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# Developing the roadmap for collaboration between patients and researchers about genome ELSI on clinical research and policy

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### Objective

- > Though the use of genomics, rare diseases (RDs) are an area of significant opportunity for improved diagnosis and treatment.
- → The way of recruiting patients and patient participation in genome research has slowly become more important and established at RD field in Japan.
- But RD patients don't have enough knowledge about genome research and ethical, legal, and social issues (ELSI) around genome research.
- Also genome researchers seldom have chances directly to meet RD patients and to understand their needs and issues about genome research.
- > Our aim is to develop the long-term roadmap for collaboration between patients and researchers about genome ELSI on clinical research and policy.

#### Method

- Collecting data from eligible participants (see below) in the following 2 ways:
   1) Through semi-structured interviews
- 2) Through our workshop series about patient participation in genome researchEligible participants:
  - Patient side : RD patients, their family members, and patient group/association leaders
- Researcher side : Skilled genome research and development personnel, clinicians, medical and/or ELSI researchers, and executive officers
  - Data analysis: Classified interview data and opinions from workshop series into related theme categories by content analysis method.
- > Developing the roadmap for collaboration with patients about genome ELSI on clinical research and policy:
- The authors discussed the long-term visions and action items of collaboration for genome research and its ELSI, and constructed a pilot version of roadmap. - Then we discussed this roadmap with both patient and researcher side and modified it referring to their opinions.
- Ethical consideration: We got an approval from ASrid IRB committee before starting to collect data.

#### Result

- Interview data from 23 people and opinions from 65 people participating workshop series were analyzed. (Table1)
- Researcher side includes genome analysts, R&D researchers, ELSI researchers, physicians, nurses, science communicators, and journalists.

Table 1. Background of research participant											
	Interview	WS1	WS2	WS3	WS4	Total					
Patient side, n	9	8	8	5	9	39					
Researcher side, n	14	9	8	7	11	49					
					WS: Workshop						

- As a result of content analysis, 6 categories and 33 subcategories about current recognition of collaboration for genome ELSI were identified. After repeated discussions by authors, we arranged these categories into 5 visions that should be accomplished in the next 10 years for the collaboration. (Figure 1)
- In the roadmap, the horizontal axis shows the targets in whom ELSI researchers and/or other entities intervene; Patient and Researcher side. The vertical one indicates the time and it is divided into 4 phases, each of which
- has a small goal to achieve. (Figure 2) Phase 1: ELSI researchers conduct surveys about the current situation and
- search previous literatures for the evidence of the following phases.
- Phase 2: ELSI researchers develop training programs and guidelines.
- Phase 3: ELSI researchers continuously execute training programs for both sides and issue a certificate for the completion of these training series to the person. Certificated people collaborate with each other as equal partners to promote.
- Phase 4: Certificated people conduct cooperative and collaborative activities for clinical research.
- According to the goals at each phase, we identified concrete items showing who does what to whom.

#### Conclusion

- This is the first roadmap in Japan for collaboration between RD patients and researchers about genome ELSI on clinical research and policy.
- We show 5 specific visions and concrete action items to achieve these visions in 4 phases in the next 10 years.
- We hope this roadmap facilitates further discussion about patient participation and the collaboration with researchers in genome research and its ELSI.



Asked about knowledge and understanding about genome research and

ELSI for participating in research, and the recognition of the other side.

Figure1 : Present issues about collaboration for genome ELSI, 5 goals to resolve these issues and basic ideas of the roadmap

	< Roadmap for	Collaboration wi	th Patients al	oout Geno	me ELSI to	Achieve "the	e 10 year visions" >					
	Toward Patients, Families, and						International Collabolations					
	ELSI researchers conduct survey	ELSI researchers conduct surveys about the current situation of both sides and search previous literatures for the evidence of the following phase										
<ul> <li>Phase 1 [1 year] Basic Survey</li> </ul>	[Conduct Surveys about P (DSurvey of recognition about the relating patients' disease experiences and gen (Survey of knowledge and attitude ab science, RD, medical application of gen (Survey of experience of the participal research	[Collect previous literatures] OSummarize the situation about Pt-side participation in genomic research and ELSI based on public information and literature. OExpose these info. on the web after obtaining permission.			[ Conduct Surveys about Ex-side ] (3)Survey of recognition about Pr-side participation in genomic research (2)Survey of experience of collaborating with Pt-side on genomic research.		Identify International Partners for Collaboration					
	ELSI researchers develop training programs and guidelines for providing knowledge and stimulating interests for collaboration about genome ELSI.											
h sides	Develop and Establish Training Programs for Both-Side]     [Accumulate Cases]     [ELSI researchers develop a prototype of Graining program based on the evidence obtained in Phase 1, evaluate, and improve It.     [Figure 1: 1] researchers     accumulate cases where     accumulate cases						41	Share and Disse- minate Issues				
rbot	[Pt-Side Training Programs for Patient P	mic Research]	collaboration with		ograms for Patients in			Import Good Foreign Cases Partners IRDIRC RDI UDN				
Phase 2 [3 years] Develop training programs and guidelines for both	Training for Knowledgel sarating terms Research Literacy Casearch Literacy General research ethics - ELSI Method of participation in research Basis in knowledge of genome Genomic research process Genome KLSI, patient rights and risks StProvide training via workshops, seminar	[Training for A <training g<br="">Pt-side can objective side of their disease (Visualize and verbal implicit experience) Pt-side can understa relationship betweer experience and genc and then can have a about research</training>	bals> experience. lize their and the a their pmic research,	Genomic Research) [Training for Koveldage] Chaining Items: why Hold of Needs, whit Menore and the set of the set of the set involvement in research How to explain genomic How to explain genomic informat consent from them. Informat consent from them. Informat consent from them. Informat consent from them. Informat consent from them. CLST executions and a set of the set ficture set of a set of the set of the ficture set of a set of the set of the set bod showed ELSI for Both-Bidling to knowledge and at attube which both- bod showed ELSI for Both-Bidling								
Develop traini	(ELSI researchers also examine an effic %Training programs are mainly for patien They can teach their association membra C Based on the activities above, ELSI m	ent way to deliver the t association leaders, irs about the contents Formulate a Guideline esearchers formulate	of the programs. of collaboration a ed a guideline at									
Fich	ELSI researchers continuously exec to promote discussion about ELSI.	ute training program	s for both side. T	hen Certificat	ed Pt/Ex-side	people collabora	te with each other as equal p					
 iate Inform	[Execute Training Programs Continuously] [Produce certificated people who complete the training programs] CELSI researchers execute training programs developed at phase 2 to both side and issue a certificate for the completion of training to the person.								Dissemin- ate and Export			
Execute Programs and Disseminate Information	by Certificate People Network) Certificate People Network Bits Involvedge of genome science, research process, ELSI, and initidet as a research participant to ordinary patients at association meetings and open lectures. Specific Cinical genomic research, certificated people network tells a significance of the research, risks, and rights to the association members.		Disseminate Information to Public) Construct website for archive the ELSI contents such as surveys, guidelines, cases, training session contents, etc. E.C. Existép seople can Case this website as a reference for their activity. Increase knowledge and interests of patients who don't belong to patient avoid on the total patient association.		e the ays, ession le can nce for erests	[Provide Closed Discussion Opportunities [Build a Network of Certificated Reopie] Build a relationship of mutual trust by sharing their knowledge and experience. OHoid case conferences about ELSI. Ocreate a broad range of personal connections from both sides.			the Programs			
Bar]	Certificated people conduct cooperative and collaborative activities for clinical research. ELSI researchers coordinate and follow-up both side as necess											
Phase 4 [2 year] Honase Cases International Deservation	[Pt-side can collaborate on genome ELSI matters of clinical research and/o policy etc. with Ex-side on the basis of patient standpoint]				hen Certificated 2 Network]	[Export Japanese Case of collaboration with Patients abo Genome ELSI] To IRDIRC, RDI, and UDN etc		about				
*Blue c	Blue cells on roadmap: Items for Pt-side. Red: for Ex-side. Yellow: for both sides. Purole: for international collaboration, and arean cells summarize the contents of each phase.											

Figure2 : The roadmap for collaboration between patients and researchers about genome ELSI

on clinical research and policy (for further discussion)

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