese). We selected 14 adjective sets based on these re-

sponses as well as adjectives that were used by Condit et al. (10). The 14 selected adjective sets were good/bad, sudden/not sudden, changing/unchanging, evolved/degenerated, normal/abnormal (not normal), natural/unnatural, avoidable/unavoidable, healthy/unhealthy, intended/unintended, adapted/maladapted, scary/not scary, desirable/undesirable, fast/slow and able to develop/unable to develop. Values given were 5 for the adjectives on the left and 1 for the adjectives on the right.

#### *Knowledge about the concept of mutation*

Thirteen items were designed to measure knowledge regarding the basic concept of mutation. Four of 13 items were original questions based on high school biology textbooks approved by the Japanese Ministry of Education, and 9 of 13 items were questions that were used by Condit et al. (10). Participants were asked to respond to the 13 questions by choosing one of the three responses (*true*, *false*, and *I don't know*). Value of 1 was given for correct answer (*all true*) and 0 for the others.

### Procedure

Participants were asked to anonymously rate their impressions regarding the three terms and to complete a questionnaire concerning the basic concept of mutation. The three terms were indicated randomly in the *Impressions regarding the terms* questionnaire to counterbalance the order effects. The procedure of the study was approved by institutional review board.

### Data and statistical analyses

Firstly, the percentage of correctly answered questions and the mean and standard deviation (SD) of the total number of questions answered correctly in the knowledge assessment questionnaire were calculated. Secondly, to reveal the factor structure of the three terms that connoted the same concept of changes in genetic information, we performed a factor analysis (varimax rotation) on impressions regarding the three terms. Finally, we calculated the percentage of questions answered correctly to assess the knowledge of the participants regarding mutation, and they were divided into three groups based on the total score for knowledge about mutation: low-knowledge group ( $n = 53$ , 3–7 correct answers), middle-knowledge group ( $n = 51$ , 8–9 correct answers) and high-knowledge group ( $n = 71$ ,

## Knowledge and impressions of mutation

10–13 correct answers) based on the 33.3 percentile (8 questions answered correctly) and the 66.7 percentile (10 questions answered correctly). To reveal whether knowledge about mutation had an effect on the impressions of the three terms, a two-way analysis of variance (ANOVA) [Term (*mutation of gene*, *change of gene*, and *lesion of gene*) × Knowledge Group (low, middle, and high)] was conducted on the factor score of each. Data analyses were performed using SPSS statistical software version 15.0.

### Results

#### Knowledge about the basic concept of mutation

The mean of correctly answered questions was 8.77 (percentage of correct answer: 67.7%) and SD was 2.24. The percentage of correct responses to each question in this study in comparison to Condit et al.'s study is shown in Table 1 (10). It can be seen that the correct response rate for 10 of 13 question statements was above 64.0%. However, correct response rates to the statements, 'changes in genes can occur over a lifetime', 'changes in genes can be caused by the sun', and 'changes in genes of somatic cells cannot be inherited' were below 37.1%.

#### Factor structure of the three terms *mutation of gene*, *change of gene*, and *lesion of gene*

Firstly, to reveal the factor structure of the three terms, an unconstrained factor analysis for the

number of factors was conducted dependently for the three terms. Three factors were extracted for the three terms, and the cumulative rate was 44.7% (Table 2). Factor 1 was interpreted as positive–negative connotations and was named the Value Factor. Factor 2 was interpreted as changing rate and was named the Change Rate Factor. Factor 3 was interpreted as going as planned and was named the Intention Factor. Cronbach's alpha scores for each factor were 0.87, 0.48 and 0.62, respectively.

#### Knowledge effect on the impression of the three terms

To reveal whether knowledge about mutation had an effect on impressions regarding the three terms, two-way ANOVA [Term (*mutation of gene*, *change of gene*, and *lesion of gene*) × Knowledge Group (low, middle, and high)] was conducted on the factor scores for each extracted factor. A significant main effect of Term was observed for the Value Factor ( $F_{2,344} = 212.4$ ,  $p < 0.01$ ). Post-hoc analysis (Bonferroni method) revealed that the factor score of the Value Factor for *lesion of gene* was significantly higher than those for *mutation of gene* and *change of gene* and that the factor score for the Value Factor of *change of gene* was significantly lower than those for *mutation of gene* and *lesion of gene* (Fig. 1a). The interaction of the Change Rate Factor (Term × Knowledge Group) was significant ( $F_{4,344} = 3.619$ ,  $p < 0.01$ ). The simple main effect of Knowledge Group on *mutation of gene*

Table 1. Understanding of the basic concept of *mutation*: the percentages of correct responses for each question are shown

Question	Question statements	Percentage of questions answered correctly	
		This study, $n = 175$	Condit et al.'s study (10), $n = 848$
Q1	Changes in your genes can be inherited <sup>a</sup>	78.9	81.0
Q2	Changes in genes can lead to disease <sup>a</sup>	92.6	83.1
Q3	Changes in genes can be caused by radiation <sup>a</sup>	88.6	76.2
Q4	Changes in genes can be caused by chemicals <sup>a</sup>	71.4	77.0
Q5	Every gene is able to mutate or change <sup>a</sup>	80.0	69.2
Q6	Chemicals can change the genes in the sperm of a man <sup>a</sup>	68.0	75.9
Q7	Chemicals can change the genes in the reproductive eggs of a woman <sup>a</sup>	65.1	76.6
Q8	Changes in genes can be spontaneously caused at a constant frequency <sup>b</sup>	64.0	
Q9	Changes in genes can occur over a lifetime <sup>a</sup>	27.4	63.3
Q10	Changes in genes can be caused by the sun <sup>a</sup>	37.1	41.6
Q11	Changes in genes of somatic cells cannot be inherited <sup>b</sup>	32.0	
Q12	Changes in genes concern biologic evolution <sup>b</sup>	88.0	
Q13	Some changes in genes cannot lead to disease <sup>b</sup>	84.0	
Percentage of common questions answered correctly		67.7	71.5

<sup>a</sup>Cross-questions with Condit et al. (10).

<sup>b</sup>Original questions in this study.

Table 2. Factor loading of *mutation of gene*, *change of gene*, and *lesion of gene* (varimax rotation): the values in bold indicate the items that loaded on each Factor

	Factor 1, Value	Factor 2, Change Rate	Factor 3, Intention	Communality
Good/bad	<b>0.836</b>	-0.040	-0.036	0.701
Healthy/unhealthy	<b>0.747</b>	-0.177	-0.103	0.600
Desirable/undesirable	<b>0.743</b>	-0.206	-0.024	0.595
Adapted/maladapted	<b>0.676</b>	0.027	-0.040	0.459
Scary/not scary	<b>0.619</b>	-0.333	-0.063	0.498
Normal/not normal	<b>-0.615</b>	0.268	0.034	0.452
Evolved/degenerate	<b>0.599</b>	-0.299	0.195	0.485
Natural/unnatural	<b>0.586</b>	0.131	0.039	0.361
Able to develop/unable to develop	<b>0.465</b>	0.130	0.022	0.233
Sudden/not sudden	-0.101	<b>0.577</b>	0.049	0.346
Fast/slow	-0.371	<b>0.474</b>	0.008	0.362
Changing/unchanging	-0.239	<b>0.395</b>	0.163	0.240
Avoidable/unavoidable	0.027	0.094	<b>0.719</b>	0.526
Intended/unintended	-0.075	0.048	<b>0.627</b>	0.402
Cumulative percent (%)	29.681	37.592	44.723	

was also significant ( $F_{2,172} = 7.202, p < 0.01$ ). That is, the middle- and high-knowledge groups had a significantly higher score than the low-knowledge group (Fig. 1b). Concerning the Intention Factor, the main effect of Knowledge-Group was significant ( $F_{2,172} = 7.909, p < 0.01$ ). The low-knowledge group had higher factor scores for the Intention Factor (Fig. 1c) than the middle- and high-knowledge groups (Fig. 1c).

**Discussion**

Knowledge about the basic concept of mutation

Participants' knowledge about the basic concept of mutation was significantly different for each question, with the correct response rate ranging between 92.6% and 27.4%. The correct response rate for 10 of 13 questions was above 64.0% and that for 3 of 13 questions was below 37.1%. When compared with Condit et al.'s results (10), the correct response rates to the nine questions

that were common to the two questionnaires were similar. However, close examination of details revealed differences in responses to Q9 (changes in genes can occur over a lifetime), and the rate of correct response for Q9 in our study (27.4%) was lower than that of Condit et al.'s study (63.3%) (10). The low correct response rate for Q10 and Q11, observed in both studies, indicated that lay people had little knowledge regarding somatic mutation. These results also indicated that Japanese university students in our study had less knowledge of somatic mutation compared with American students who participated in Condit et al.'s study. A comparison of Japanese and American high school biology textbooks indicated that American biology textbooks contain more information about genetics than Japanese textbooks (12, 13). In particular, American biology textbooks included information on cancer genetics and human genetics, but this was not the case in Japanese books. These facts suggested that the quality of biology

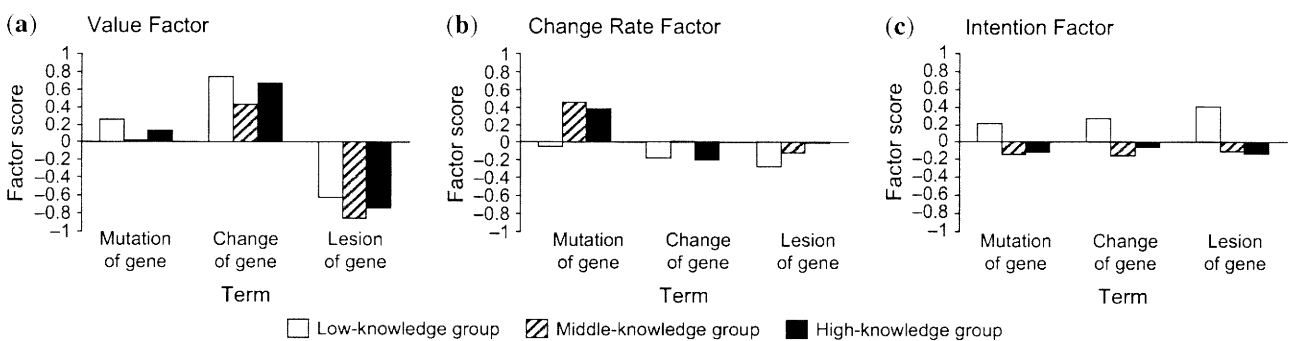


Fig. 1. Factor scores of *mutation of gene*, *change of gene*, and *lesion of gene*: means of the factor scores for the three terms in each knowledge group are shown.

education affected knowledge of genetics. Moreover, new educational guidelines from the Japanese Ministry of Education (2003) have deleted the topic of mutation from Biology Course 1 taken by many high school students. There is concern that lay people have impediments to understanding genetic diseases caused by mutation, which is a central point in genetic counseling. Moreover, it is essential to have knowledge regarding somatic mutation because cancers are often caused by somatic mutation. Knowledge regarding somatic mutation is very important when we consider genetic counseling for familial cancers.

Some studies assessing knowledge of genetics among lay people have revealed that knowledge of lay participants (14–16), patients (17) and their spouses (18) was limited. Moreover, even physicians, with the exception of geneticist (19, 20), had insufficient knowledge of genetics. Knowledge on genetics in physicians and lay people has become necessary because of the recent Human Genome Project as well as because of the discovery that genetic knowledge is helpful for human healthcare.

Factor structure of the three terms *mutation of gene*, *change of gene*, and *lesion of gene*

As the result of factor analysis, three factors were extracted for the three terms: Value Factor contained the general positive–negative concept, Change Rate Factor contained the static–dynamic concept and Intention Factor contained the accidental–intentional concept.

Results of two-way ANOVAs (Term × Knowledge Group) suggested different impressions of the three Japanese terms for the concept of mutation: *mutation of gene*, *change of gene*, and *lesion of gene*. Firstly, *lesion of gene* in Japanese had the most negative impression without relation to knowledge on Value Factor. According to the Japanese dictionary (21), the term *lesion* has the meaning of (i) physical damage or psychological damage, (ii) breach, break, cleft, fissure, fracture or split line, (iii) fault or imperfection and (iv) abasement, shame or disgrace. The term *lesion* (in Japanese: *Kizu*) includes concepts relating to physical wounds and also mental trauma. It seems that the negative impression of the term *lesion of gene* depended on the fundamental negative impression of the term *lesion* (*Kizu*). Japanese healthcare professionals might think that it is easy for lay people to understand mutation by using the term *lesion of gene* during genetic counseling, or at a heredity

clinic, because *Kizu* is a familiar term for lay people. But using a negative term might lead to psychological distress for clients with anxiety and stress who are recipients of genetic counseling (22). Healthcare professionals giving genetic counseling to clients and working in heredity clinics should keep in mind that clients and patients might have a negative impression of the term *lesion of gene* regardless of their knowledge. The Japanese term for *change of gene* resulted in the most positive impression without relation to knowledge on Value Factor, probably because the term *change* has a neutral impression.

Secondly, an interaction between Term and Knowledge was observed on Change Rate Factor such that the low-knowledge group had a lower impression of *sudden*, *fast*, and *changing* with *mutation of gene* on Change Rate Factor because *Totsuzen*, in the Japanese term for *mutation* (*Totsuzen-Hen'i*), means sudden. Japanese high school biology textbooks teach about the concept of mutation (*Hen'i*) by using *Kankyo-Hen'i* (environmental variation) and *Totsuzen-Hen'i* (gene mutation and chromosomal mutation), so it appears that middle- and high-knowledge groups had an impression of the term *Hen'i* that included sudden.

Finally, data on the Intention Factor indicated that the low-knowledge group perceived mutation as being more intentional compared with the middle- and high-knowledge groups, suggesting that the middle- and high-knowledge groups held a stronger belief that mutation is a natural phenomenon and therefore is unavoidable in comparison to the low-knowledge group. Condit and O'Grady (23) have explored interpretations of the term *mutation* held by lay and expert audiences. Both groups regarded mutations as being a variation but not as a planned or an intentional one. Experts regarded mutations as being necessary and not as being undesirable, whereas the lay group tended to be significantly less likely to see mutations as being necessary or as desirable. That the factor score for the Intention Factor, including intended/unintended and avoidable/unavoidable, was affected by knowledge about mutation suggests that the intentional impressions of the term *mutation* might be different in lay people. Differences of knowledge regarding the concept of mutation might affect the acceptance of the process of genetic diseases. Iwamitsu (24) has stated that it was important for patients and their families to be provided with at least a minimum amount of medical information about their diseases as soon as possible. This is also true in the area of genetic counseling and educational intervention.