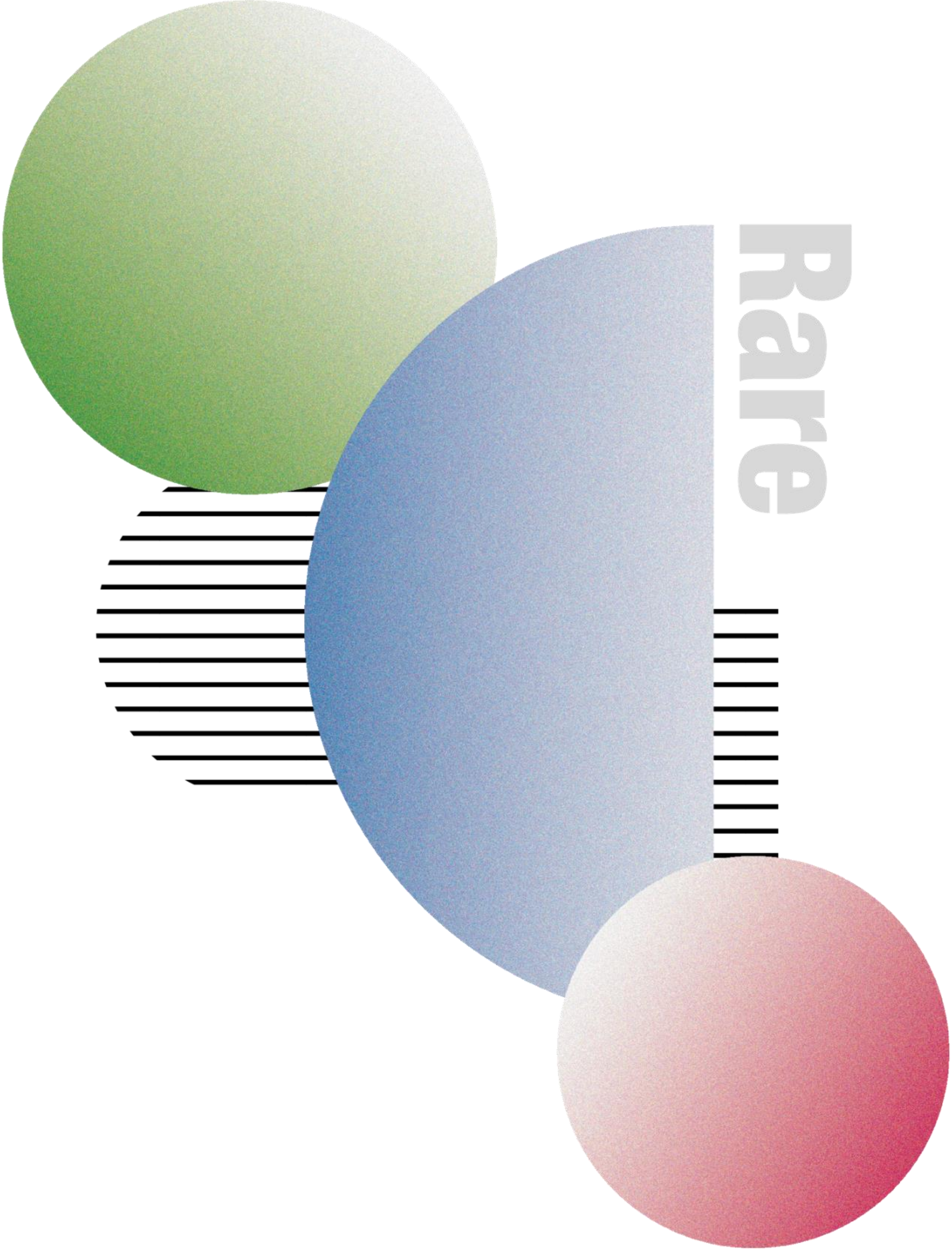

Survey on challenges faced by healthcare professionals in rare diseases [Data part]

Initiative on Rare and Undiagnosed Diseases (IRUD)
Rare Disease Consortium Japan (RDCJ)
Japan Pharmaceutical Manufacturers Association (JPMA)

IRUD Rare Disease Consortium Japan JPMA

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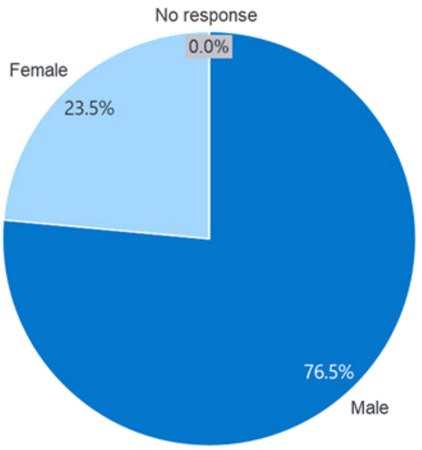
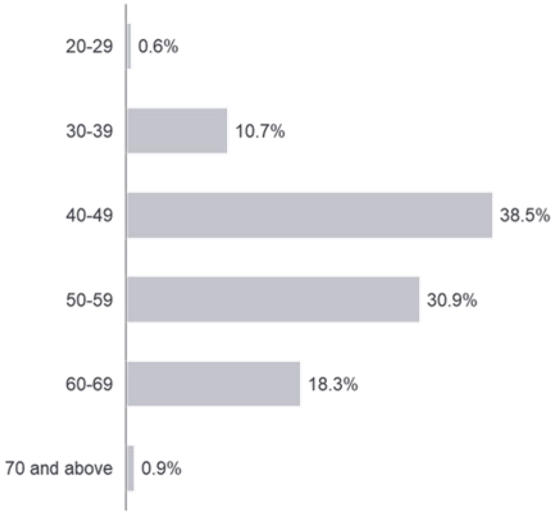
3. Survey methodology overview: Respondent demographics

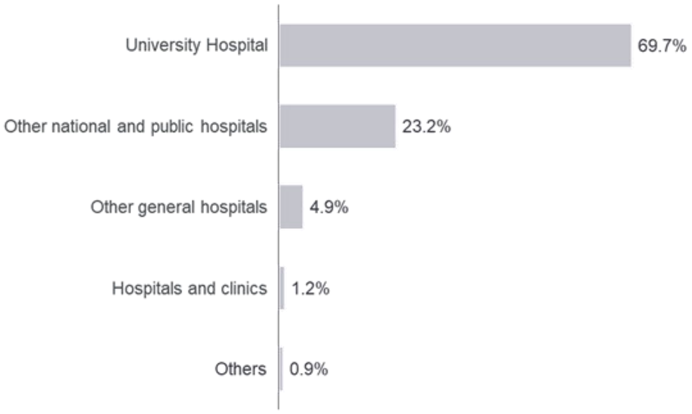
Gender



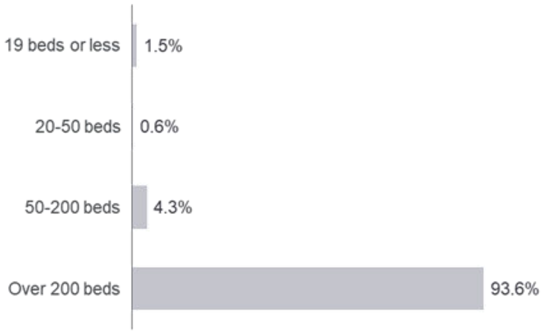
Age

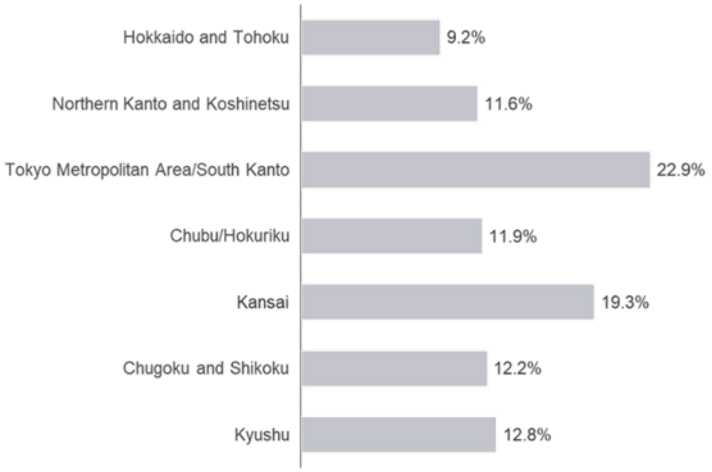


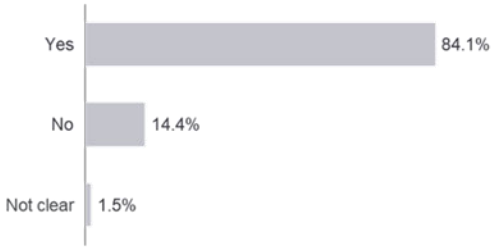
Type of facility

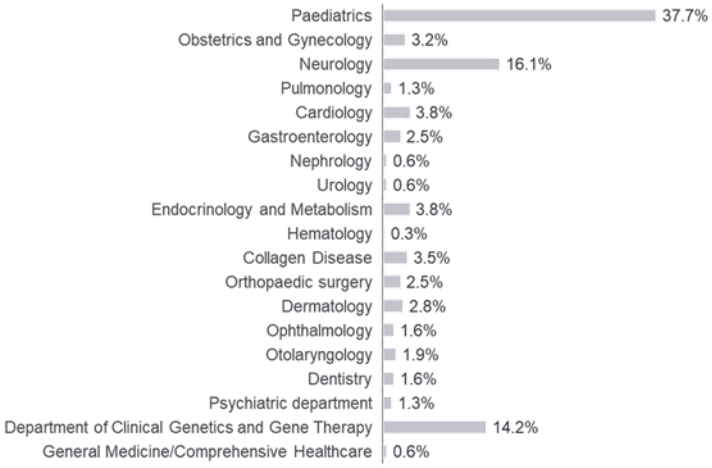


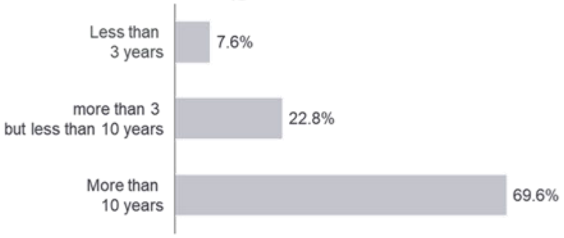
Number of hospital beds at affiliated facilities

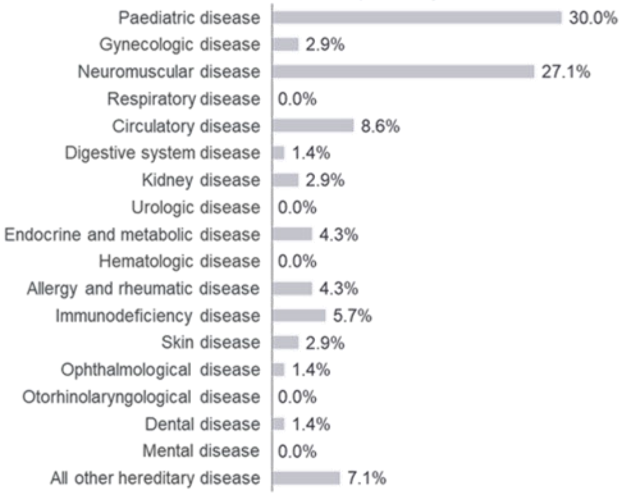


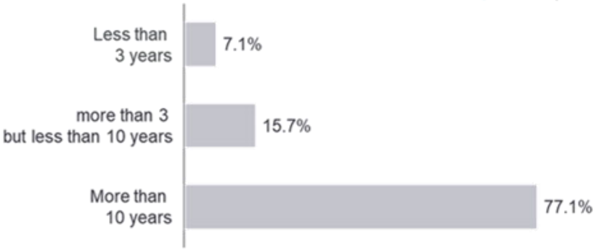

Region/area of affiliated facility

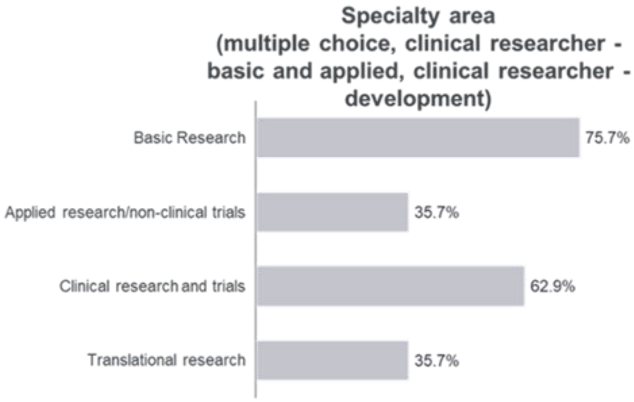

Availability of specialized staff at affiliated facilities


Departments
Specialists, non-specialists, other HCPs (genetic counselors, nurses)


Years of experience in rare disease treatment for support specialists, non-specialists, and other HCPs (genetic counselors and nurses)


Disease area (Clinical researcher - basic and applied, clinical researcher - development)


Years of experience in rare disease R&D (Clinical researcher - basic and applied, clinical researcher - development)



Specialty area (multiple choice, clinical researcher - basic and applied, clinical researcher - development)

Major rare diseases involved in the past year (disease name and number of respondents) *These results are based on the names of diseases reported by healthcare professionals

#	Disease name	Number of respondents [people]
1	Amyotrophic lateral sclerosis	24
2	Spinocerebellar degeneration	22
3	Noonan syndrome	18
4	Muscular dystrophies	18
5	Marfan's syndrome	15
6	Spinal muscular atrophy	15
7	Fabry's disease	14
8	Duchenne muscular dystrophy	14
9	mitochondrial disease	13
10	achondroplastic dwarfism	13
11	Hypophosphatasia	13
12	Sotos syndrome	12
13	Ehlers-Danlos syndrome	12
14	Kabuki Syndrome	12
15	Rett syndrome	12
16	Parkinson's disease	11
17	Familial Mediterranean Fever	11
18	Neurofibromatosis	11
19	Osler disease	10
20	Coffin-Siris syndrome	9
21	Tuberous sclerosis	9
22	Osteogenesis imperfecta	9
23	Mucopolysaccharidosis type 2	9
24	Short limbs	9
25	CHARGE syndrome	8
26	Williams Syndrome	8
27	Myasthenia gravis	7
28	Hypertrophic cardiomyopathy	7
29	22q11.2 deletion syndrome	6
30	Prader-Willi syndrome	6
31	Huntington's disease	6
32	Mucopolysaccharidoses	6
33	Multiple system atrophy	6
34	Dravet syndrome	5
35	MELAS	5
36	Alport's syndrome	5
37	Citrin deficiency	5
38	Epileptic encephalopathy	5
39	Spinal and bulbar muscular atrophy	5
40	Xeroderma pigmentosum	5
41	Multiple sclerosis	5
42	Pulmonary arterial hypertension	5
43	Epidermolysis bullosa	5
44	Fukuyama type congenital muscular dystrophy	5
45	Von Hippel-Lindau disease	5
46	cardio-facio-cutaneous syndrome	4
47	Hereditary transthyretin (ATTRv) amyloidosis	4
48	MEN	4
49	OTC deficiency	4
50	Rubinstein-Taybi syndrome	4
51	Wilson's disease	4
52	XLH	4
53	Angelman Syndrome	4
54	Cryopyrin-associated periodic syndrome	4
55	Epilepsy	4
56	Nephronophthisis	4
57	Paraganglioma	4
58	Rett's syndrome	4
59	Dilated cardiomyopathy	4
60	Primary immunodeficiency disease	4
61	Progressive supranuclear palsy	4
62	sex differentiation disorder	4
63	Congenital malformation syndrome	4
64	Congenital disorder of glycosylation	4
65	Congenital myopathy	4
66	Corticobasal degeneration	4
67	Glycogen storage disease	4
68	Inclusion body myositis	4
69	Baraitser-Winter syndrome	3
70	FGF23-related hypophosphatemic osteomalacia	3
71	MECP2 Duplication Syndrome	3
72	Pompe's disease	3
73	VEXAS syndrome	3
74	WDR1 Deficiency	3
75	Down Syndrome	3
76	Phenylketonuria	3
77	Prion disease	3
78	Bloom Syndrome	3
79	Propionic acidemia	3
80	Moyamoya disease	3
81	Lysosomal storage disease	3
82	Rosmund-Thomson syndrome	3
83	Hereditary amyloid polyneuropathy	3
84	Hereditary dystonia	3
85	Primary sclerosing cholangitis	3
86	Autoinflammatory syndrome	3
87	Hereditary diffuse leukoencephalopathy with spheroid	3
88	Vertebral amorphosis	3
89	Congenital under-loss of GPI	3
90	Nephrotic syndrome	3
91	Chromosomal abnormality	3
92	Congenital cerebral hypomyelination	3
93	Craniofacial dysostosis	3
94	cystic fibrosis	3
95	Pneumocyliosis	3
96	Adrenoleukodystrophy	3
97	common variable immunodeficiency	3
98	Aniridia	3
99	Systemic lupus erythematosus	3
100	Trisomy 18 syndrome	2

#	Disease name	Number of respondents [people]
101	Aarskog-Scott syndrome	2
102	Aicardi-Goutiéres syndrome	2
103	ATRX Syndrome	2
104	CLIFFORD syndrome (NALCN)	2
105	Fanconi syndrome	2
106	HBOC	2
107	HNRNP disease	2
108	IgG4-related disease	2
109	Marinesco-Sjogren syndrome (SIL1)	2
110	MEN1	2
111	MSA	2
112	Schaaf-Yang syndrome	2
113	Systemic scleroderma	2
114	Weaver syndrome	2
115	X-linked hypophosphatemic rickets	2
116	Usher syndrome	2
117	Imprinting Diseases	2
118	Galactosalidosis	2
119	Klippel-Trenonnay-Weber syndrome	2
120	Crohn's disease	2
121	Cockayne syndrome	2
122	Cornelia Derange's syndrome	2
123	Joubert syndrome	2
124	Sturge-Weber syndrome	2
125	Valde-Biedl's syndrome	2
126	Brugada syndrome	2
127	Beckwith-Wiedemann syndrome	2
128	Porphyria	2
129	Myopathy	2
130	Mitochondrial encephalomyopathy	2
131	Methylmalonic acidemia	2
132	Lipoid adrenal hyperplasia	2
133	Label's hereditary optic neuropathy	2
134	Loeys-Dietz syndrome	2
135	Waardenburg's syndrome	2
136	Hereditary neuropathy	2
137	Hereditary deafness	2
138	Familial hemophagocytic syndrome	2
139	Oculocutaneous albinism	2
140	Facioscapulohumeral muscular dystrophy	2
141	Pseudoparathyroidism	2
142	Ankylosing spondylitis	2
143	Vascular Ehlers-Danlos syndrome	2
144	Eosinophilic gastrointestinal disorder	2
145	Restrictive cardiomyopathy	2
146	Left ventricular densification disorder	2
147	Relapsing polychondritis	2
148	Insufficiency of the limbs	2
149	Fatty acid metabolism disorders	2
150	Neuromyelitis optica spectrum disorder	2
151	Autoimmune hepatitis	2
152	Severe combined immunodeficiency	2
153	Palmoplantar keratoderma	2
154	Cardiac amyloidosis	2
155	Progressive familial intrahepatic bile stagnation	2
156	Nephrogenic diabetes insipidus	2
157	Neuropsychiatric retardation	2
158	acromegaly	2
159	Congenital diaphragmatic hernia	2
160	Congenital hypopituitarism	2
161	Congenital ichthyosis	2
162	Congenital myasthenia	2
163	Congenital hyperinsulinemia	2
164	Congenital adrenal hypoplasia	2
165	Anterior segment ocular dysgenesis	2
166	Frontotemporal dementia	2
167	Idiopathic interstitial pneumonia	2
168	Spina bifida	2
169	Adrenocortical carcinoma	2
170	Chronic inflammatory demyelinating polyneuropathy	2
171	Chronic granulomatosis	2
172	Anhidrotic ectodermal dysplasia	2
173	Retinal pigment degeneration	2
174	Disorders of organic acid metabolism	2
175	Gliform droplet corneal dystrophy	2
176	Trisomy 13 syndrome	1
177	Chromosome 15 tetrasomy	1
178	Chromosome 15 marker gene	1
179	17q12 demented syndrome	1
180	1p36 micro-deficiency dementia syndrome	1
181	22Q11.3 Deficit Dementia Syndrome	1
182	2q17 Minimal Deficiency Syndrome	1
183	Type 2 collagen disorder	1
184	3Q21 Microdeletion Syndrome	1
185	3q29 is demented syndrome	1
186	4p deletion syndrome	1
187	Partial deletion of chromosome 4 long arm	1
188	5P-Syndrome	1
189	Trisomy mosaic 9	1
190	A20 Haploinsufficiency	1
191	ADNP-related disorders (ADNP gene abnormalities)	1
192	ADTKD	1
193	Alagille syndrome	1
194	AMeDS	1
195	ARID1B Related Disorders	1
196	ATR-X	1
197	Bainbrdge-Ropers syndrome	1
198	BO Syndrome	1
199	BPAN	1
200	CACNA1A related disorder	1

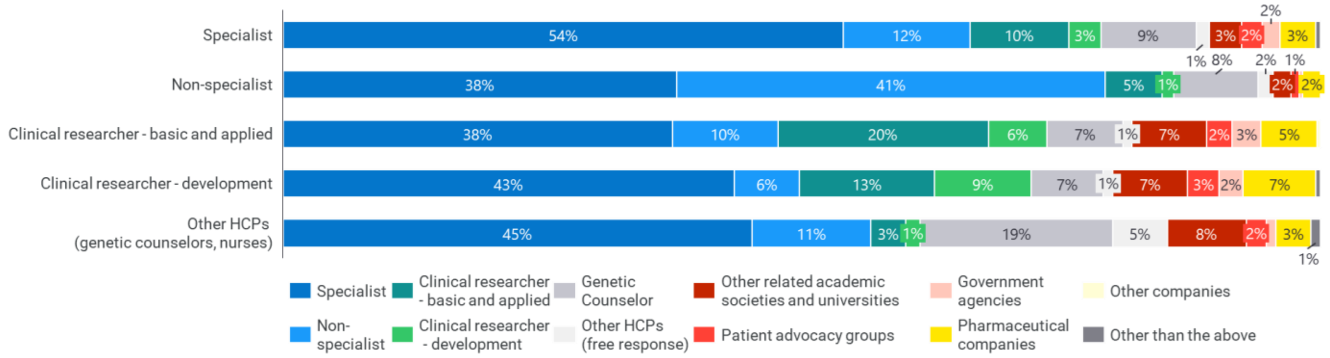
#	Disease name	Number of respondents [people]
201	CALJA	1
202	Dysbacteriosis CASK	1
203	CHDED	1
204	CMT1A	1
205	Craniolenticulosutural dysplasia	1
206	DM1	1
207	FG syndrome	1
208	FLNA-associated periventricular nodular heterotopia	1
209	Floating Harbor syndrome	1
210	Good syndrome	1
211	Gorlin syndrome	1
212	GPI anchor deficiency	1
213	GRIN1 Gene-Related Disorders	1
214	GSDIa	1
215	GSDIb	1
216	Hb hammersmith	1
217	HDLS	1
218	HLRCC	1
219	HNRNPK disorder	1
220	HTLV-1 associated myelopathy	1
221	HTRA1-associated cerebral small vessel disease	1
222	hypomyelinating leukodystrophy-24 (HLD24)	1
223	IgA vasuculitis	1
224	Intellectual disability-facial dysmorphism syndrome	1
225	Jubert syndrome	1
226	KID syndrome	1
227	KMS	1
228	L1 syndrome	1
229	LRBA deficiency	1
230	LZTR1-related disorder	1
231	Malan syndrome	1
232	MAPK8IP3-related neurodevelopmental disorders	1
233	MBD5 gene duplication	1
234	MCAD deficiency	1
235	MCT8 Disorders	1
236	MECOM-related diseases	1
237	MED13L	1
238	Megalencephaly-Polydactyly syndrome	1
239	Microcephaly, seizures, and developmental delay(PNKP gene)	1
240	MODY (Familial Onset Diabetes of the Young)	1
241	MOG antibody-associated neuropathy	1
242	Myhre syndrome	1
243	NBIA(BPAN)	1
244	NCL	1
245	NF1	1
246	NLRC4 Defects	1
247	NMDAR encephalitis	1
248	OZEMA (Oocyte-zygote-embryo maturation arrest)	1
249	PALLISTER-KILLIAN SYNDROME	1
250	Phelan-McDermid syndrome	1
251	PIC3CA	1
252	Pitt-Hopkins syndrome	1
253	PKU	1
254	PLCG2 Disorders	1
255	MCSZ due to PNKP gene mutation	1
256	Potocki-Lupski syndrome	1
257	PROD3 Genetic Disorders	1
258	PSP	1
259	Long QT syndrome	1
260	Rasopathy	1
261	RhoBTB2-associated neurodevelopmental disorders	1
262	Ritscher-Schinzel syndrome	1
263	ROHHAD SYNDROME	1
264	RORγT deficiency	1
265	Russell-Silver syndrome (maternal UPD7)	1
266	SAVI	1
267	SCA6	1
268	SCA8	1
269	SCN8A-Related Developmental and Epileptic Encephalopathy	1
270	SENDA/BPAN	1
271	SETD5 Gene Disorders	1
272	SHOX Disorders	1
273	Shwartz Jampel Syndrome	1
274	small vessel deisease with or without ocular anomalies	1
275	SPG80	1
276	Shprintzen-Goldberg syndrome	1
277	Stickler syndrome	1
278	TAFRO Castleman disease	1
279	TAFRO SYNDROME	1
280	Takenouchi-Kosaki syndrome	1
281	Temple syndrome	1
282	VCTERL Union	1
283	Ververi-Brady syndrome	1
284	Vici syndrome	1
285	Walfram syndrome	1
286	WDR45 Abnormality (BPAN)	1
287	Wiedemann-Steiner syndrome	1
288	Xia Gibbs syndrome	1
289	X-linked syndromic neurodevelopmental disorders	1
290	X-linked severe complex immunodeficiency	1
291	ZTTK syndrome	1
292	Amino acid metabolism disorders	1
293	Argininosuccinic aciduria	1
294	West Syndrome	1
295	Werner syndrome	1
296	Ulrich-type congenital muscular dystrophy	1
297	AIDS-related complex	1
298	Albright's syndrome	1
299	Occipital Horn Syndrome	1
300	Ornithine transcarbamylase deficiency disease	1

#	Disease name	Number of respondents [people]
301	Carney complex	1
302	CADASIL	1
303	catecholamine-induced polymorphic ventricular tachycardia	1
304	Galactosemia type IV	1
305	Carbamyl phosphate synthase 1 deficiency	1
306	Kallmann syndrome	1
307	Galloway-Mowat syndrome	1
308	Cushing's disease	1
309	Kriefstra syndrome	1
310	Klippel-Feil syndrome	1
311	Glucose transporter type 1 deficiency	1
312	Hypoglycemia due to glucokinase gene abnormalities	1
313	Cretinism	1
314	Creutzfeldt-Jakob disease	1
315	Gaucher's disease	1
316	Gorin syndrome	1
317	Costello's syndrome	1
318	sarcoidosis	1
319	Sialidosis	1
320	Sjogren's syndrome	1
321	Cystine storage disease	1
322	Cystinuria	1
323	Dystonia	1
324	Dystrophinopathy	1
325	Silver Russell syndrome	1
326	sudanophilic leukodystrophy	1
327	Stevens-Johnson syndrome	1
328	Swyer syndrome	1
329	Other autosomal abnormalities	1
330	Turner's syndrome	1
331	Thanatoholic osteodysplasia	1
332	Treacher Collins syndrome	1
333	Niemann-Pick disease type C	1
334	Nemaline myopathy	1
335	Barth's syndrome	1
336	Birt-Hogg-Dube syndrome	1
337	Byrne syndrome	1
338	Hutchinson-Gilford syndrome	1
339	Paramyotonia	1
340	Hyaline fibroma syndrome	1
341	Pitt-Hopkins syndrome	1
342	Hirschsprung's disease related diseases	1
343	Fanconi's anemia	1
344	Blount's disease	1
345	Freeman-Sheldon syndrome	1
346	Prolactinoma	1
347	Behcet's disease	1
348	Bethlem myopathy	1
349	Becker muscular dystrophy	1
350	Homocystinuria	1
351	Myotubular myopathy	1
352	Menkes' disease	1
353	Mendelian genotype Mycobacterium susceptible to infection	1
354	Molybdenum coenzyme deficiency	1
355	Mowat Wilson Syndrome	1
356	Ewing's sarcoma	1
357	Lasopathy	1
358	Libman-Sacks endocarditis	1
359	Liframeny syndrome	1
360	Lynch syndrome	1
361	Lymphoproliferative disorder	1
362	Lymphangioleiomyomatosis	1
363	Lennox-Gastaut syndrome	1
364	Subacute necrotizing encephalomyelopathy	1
365	Ectopic ACTH-producing thymic carcinoids	1
366	Hereditary phosphorus metabolism disorders	1
367	Hereditary spherocytosis	1
368	Hereditary coagulation factor deficiency	1
369	Hereditary angioedema	1
370	Hereditary thrombocytopenia	1
371	Hereditary thrombotic tendency	1
372	Hereditary autoinflammatory diseases	1
373	Hereditary periodic paralysis	1
374	Hereditary neuromuscular diseases	1
375	Hereditary spinocerebellar degeneration	1
376	Hereditary hematopoietic disorders	1
377	Hereditary multiple exostosis	1
378	Hereditary intellectual disability	1
379	Hereditary dementia	1
380	Relentless smooth tendon swelling syndrome of renal cells	1
381	Rhabdomyosarcoma	1
382	ossification of ligamentum flavum	1
383	Macular dystrophy	1
384	Familial amyloid neuropathy	1
385	Familial hypercholesterolemia	1
386	Ectodermal hypoplasia	1
387	Various chromosomal microdeletion (or duplicate) syndromes	1
388	Hepatic sarcoidosis	1
389	Eyes, teeth, and digital dysplasia	1
390	Basal cell nevus syndrome	1
391	Megalocephaly	1
392	Macromaly-trichotomy syndrome	1
393	Pontine cerebellar hypoplasia	1
394	Fulminant hepatitis	1
395	Polyarteritis nodosum	1
396	hemophilia	1
397	Primary biliary cholangitis	1
398	Primary eruption insufficiency	1
399	Cancer of unknown primary	1
400	Isolated lens luxation	1

#	Disease name	Number of respondents [people]
401	Good acid globule granulogranulotrus multiplea vasculitis	1
402	Eosinophilic sinusitis	1
403	Neutropenia	1
404	Anti-MuSK antibody positive myasthenia gravis	1
405	Antiphospholipid antibody syndrome	1
406	Antineutrophil cytoplasmic antibody-associated vasculitis	1
407	Thyroid ophthalmopathy	1
408	Hyperinsulin-hyperammonemia	1
409	Hypercitrullinemia	1
410	Osteomalacia	1
411	hypoplastic left heart syndrome	1
412	Reticulodysplasia	1
413	trifunctional protein deficiency	1
414	Limb girdle muscular dystrophy	1
415	Lipoatrophy	1
416	Neuromyelitis optica	1
417	Autoinflammatory disease-associated enteritis	1
418	Juvenile Alzheimer's disease	1
419	Juvenile Parkinson's disease	1
420	Juvenile-onset bilateral sensorineural hearing loss	1
421	Periodic fever	1
422	Severe congenital neutropenia	1
423	Severe drug eruption	1
424	Childhood hereditary disorders	1
425	Alternating hemiplegia of childhood (ATP1A3 abnormality)	1
426	Autosomal episomal (superior) polymorphic anchocytic kidneys	1
427	Autosomal latent (inferior) polycytic nepholis	1
428	Autosomal latent trichosis/attrichopathy	1
429	Autosomal latent polycystic kidney disease	1
430	Autosomal predominantly interductal renal disease	1
431	Cardiac sarcoidosis	1
432	Cardiac Fabry disease	1
433	Cardiomyopathy, bradyarrhythmia	1
434	Neonatal diabetes mellitus	1
435	Neuroblastoma	1
436	Intranuclear-inclusion body disease	1
437	Neuroendocrine tumor	1
438	Neurodevelopmental disorders	1
439	Progressive supranuclear palsy	1
440	Progressive myoclonic epilepsy	1
441	fibrodysplasia ossificans progressiva	1
442	Progressive leukoencephalopathy	1
443	Bullous pemphigoid	1
444	Meningeal amyloidosis	1
445	Segawa disease	1
446	Growth Disorder Disease	1
447	Congenital long QT syndrome	1
448	Congenital QT shortening syndrome	1
449	Congenital thrombomodulin disorder	1
450	Congenital hepatic fibrosis	1
451	Congenital tracheal stenosis	1
452	Congenital bone marrow failure syndrome	1
453	Congenital heart disease	1
454	Congenital epidermal vesicular disease	1
455	Congenital corticosteroid enzyme deficiency	1
456	Congenital edentulous	1
457	Congenital immunodyspathy	1
458	Congenital immunodeficiency-associated enteritis	1
459	Frontotemporal degeneration	1
460	Early repolarization syndrome	1
461	Total excretion empty remnants	1
462	Multisystem proteinosis	1
463	idiopathic multicentric Castleman's disease	1
464	Multicentric hand root bone foot root osteolysis	1
465	multiple abnormality	1
466	multiple lentigines syndrome	1
467	Multiple endocrine neoplasia type 1	1
468	Multiple endocrine neoplasia type 2	1
469	Basal ganglia degeneration	1
470	Cerebral leukodegeneration	1
471	single ventricle	1
472	Central diabetes insipidus	1
473	Nakajo-Nishimura syndrome	1
474	Ulcerative colitis	1
475	Hyposodicemia and osteosodicoidosis	1
476	Hyaprotic ectodermal hypoplasia	1
477	Pemphigus	1
478	chondrodysplasia punctata	1
479	Idiosyncratic/relegacious pulmonary arterial pulmonary hyperhememia	1
480	idiopathic basal ganglia calcification	1
481	Idiopathic small bowel ulcer	1
482	Idiopathic cardiomyopathy	1
483	heterotaxy syndrome	1
484	Refractory brain formation disorder	1
485	Infantile epileptic spasm syndrome	1
486	Infantile liver failure syndrome type 1	1
487	urea cycle disorder	1
488	Disorders of brain formation	1
489	Cerebral small vessel disease	1
490	cerebrotendinous xanthomatosis	1
491	Pustular psoriasis	1
492	Seeded epidermic actinic pokeratosis	1
493	Pulmonary Langerhans histiocytosis	1
494	Panhypopituitarism	1
495	Dermatosis leukoplasma	1
496	Hypertrophic duritis	1
497	pachydermoperiostosis	1
498	Atypical hemolytic uremic syndrome	1
499	Microchromosomal structural abnormalities	1
500	Arrhythmogenic right ventricular cardiomyopathy	1

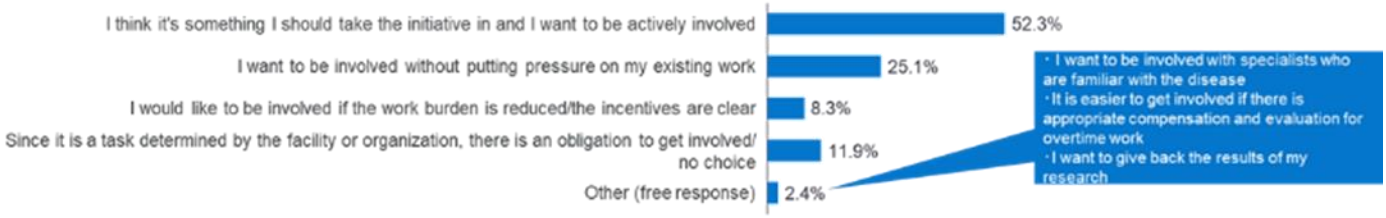
#	Disease name	Number of respondents [people]
501	Complicated congenital heart disease	1
502	hemimegalencephaly	1
503	Chronic thromboembolic pulmonary hypertension	1
504	asplenia syndrome	1
505	Immune thrombocytopenia	1
506	immunodeficiency disease	1
507	Ataxia telangiectasia	1
508	Hair-hepatic-bowel syndrome	1
509	Spastic paraplegia	1
510	Spastic paraplegia	1

Figure 3-1: Percentage of people with experience of collaboration with other professionals and stakeholders



■Survey: Web survey
■Question: Regarding your activities related to rare diseases in the past year, have you collaborated with others? Please answer with an integer between 0 and 10 so that the total for the people you collaborated with is 100%.
■ Subjects: 327 specialists, non-specialists, clinical researchers (basic and applied), clinical researchers (development), and other HCPs (genetic counselors and nurses)

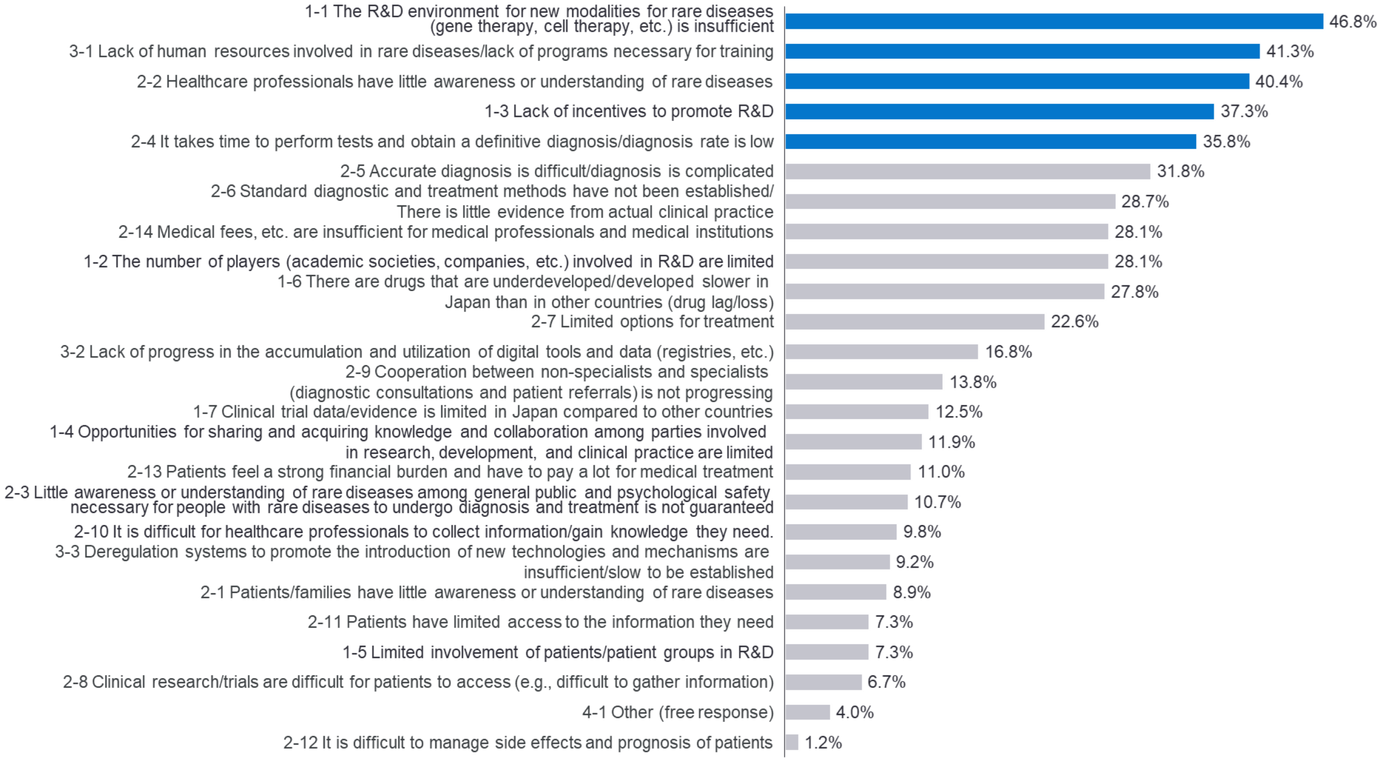
Figure 3-2: Attitude and motivation towards activities related to rare diseases – Top selection result



■Survey: Web survey
■Question: Please choose the top three that apply to you regarding your attitude and motivation for participating in activities related to rare diseases (ranking format)
■Subjects: 327 specialists, non-specialists, clinical researchers (basic and applied), clinical researchers (development) and other HCPs (genetic counselors and nurses)

4.1.1 Overall landscape of challenges in rare diseases in Japan

Figure 4.1.1-1: Overview of the challenges facing rare diseases in Japan

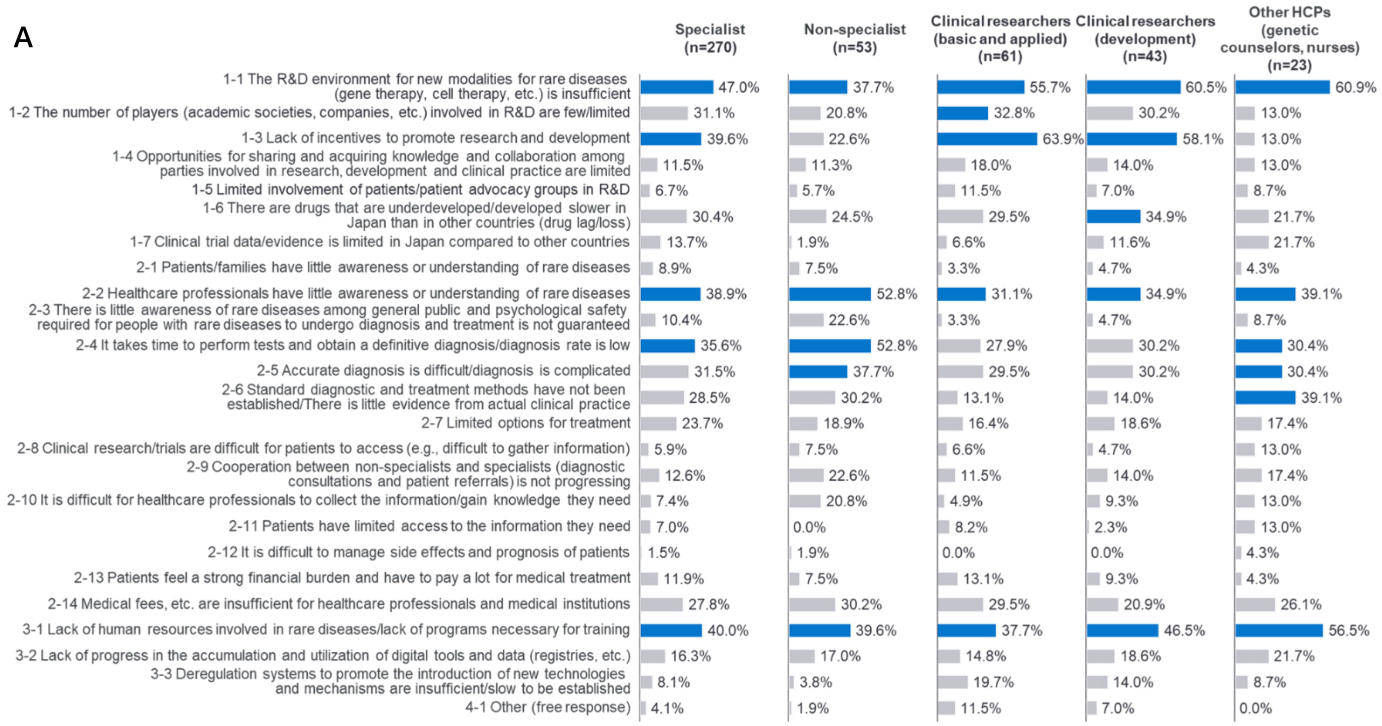


■Survey: Web survey

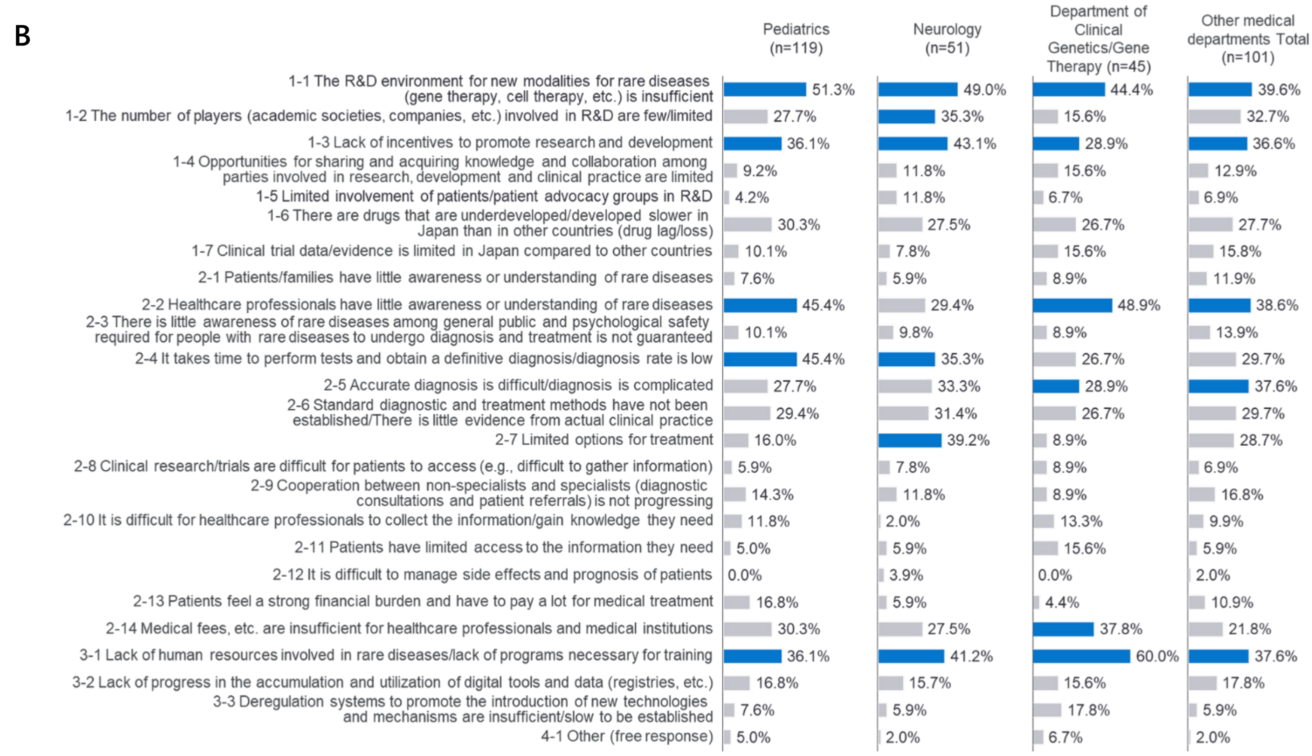
■Question: What are the most pressing challenges regarding rare diseases in Japan? (Select 5)

■Subjects: 327 specialists, non-specialists, clinical researchers (basic and applied), clinical researchers (development) and other HCPs (genetic counselors and nurses)

Figure 4.1.1-2: Overall landscape of the challenges facing rare diseases
・A by occupation ・B by medical department ・C by disease research area ・D by region ・E by professional staff availability

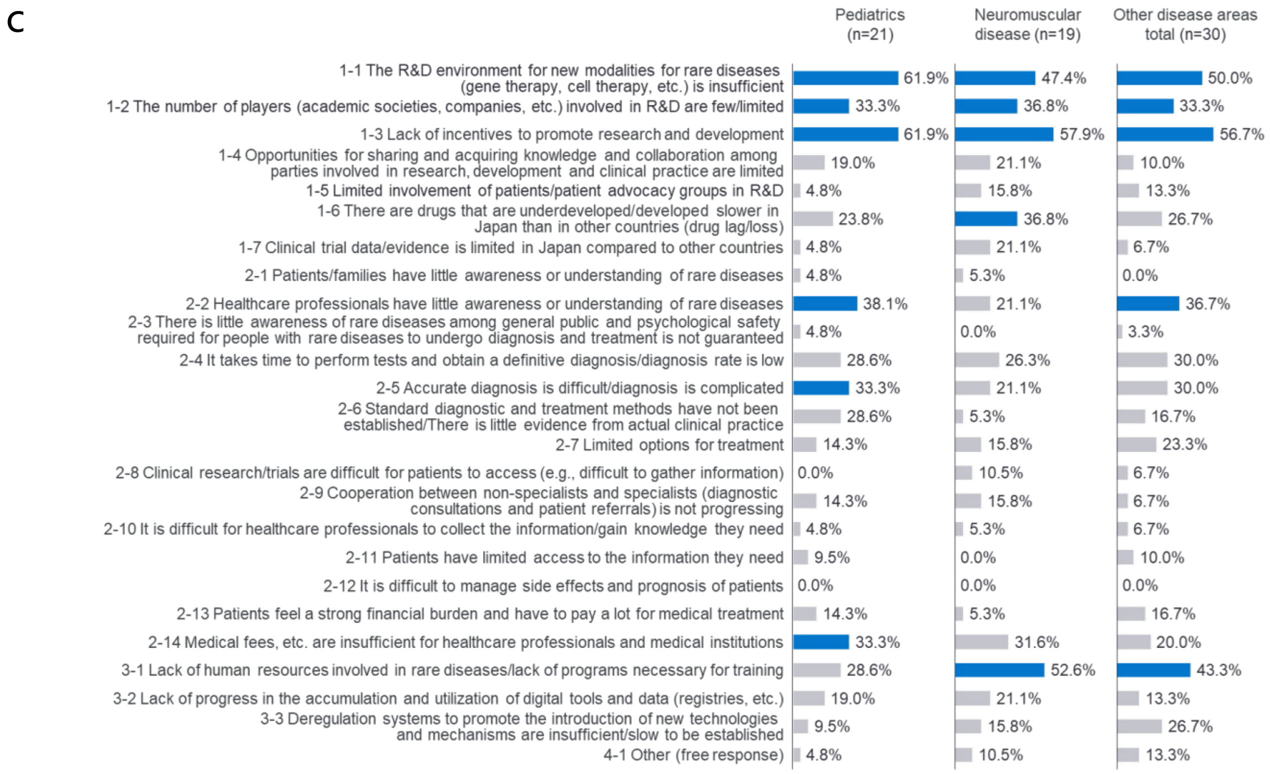


■Survey: Web survey
■Question: Please answer the most important issue you feel is related to rare diseases in Japan (choose 5, multiple choice)
■Subjects: 327 specialists, non-specialists, clinical researchers (basic and applied), clinical researchers (development) and other HCPs (genetic counselors and nurses)



*Medical departments with 15 or more respondents selected

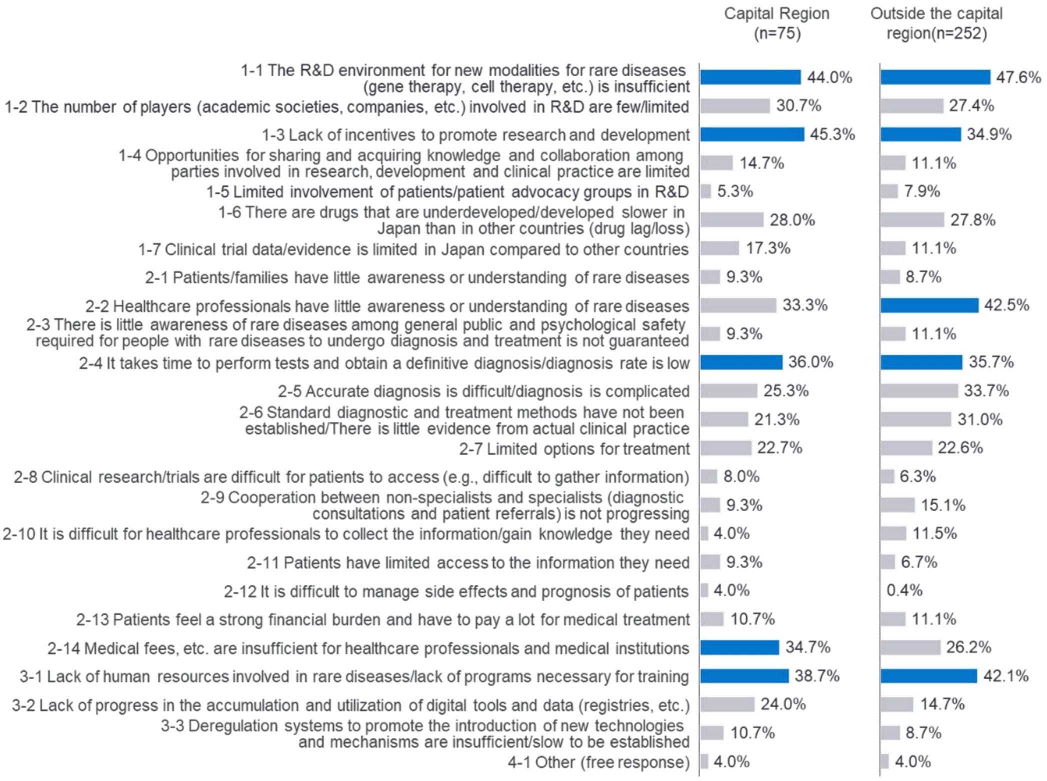
■Survey: Web survey
■Question: Please answer the question about the most pressing challenges surrounding rare diseases in Japan (choose 5, multiple choice)
■Subjects: 316 specialists, non-specialists, and other HCPs (genetic counselors and nurses)



*Research areas with 15 or more respondents were selected

■Survey: Web survey
■Question: Please answer the question about the most pressing challenges surrounding rare diseases in Japan (choose 5, multiple choice)
■Subjects: 70 clinical researchers (basic and applied), clinical researchers (development)

D

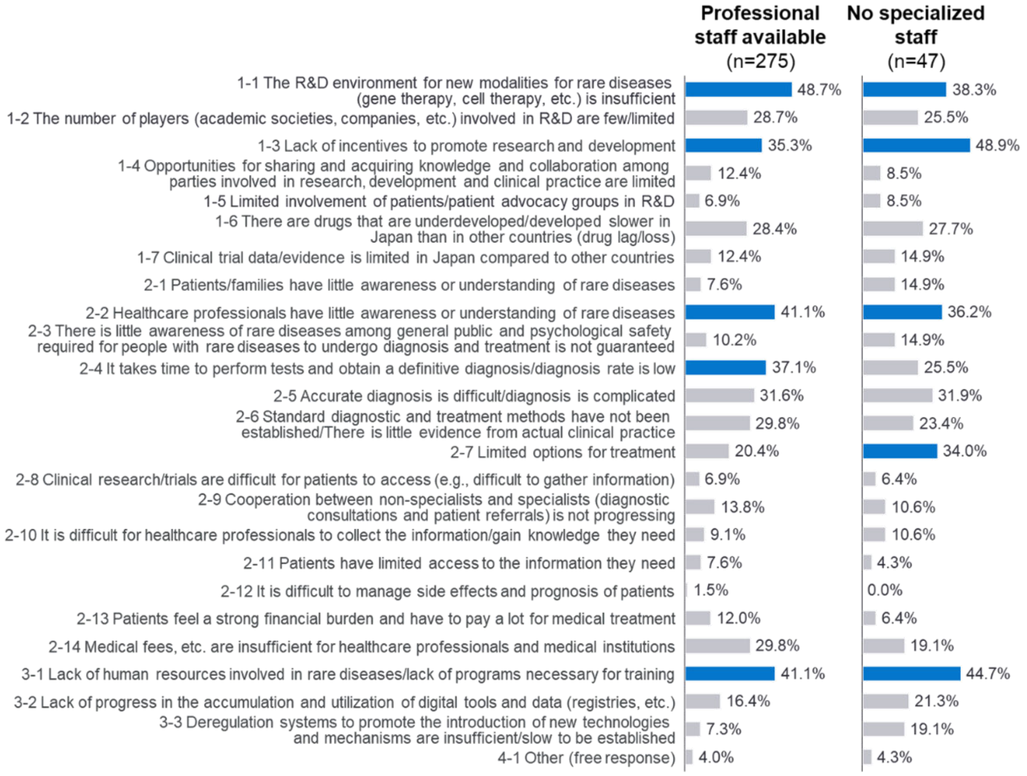


■Survey: Web survey

■Question: Please answer the most important issue you feel is related to rare diseases in Japan (choose 5, multiple choice)

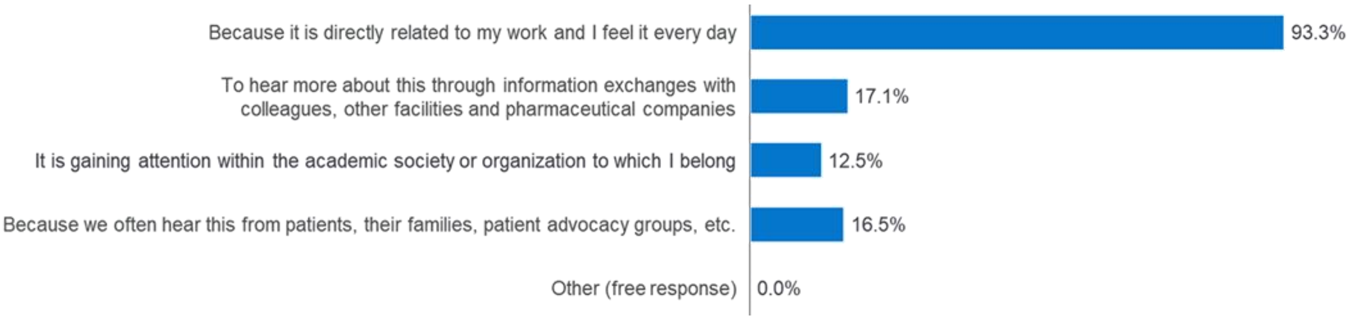
■Subjects: 327 specialists, non-specialists, clinical researchers (basic and applied), clinical researchers (development) and other HCPs (genetic counselors and nurses)

E



■Survey: Web survey
■Question: Please answer the question about the most pressing challenges surrounding rare diseases in Japan (choose 5, multiple choice)
■ Subjects: 327 specialists, non-specialists, clinical researchers (basic and applied), clinical researchers (development) and other HCPs (genetic counselors and nurses)

Figure 4.1.1-3: The big picture of the challenges facing rare diseases

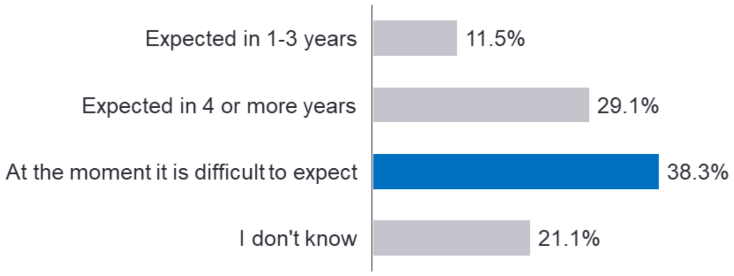


■Survey: Web survey
■Question: Please answer the question about the most pressing challenges surrounding rare diseases in Japan (choose 5, multiple choice)
■ Subjects: 327 specialists, non-specialists, clinical researchers (basic and applied), clinical researchers (development) and other HCPs (genetic counselors and nurses)

Figure 4.1.1-4: Challenges related to rare diseases and expectations of stakeholders (keywords from qualitative interviews)

4.1.2 Challenges in research and development

Figure 4.1.2-1: Expectations for progress in R&D leading to fundamental treatment of rare diseases



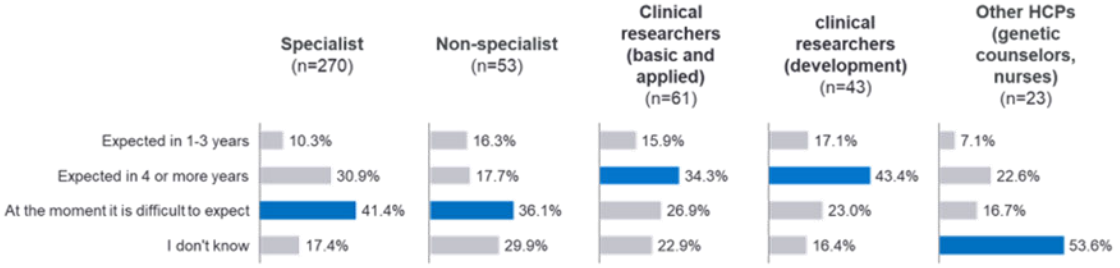
■Survey: Web survey

■Question: Please answer the question about the rare disease you answered in Q13 (Please answer the main rare disease names among your activities related to rare diseases in the past year (up to 5 names allowed)). Do you expect progress in research and development that will lead to a fundamental treatment for the rare disease?

■Subjects: 327 specialists, non-specialists, clinical researchers (basic and applied), clinical researchers (development) and other HCPs (genetic counselors and nurses)

Figure 4.1.2-2: Expectations for progress in R&D leading to fundamental treatment of rare diseases - A by occupation / B by medical department

A

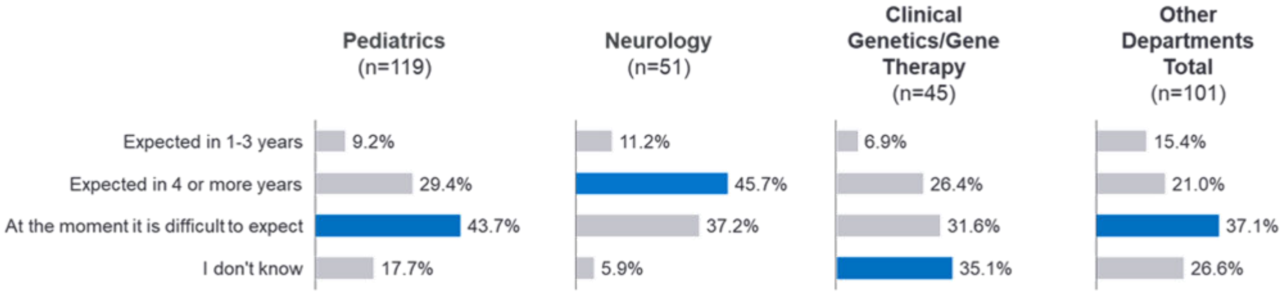


■Survey: Web survey

■Question: Please answer the question about the rare disease you answered in Q13. Do you expect progress in R&D leading to a fundamental treatment for the rare disease? (Q13: Please answer the name of the main rare disease among your activities related to rare diseases in the past year (up to 5 answers possible)

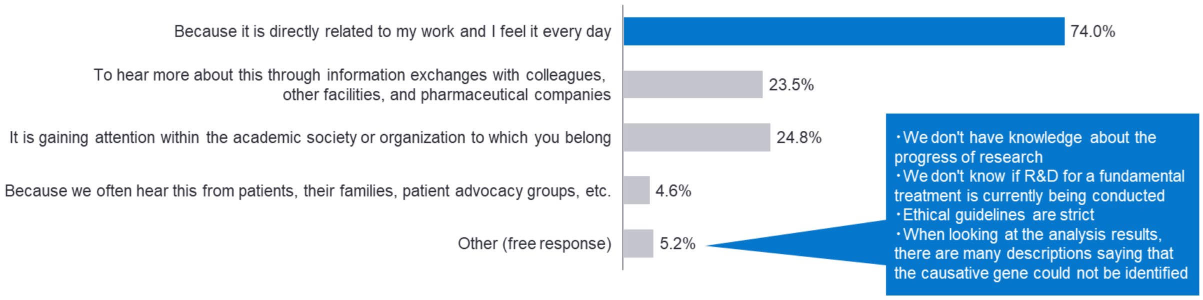
■Subjects: 327 specialists, non-specialists, clinical researchers (basic and applied), clinical researchers (development) and other HCPs (genetic counselors and nurses)

B



■Survey: Web survey
■Question: Please answer the question about the rare disease you answered in Q13. Do you expect progress in R&D leading to a fundamental treatment for the rare disease? (Q13: Please answer the name of the main rare disease among your activities related to rare diseases in the past year (up to 5 answers possible)
■Subjects: 316 specialists, non-specialists, and other HCPs (genetic counselors and nurses)

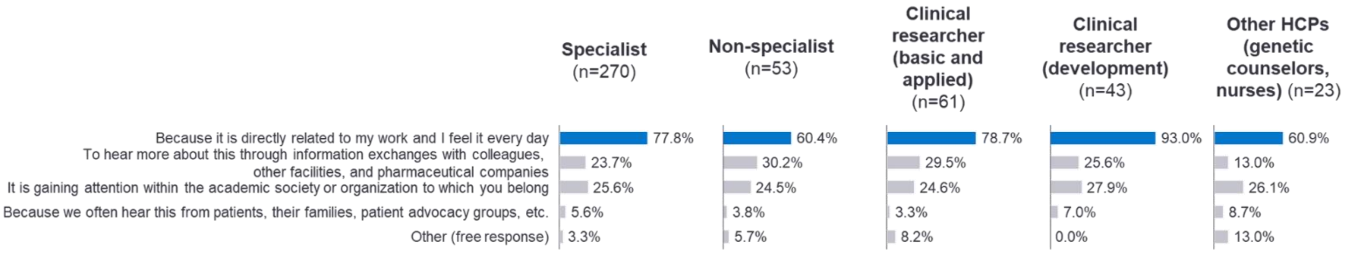
Figure 4.1.2-3: Reasons for expecting progress in R&D leading to fundamental treatments of rare diseases



・We don't have knowledge about the progress of research
・We don't know if R&D for a fundamental treatment is currently being conducted
・Ethical guidelines are strict
・When looking at the analysis results, there are many descriptions saying that the causative gene could not be identified

■Survey: Web survey
■Question: Please answer the reason (multiple choices possible)
■Subjects: 327 specialists, non-specialists, clinical researchers (basic and applied), clinical researchers (development) and other HCPs (genetic counselors and nurses)

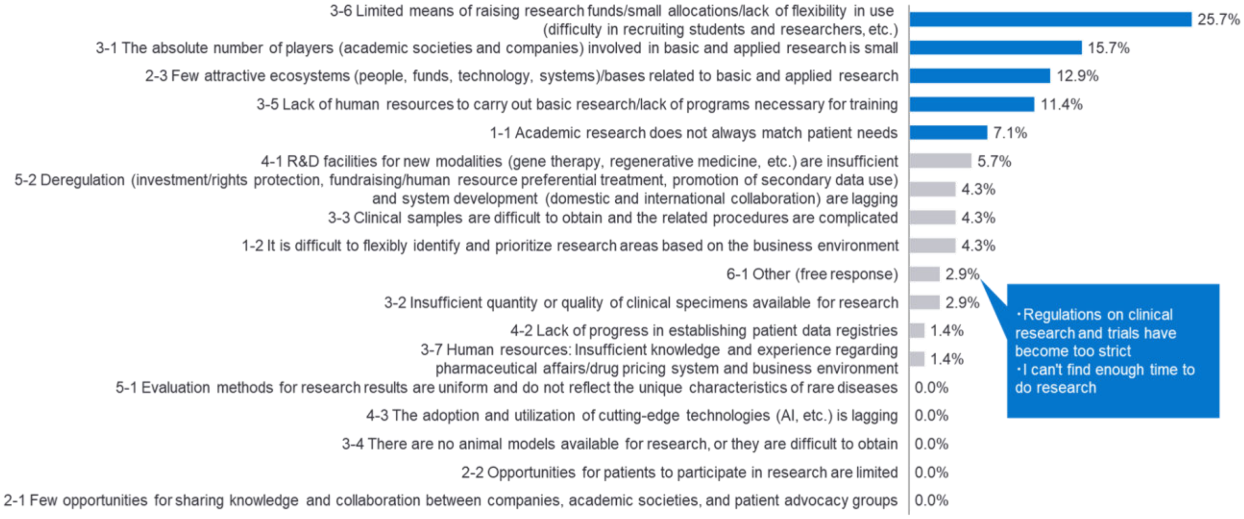
Figure 4.1.2-4: Reasons for expectation of progress in R&D leading to fundamental treatment of rare diseases – by occupation



■Survey: Web survey
■Question: Please answer the reason (multiple choices possible)
■Subjects: 327 specialists, non-specialists, clinical researchers (basic and applied), clinical researchers (development) and other HCPs (genetic counselors and nurses)

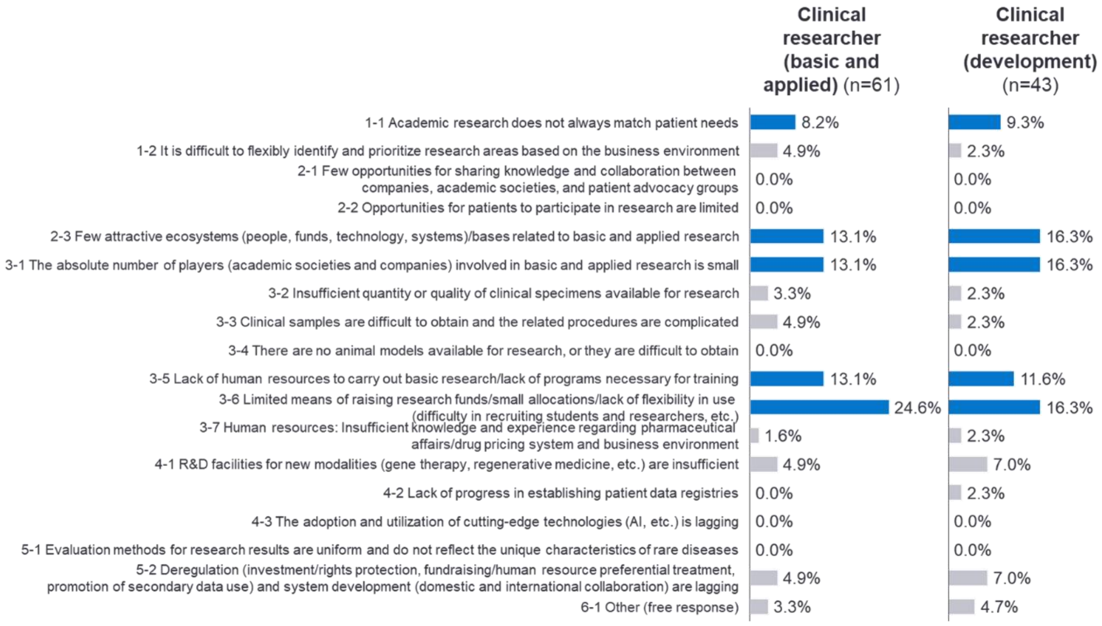
Figure 4.1.2-5: Challenges in basic and applied research – Top selection results：
A all Segments ・B by occupation ・C by disease research area ・D by specialty

A

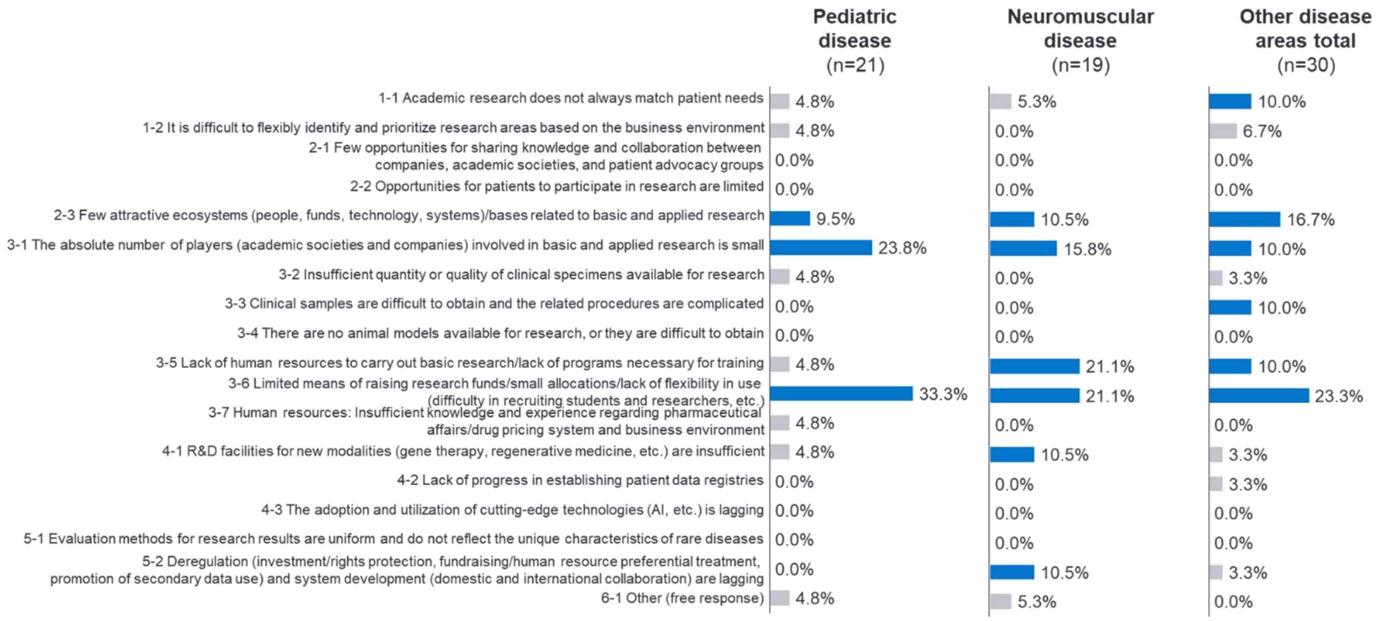


・Regulations on clinical research and trials have become too strict
・I can't find enough time to do research

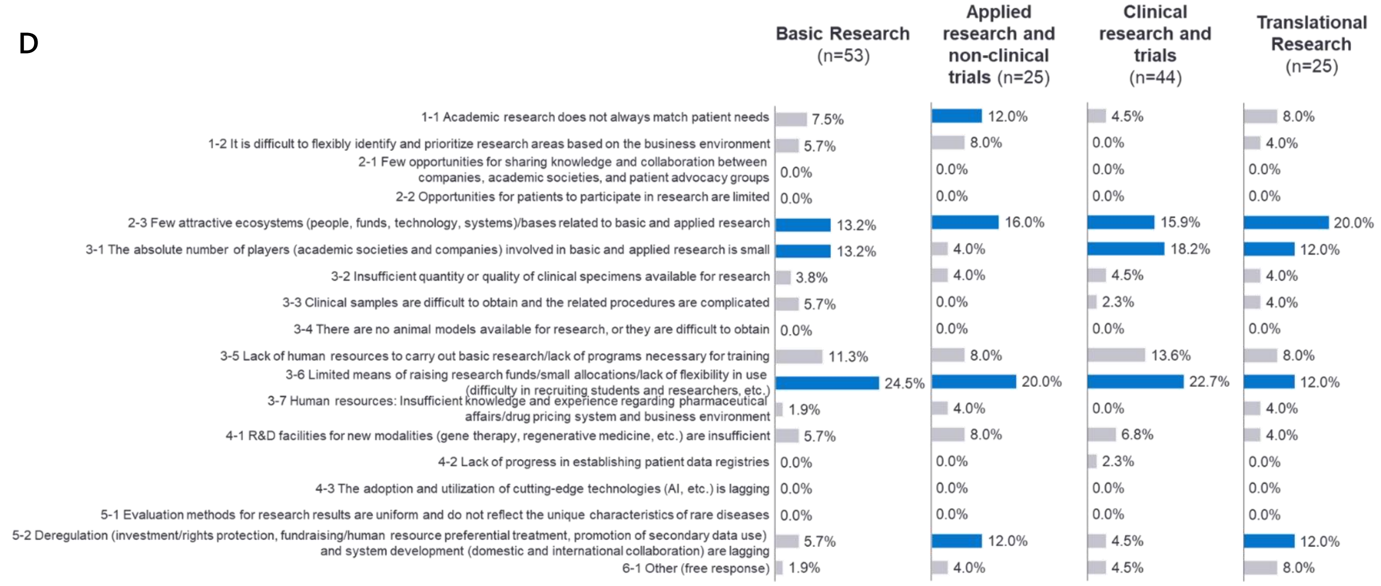
B



C

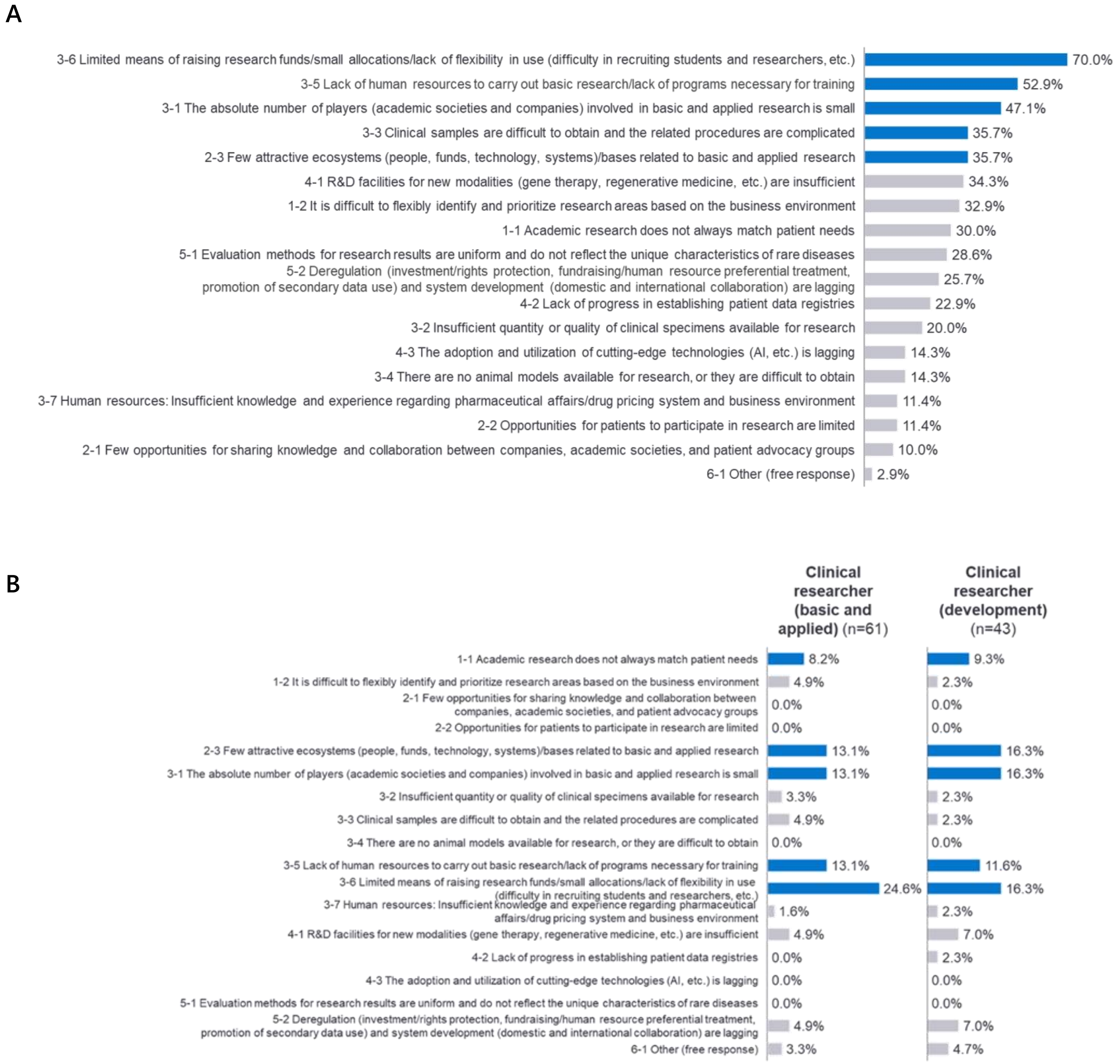


D

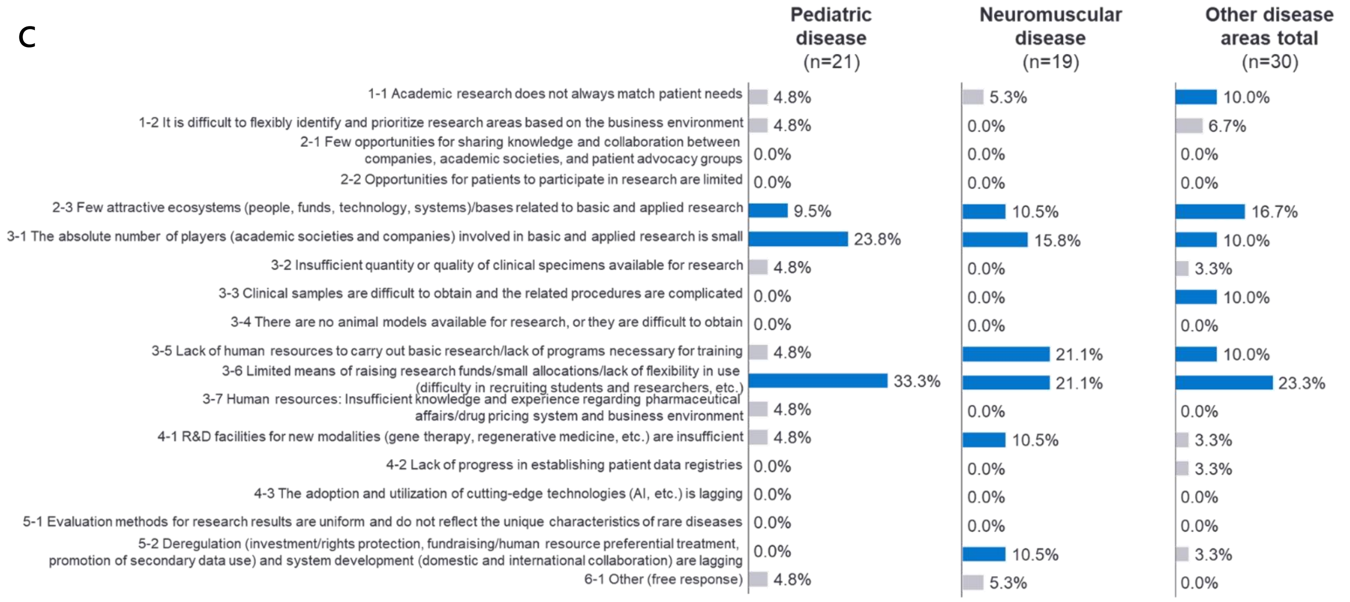


■Survey: Web survey
■Question: Please select the top 5 challenges that you feel are most important in basic and applied research (ranking format)
■Subjects: 70 clinical researchers (basic and applied) and clinical researchers (development)

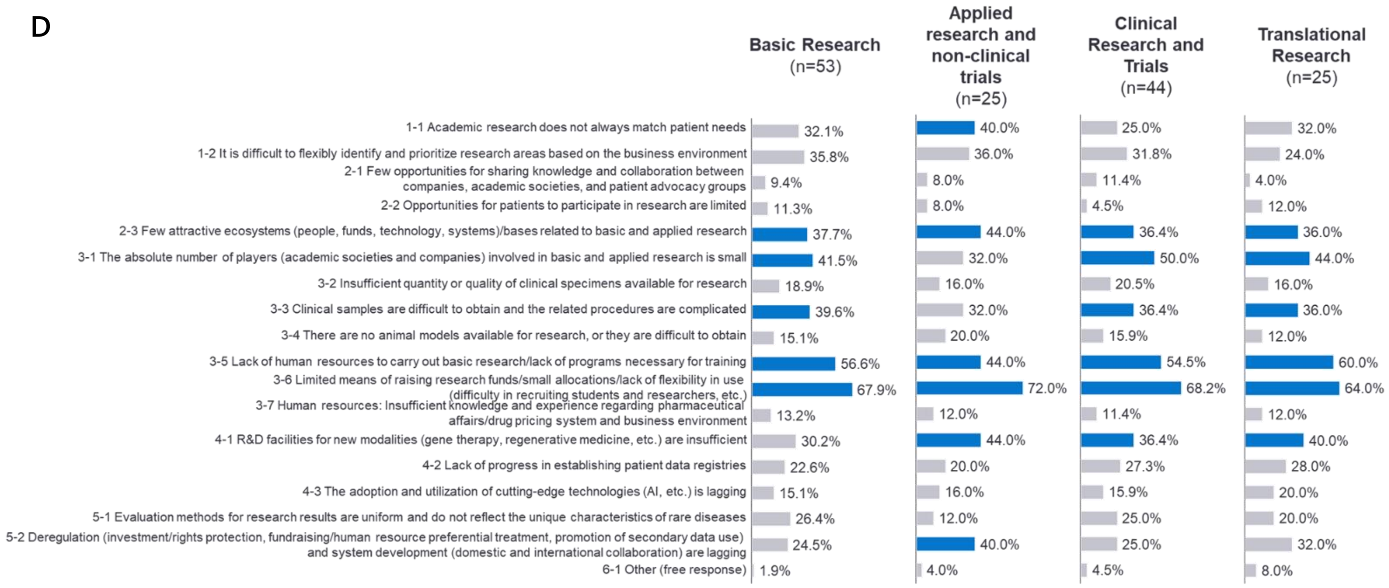
Figure 4.1.2-6: Challenges in basic and applied research – Top 5 Selection Results：
A all segments ・B by occupation ・C by disease research area ・D by specialty



C

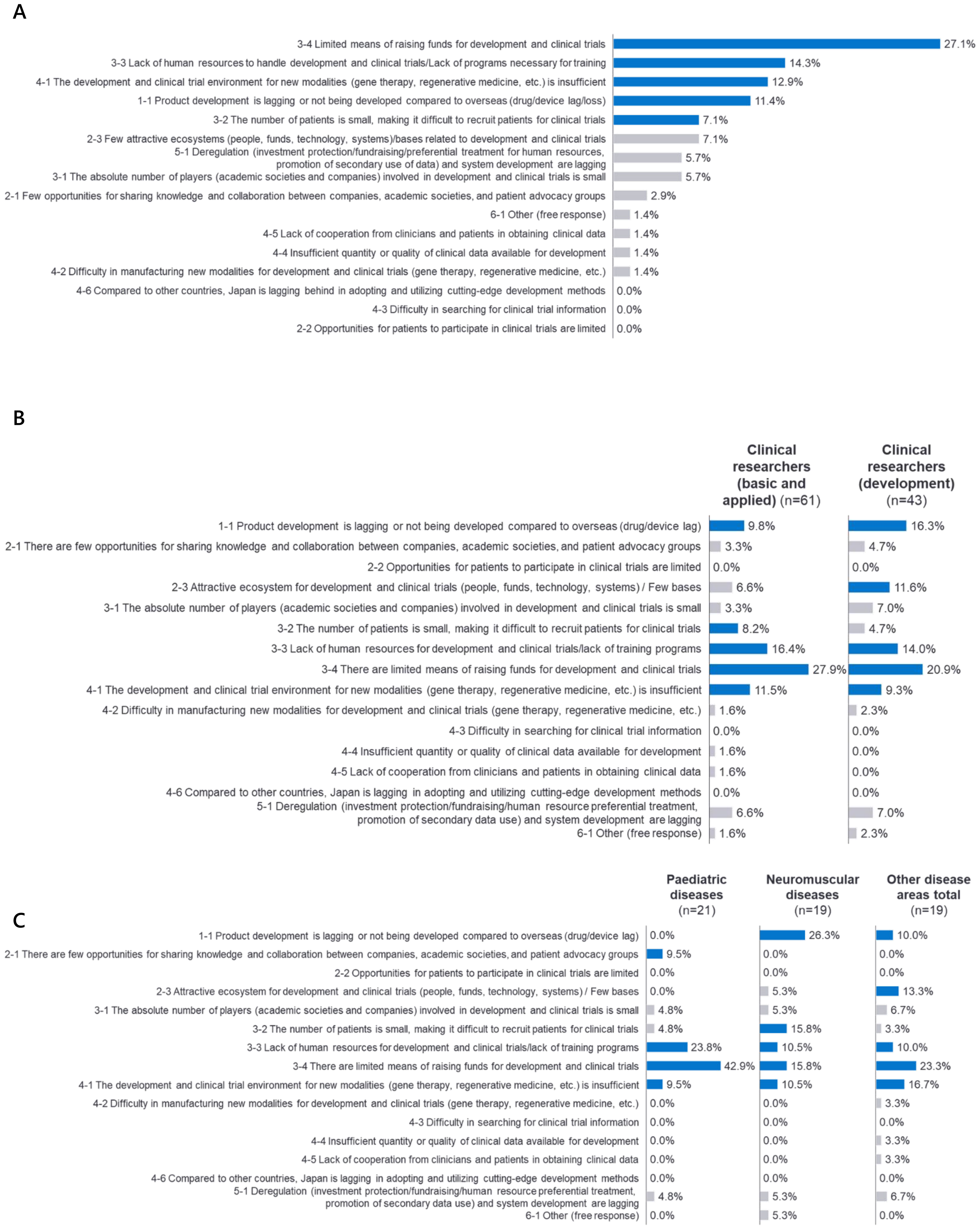


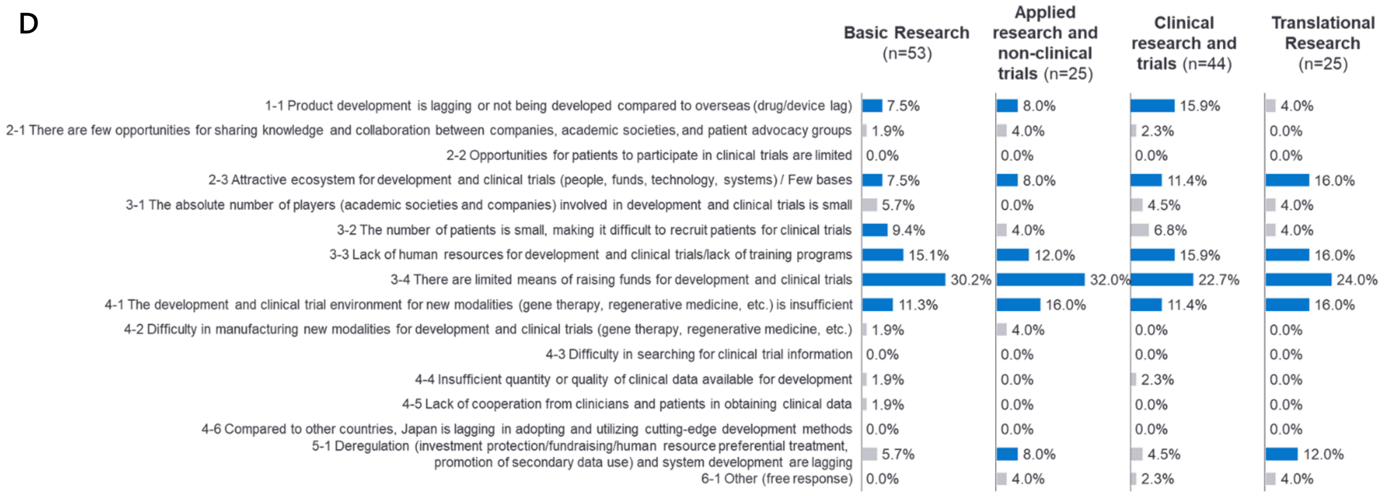
D



■Survey: Web survey
■Question: Please answer by selecting the top 5 challenges that you feel are most important in basic and applied research (ranking format)
■Subjects: 70 clinical researchers (basic and applied) and clinical researchers (development)

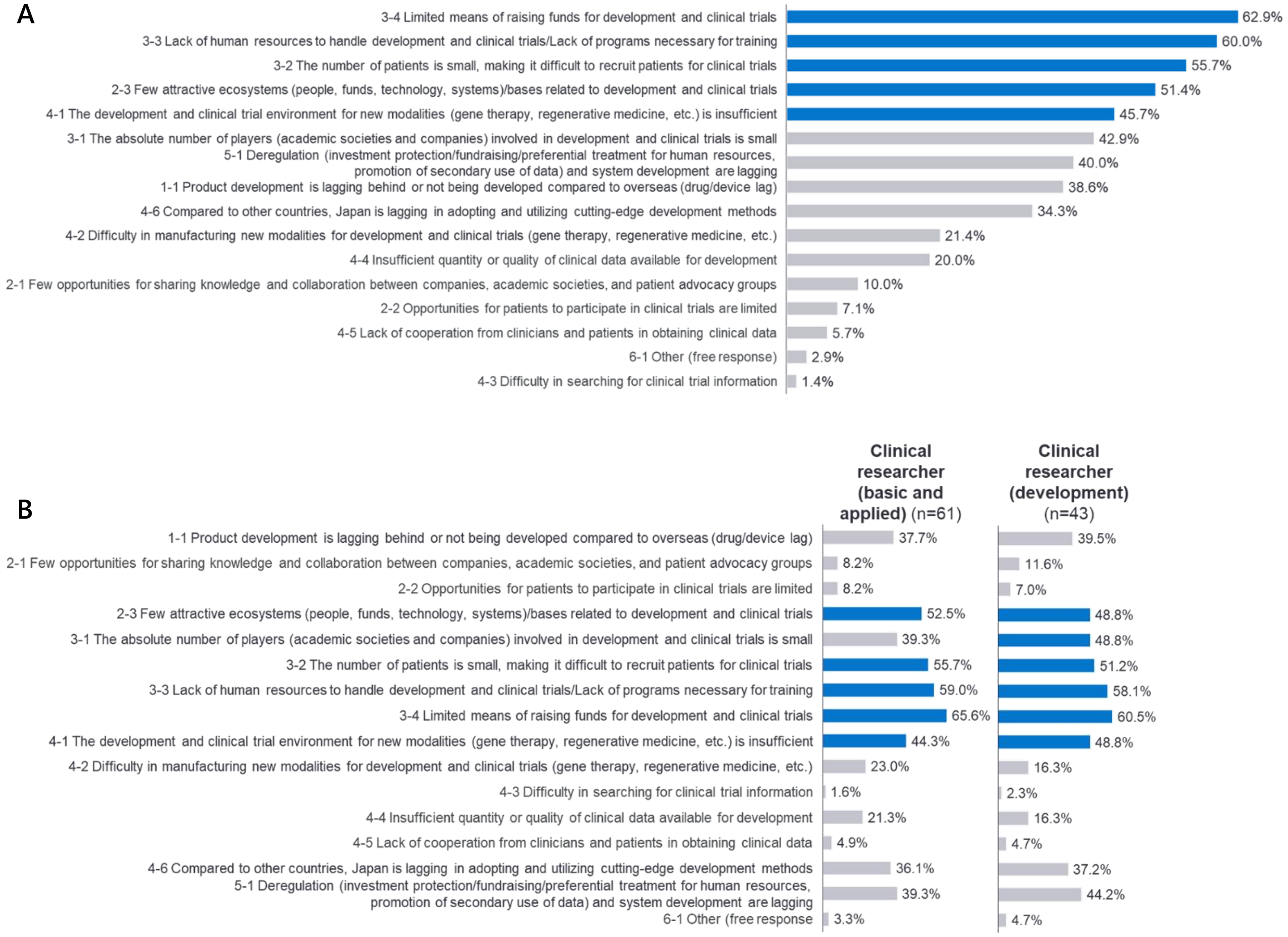
Figure 4.1.2-7: Challenges in development and clinical trials – Top selection result：
A all segments ・B by occupation ・C by disease research area ・D by specialty



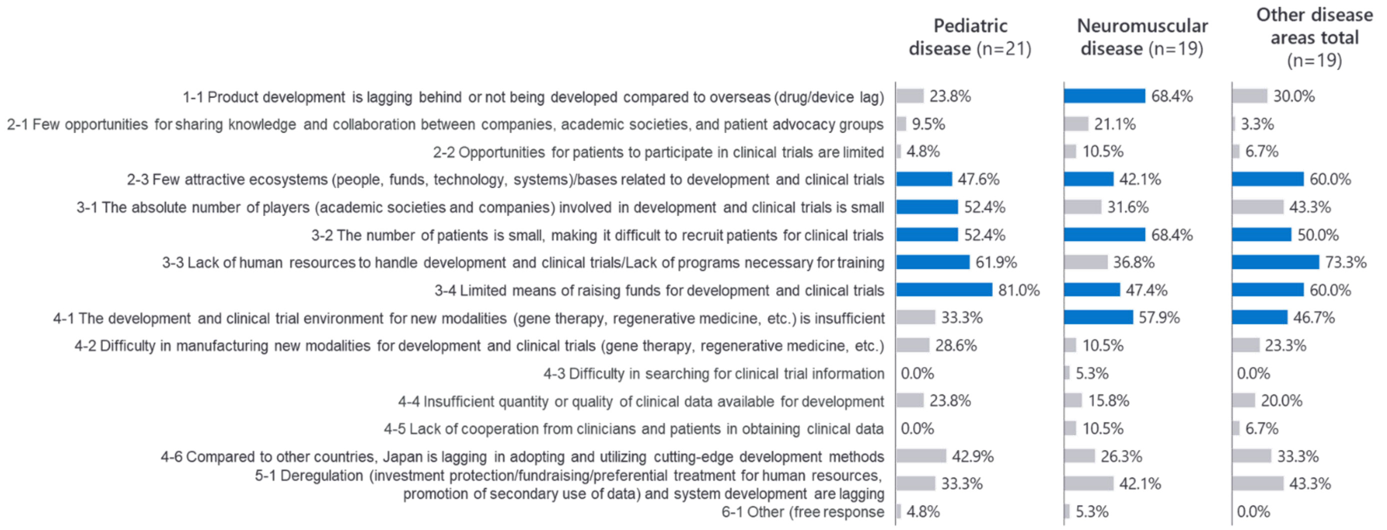


- ■ Survey: Web survey
- ■ Question: Please select the top 5 challenges you feel are most challenging regarding development and clinical trials (ranked)
- ■ 70 clinical researchers (basic and applied) and clinical researchers (development)

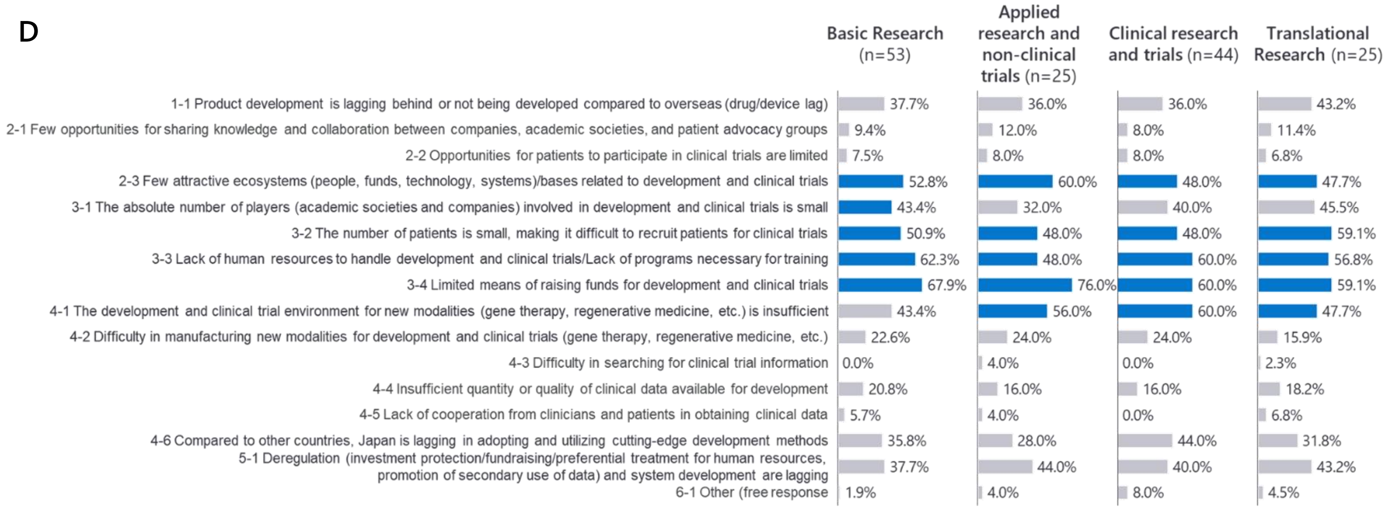
Figure 4.1.2-8: Challenges in development and clinical trials – Top 5 Selection Results：
A all segments ・B by occupation ・C by disease research area ・D by specialty



C



D



- Survey: Web survey
- Question: Please select the top 5 challenges you feel are most challenging regarding development and clinical trials (ranked)
- 70 clinical researchers (basic and applied) and clinical researchers (development)

“ *In Japan, when it comes to R&D and the social implementation of new technologies/systems, social consensus tends to take precedence, and there is a tendency for insufficient discussion on 'what should be prioritized for the patient in front of us.' Japanese society is one in which risk and challenge cannot be tolerated and there is strong pressure to conform, but if patients and their families can tolerate the risks, there should be a system in place that allows them to receive treatment and other assistance on an exceptional basis.* ***This culture of placing too much emphasis on the ethical values of society in general over the will of the patient is an obstacle to R&D and clinical trials in Japan****.*
(Clinical researcher (basic and applied) / Other hereditary disease)

“ ***The construction of patient data registries has not progressed*** *due to high hurdles in academic cliques and research ethics. For example, registries for disorders of sex development already exist in about 20 countries in the EU, but Japan does not yet have one.*
(Clinical researcher (basic and applied) / Pediatrics)

“ *There is a **shortage of human resources and training programs** to carry out basic and applied research and working at a university inevitably means a large amount of administrative and clerical work unrelated to rare diseases, so the limited number of people who are hubs in the field of rare diseases need to have the time and financial flexibility to take on this challenge.*
(Clinical researcher (basic and applied) / Pediatrics)

“ ***Obtaining clinical samples is difficult and the related procedures are complicated**. When obtaining clinical samples from other facilities, they must go through the IRB, which has strict document submission requirements and even when discussing with overseas researchers and companies, the strictness of the document submission requirements can become an obstacle and cause negotiations to stall.*
*In addition, there is a lack of information exchange between different industries regarding what seeds (basic research results and technologies that lead to the development of new treatments and medical technologies) are desired, **making it difficult to flexibly identify and prioritize research areas based on the business environment**. There should be more opportunities for exchange with healthcare professionals as the hub.*
(Clinical researcher (development) / Endocrinology and Metabolic Disease)

“ ***The biggest challenge in the world of basic and applied research is always how to obtain research funds and gather colleagues to work with**.*
I feel that the lack of understanding of rare diseases among young people is leading to a shortage of human resources, so I feel that it is necessary to convey the need for development of diagnosis and treatment for rare diseases through lectures and speeches, but as a prerequisite for doing this, I would like the government to actively provide research funding for rare diseases. On the other hand, national research institutes have no university affiliations, so it is difficult to sustain a sustainable supply of young researchers, and even institutes with abundant budgets that are in urban areas have difficulty securing human resources.
(Clinical researcher (basic and applied) / neuromuscular disease)

“ ***The insufficient environment for new modalities makes it difficult to secure materials, which is an impediment to the development of new modalities.***
Specifically, while there seem to be few facilities in Japan that manufacture cells for cell therapy, in Europe and the United States, GMP manufacturing facilities/CPCs are attached to the medical schools of leading universities.
(Clinical researcher (basic and applied) / neuromuscular disease)

“ *As Japan's domestic economy and population shrinks and the number of domestic bases for foreign companies decreases, it is becoming unclear to foreign companies who can concretely discuss domestic development, and this situation is **accelerating drug loss**.*
(Clinical researcher (basic and applied) / neuromuscular disease)

“ *Compared to cancer, the national budget and personnel for research, development, and clinical practice are small, and there is **a lack of human resources, educators, and programs** in particular, which means there is no system or foundation for systematically allocating and training human resources, resulting in a lack of speed compared to Europe and the United States. In addition to educating specialists, we believe that we need to fundamentally reconsider not only the way in which diversity and genetics education is provided in primary education.*
(Clinical researcher (development) / All other hereditary disease)

“ ***Information on the progress of development should be systematically organized and made more accessible to patients and healthcare professionals.*** *This could encourage patients to seek medical treatment and motivate them to go to the hospital, which could ultimately lead to an improvement in the diagnosis rate.*

(Other HCPs (Genetic counselors and nurses) / Department of Clinical Genetics and Gene Therapy)

“ *I am involved in a clinical trial for achondroplasia, but it is* ***extremely difficult to recruit subjects who meet the conditions****. One of the reasons is that the subjects are not fully informed. If there was a system where clinical trial information was centrally collected and it was possible to narrow down clinical trial information and subject information that meets the conditions, it would be convenient for both healthcare professionals and subjects. Also, since it is often difficult to recruit subjects even if a drug that has already been approved in the US or EU is approved in Japan later, it is desirable to accelerate participation in international joint clinical trials.*

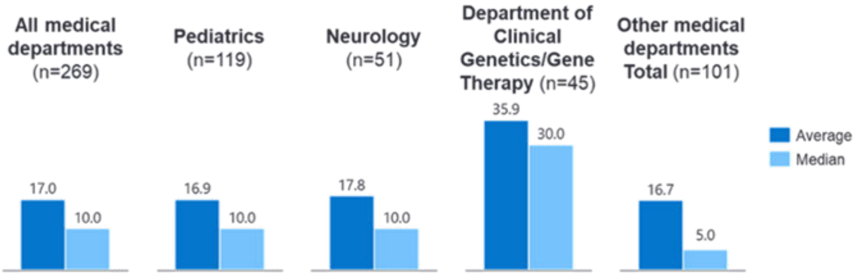
(Specialist / Pediatrics)

“ *Because rare diseases affect only a small number of patients,* ***there is little economic incentive for pharmaceutical companies, and the low motivation of industry is a clear barrier****.*

(Clinical researchers (basic and applied) / Endocrinology and Metabolic Disease)

4.1.4 Challenges in diagnosis

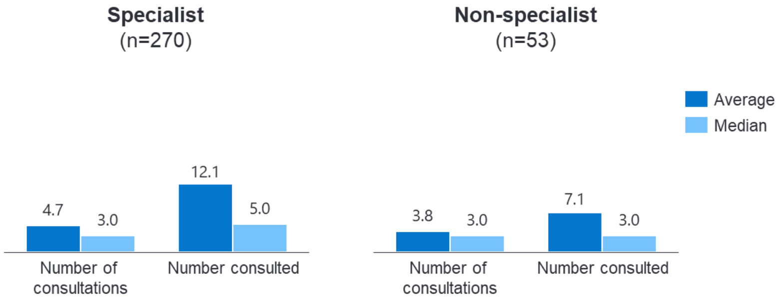
Figure 4.1.4-1: Number of patients with suspected rare diseases referred to specialists/year



*Analysis results excluding responses of 200 or more as outliers

■Survey: Web survey
■Question: Q6 If you answered ‘1. Responsible for making diagnostic and treatment decisions as a clinical doctor (specialist/quasi-specialist)’ to the question about your occupation, please tell us how many patients with suspected rare diseases you are referred to each year (numeric answer)
■Subjects: 269 specialists

Figure 4.1.4-2: Number of consultations related to diagnosis/year



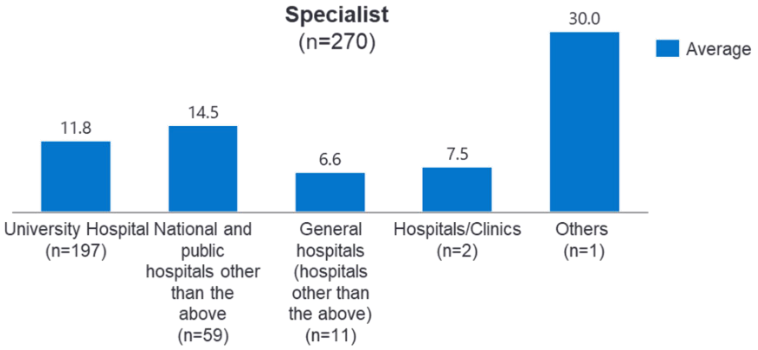
*Analysis results excluding responses of 100 or more as outliers

■Survey: Web survey
■Question: Regarding consultations related to rare disease diagnoses, how many times per year do you consult with others? (Answer with a number)
■Subjects: 302 specialists and non-specialists

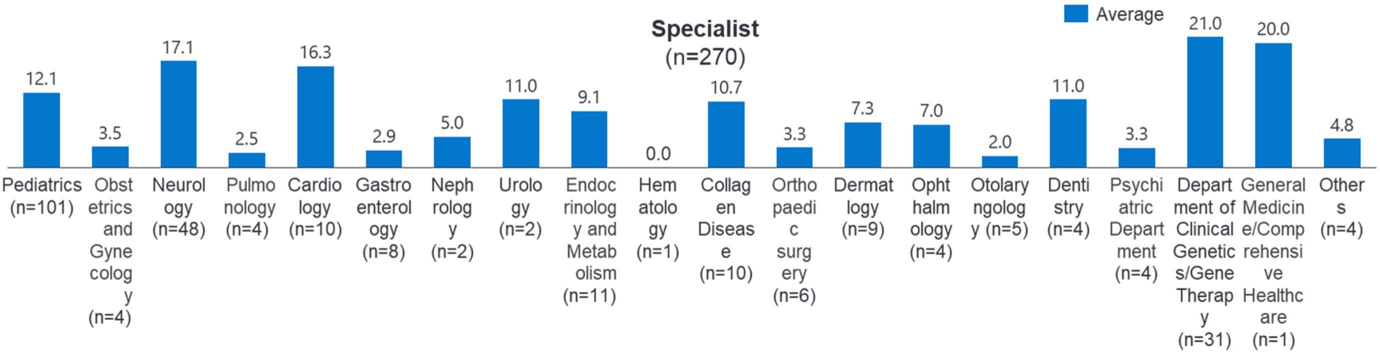
Figure 4.1.4-3: Number of rare disease diagnoses consulted by specialists/year

– A by affiliated institution ・B by medical department

A



B



■Survey: Web survey

■Question: Regarding consultations related to the diagnosis of rare diseases, please answer how many times per year you receive consultations (numeric answer)

■Subjects: 270 specialists

Figure 4.1.4-4: Duration and Number of Facilities Involved in Reaching a Definitive Diagnosis

- A: Overall, B: by medical department

A

	Less than 6 months	Less than 1 year	More than 1 year	More than 3 years	Total
1 facility	19.6%	4.5%	5.5%	1.5%	31.2%
2 facilities	13.6%	15.6%	11.1%	3.5%	43.7%
3 facilities	0.5%	3.5%	6.5%	4.0%	14.6%
4 facilities	2.0%	1.5%	2.0%	5.0%	10.6%
Total	35.7%	25.1%	25.1%	14.1%	100.0%

B

Pediatrics (n=119)

	Less than 6 months	Less than 1 year	More than 1 year	More than 3 years	Total
1 facility	25.6%	2.4%	8.5%	2.4%	39.0%
2 facilities	14.6%	12.2%	12.2%	2.4%	41.5%
3 facilities	0.0%	1.2%	7.3%	4.9%	13.4%
4 facilities and above	1.2%	1.2%	0.0%	3.7%	6.1%
Total	41.5%	17.1%	28.0%	13.4%	100.0%

Neurology (n=51)

	Less than 6 months	Less than 1 year	More than 1 year	More than 3 years	Total
1 facility	2.9%	0.0%	0.0%	0.0%	2.9%
2 facilities	8.8%	23.5%	11.8%	2.9%	47.1%
3 facilities	0.0%	5.9%	14.7%	2.9%	23.5%
4 facilities and above	2.9%	2.9%	11.8%	8.8%	26.5%
Total	14.7%	32.4%	38.2%	14.7%	100.0%

Department of Clinical Genetics/Gene Therapy (n=45)

	Less than 6 months	Less than 1 year	More than 1 year	More than 3 years	Total
1 facility	17.4%	4.3%	8.7%	4.3%	34.8%
2 facilities	13.0%	8.7%	8.7%	4.3%	34.8%
3 facilities	0.0%	8.7%	0.0%	4.3%	13.0%
4 facilities and above	4.3%	4.3%	0.0%	8.7%	17.4%
Total	34.8%	26.1%	17.4%	21.7%	100.0%

Other medical departments Total (n=101)

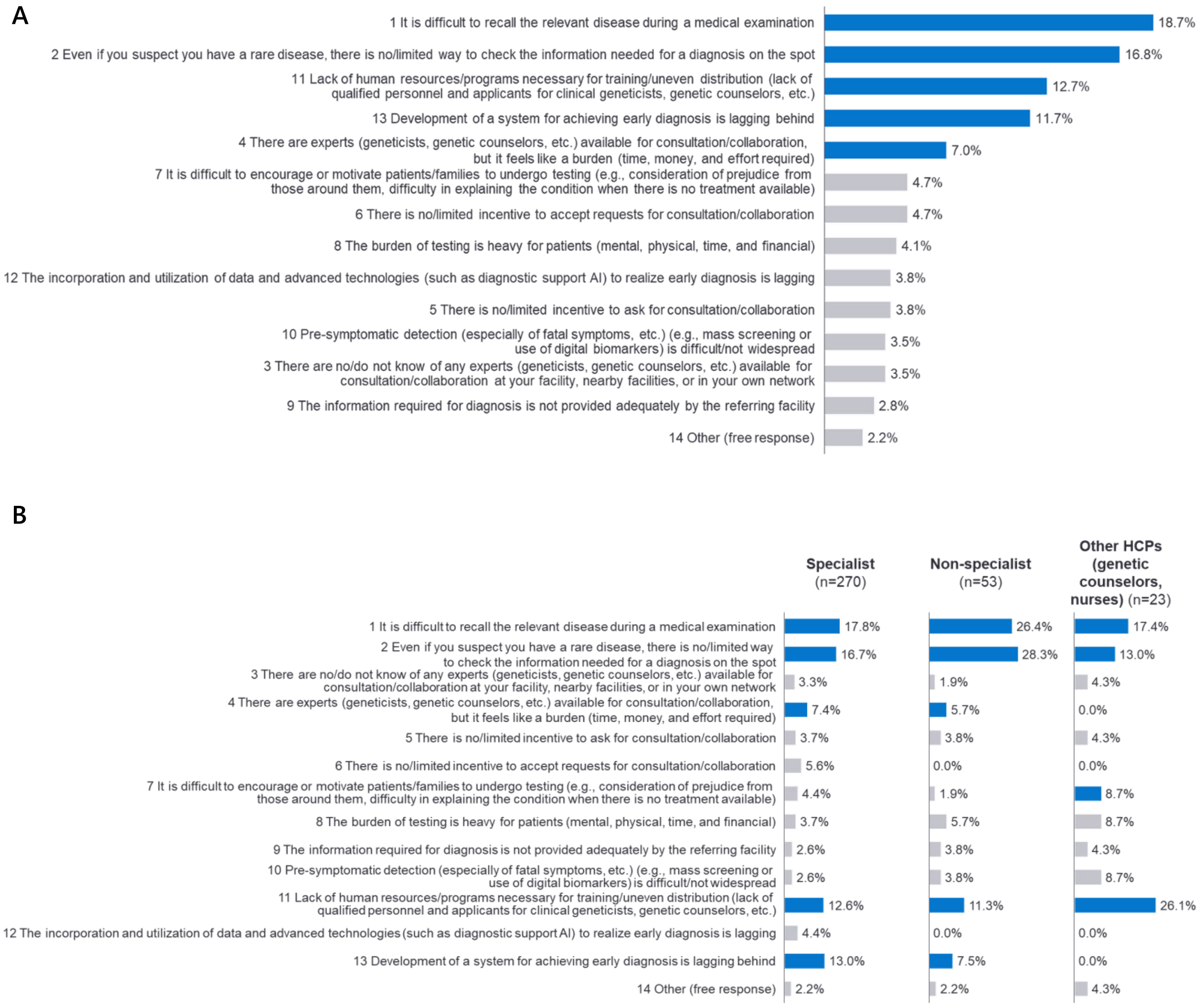
	Less than 6 months	Less than 1 year	More than 1 year	More than 3 years	Total
1 facility	21.7%	10.0%	3.3%	0.0%	35.0%
2 facilities	15.0%	18.3%	10.0%	5.0%	48.3%
3 facilities	1.7%	3.3%	3.3%	3.3%	11.7%
4 facilities and above	1.7%	0.0%	0.0%	3.3%	5.0%
Total	40.0%	31.7%	16.7%	11.7%	100.0%

■Survey: Web survey

■Question: Please tell us how long it took for the most recent rare disease patient to be diagnosed after their first visit, and to which medical institution they were referred after their first visit (select one)

■Subjects: 270 specialists

Figure 4.1.4-5: Problems in diagnosis – Top selection result：
A all segments ・B by occupation ・C by medical department



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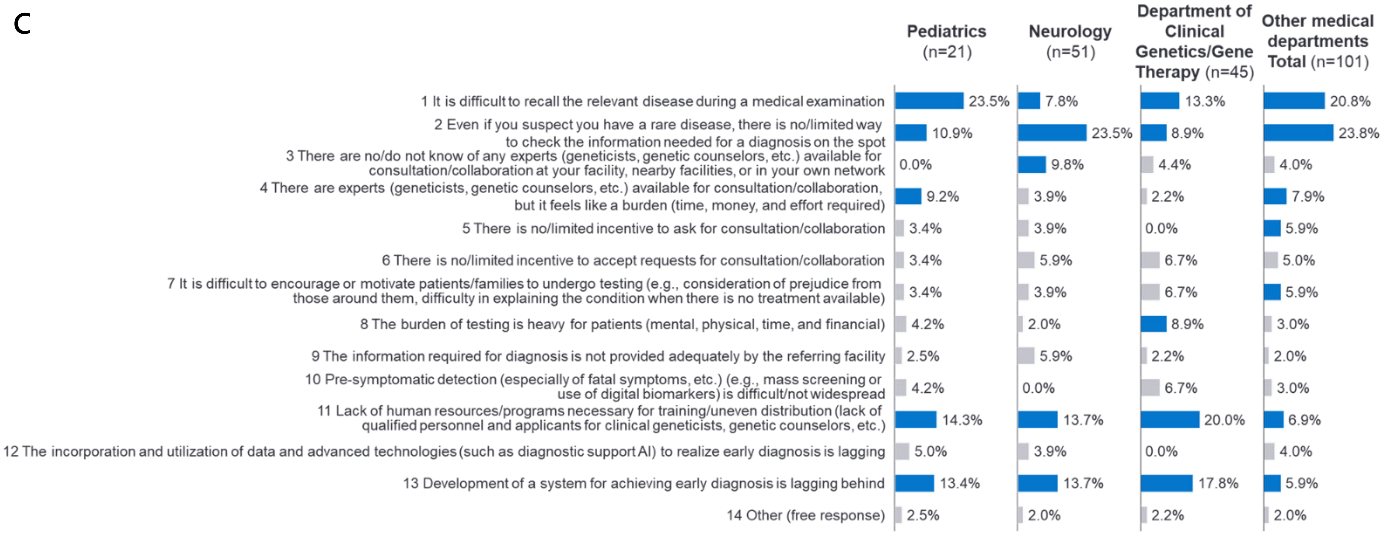


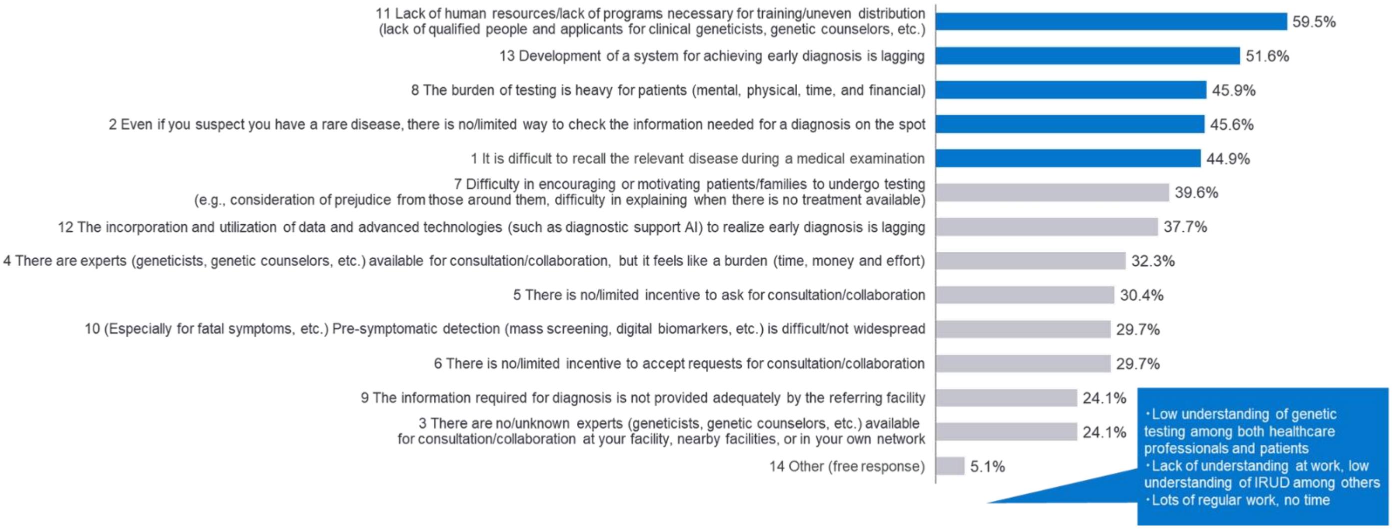
■Survey: Web survey

■Question: Please answer by selecting the top 5 most pressing challenges related to the diagnosis (ranking format)

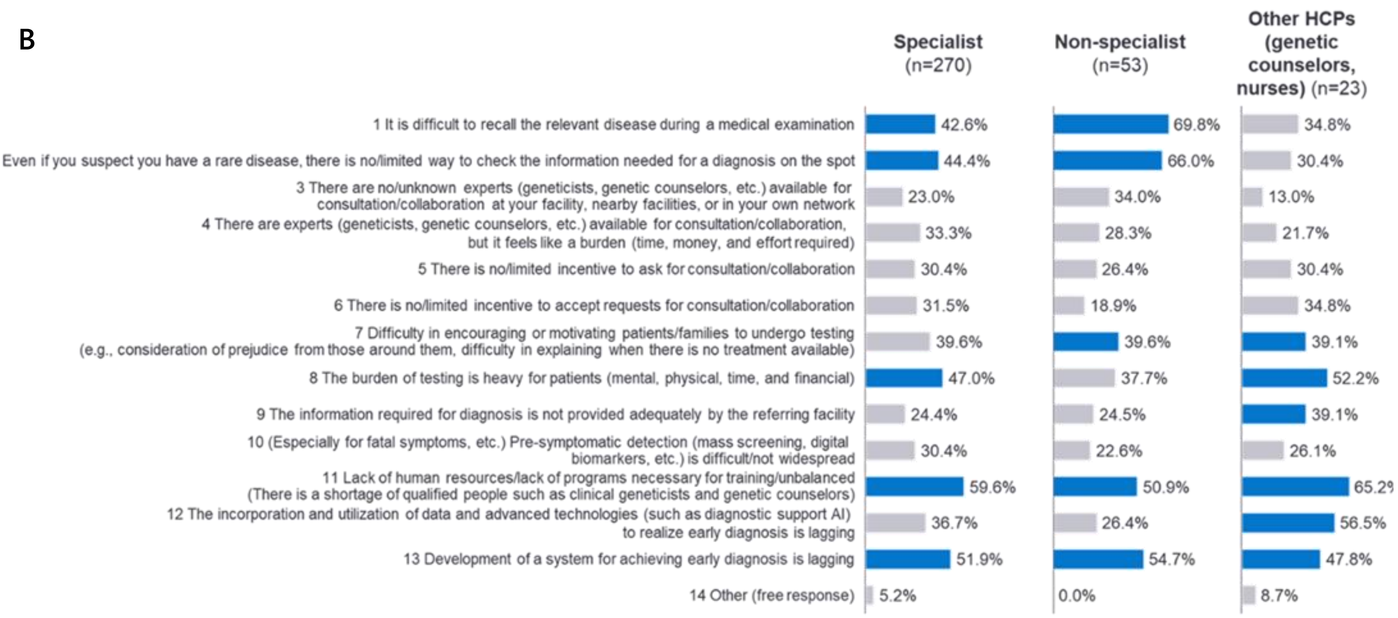
■Subjects: 316 specialists, non-specialists, and other HCPs (genetic counselors and nurses)

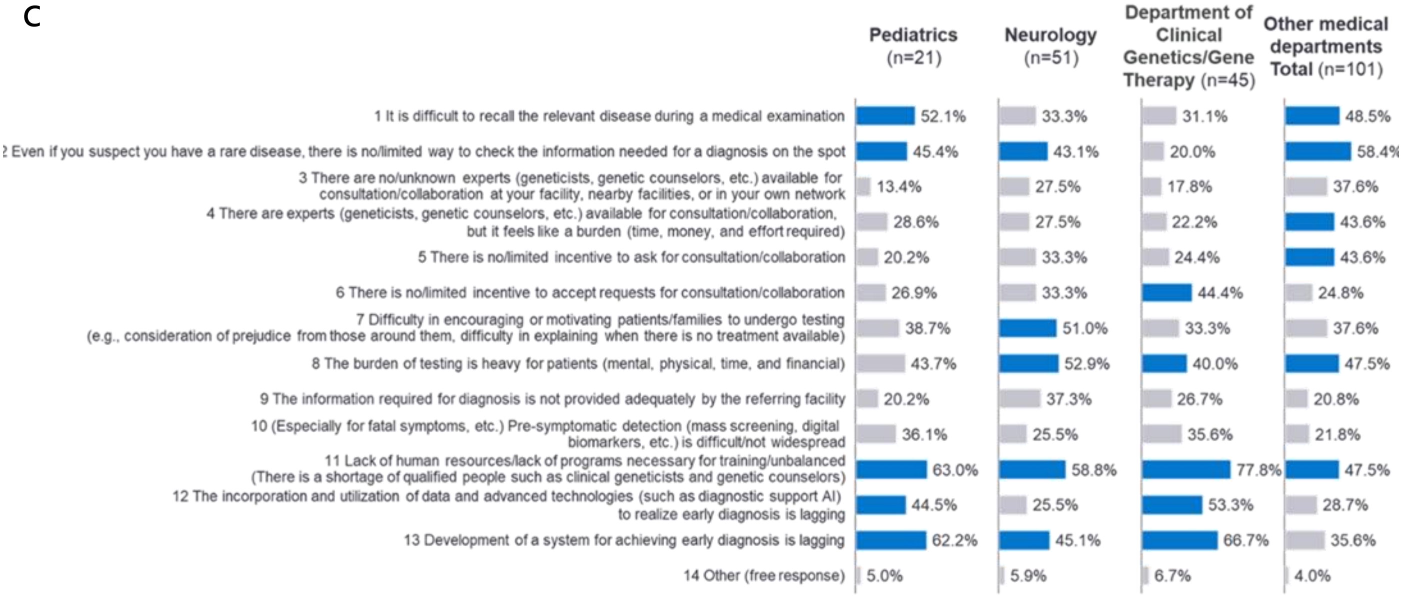
Figure 4.1.4-6: Problems in diagnosis – Top 5 Selection Results：
A all segments ・B by occupation ・C by medical department

A



・Low understanding of genetic testing among both healthcare professionals and patients
・Lack of understanding at work, low understanding of IRUD among others
・Lots of regular work, no time





■Survey: Web survey
■Question: Please select the top 5 most pressing challenges you feel are related to diagnosis (ranking format)
■Subjects: 316 specialists, non-specialists, and other HCPs (genetic counselors and nurses)

“ *There is still room for improvement in diagnosis in the field of pediatric medicine, and ideally, diagnosis would be made more accurately and quickly than it is now. Since diseases can have irreversible adverse effects on the growth of pediatric patients and on the formation of their personalities through life with their families,* ***early intervention by medical institutions and support for creating an environment where parents can feel at ease when dealing with pediatric patients should be provided****.*
(Specialist / Pediatrics)

“ *To reduce the number of facilities and the time it takes to reach a definitive diagnosis, it is necessary to make it easy for cases to accumulate and to provide good access for patients. It is unrealistic to make a definitive diagnosis at*

the first visit, but it is desirable to reach a specialized hospital such as a university hospital once and have the diagnosis confirmed there. To reduce the burden on patients, it is important to avoid repeated transfers to hospitals over a wide area, and the aim should be to complete the diagnosis at a specialized facility. In addition, ***to lower the psychological hurdle for referring doctors, it is essential to clearly indicate where facilities and specialists specializing in rare disease medicine are located, as well as the referral criteria****.*
(Specialist / Collagen Disease)

“ *I feel that the* ***high hurdles in terms of knowledge, technology, and cost (cost/effort) for both doctors and patients when it comes to genetic testing are an issue****. I think it is important to have patients understand the advantages and disadvantages of genetic testing before referring them to a hospital that can perform the test, but especially in urban areas where hospital performance is less clear than in rural areas, there is no guarantee that the hospital has a doctor with sufficient knowledge and experience in genetic testing, so it is not possible to refer patients easily or irresponsibly. Furthermore, it is rare to receive feedback on the patient's test results from the hospital, and even if an appropriate diagnosis is not made, it is difficult to grasp the situation and it is not possible to get the diagnosis back on track. Therefore, I feel that there is a need to visualize information on doctors/facilities with sufficient knowledge of genetic testing and genetic diseases that can be tested at each hospital, and a platform to share test results between specialists and non-specialists.*
(Non-specialist / Neurology)

“ ***There is no/limited access to the information necessary for diagnosis at the time of consultation****, and patients must rely on information provided by websites, papers, and pharmaceutical companies that they find by trial and error. I feel that the lack of evidence at the time of diagnosis is an issue.*
(Non-specialist / Pediatrics)

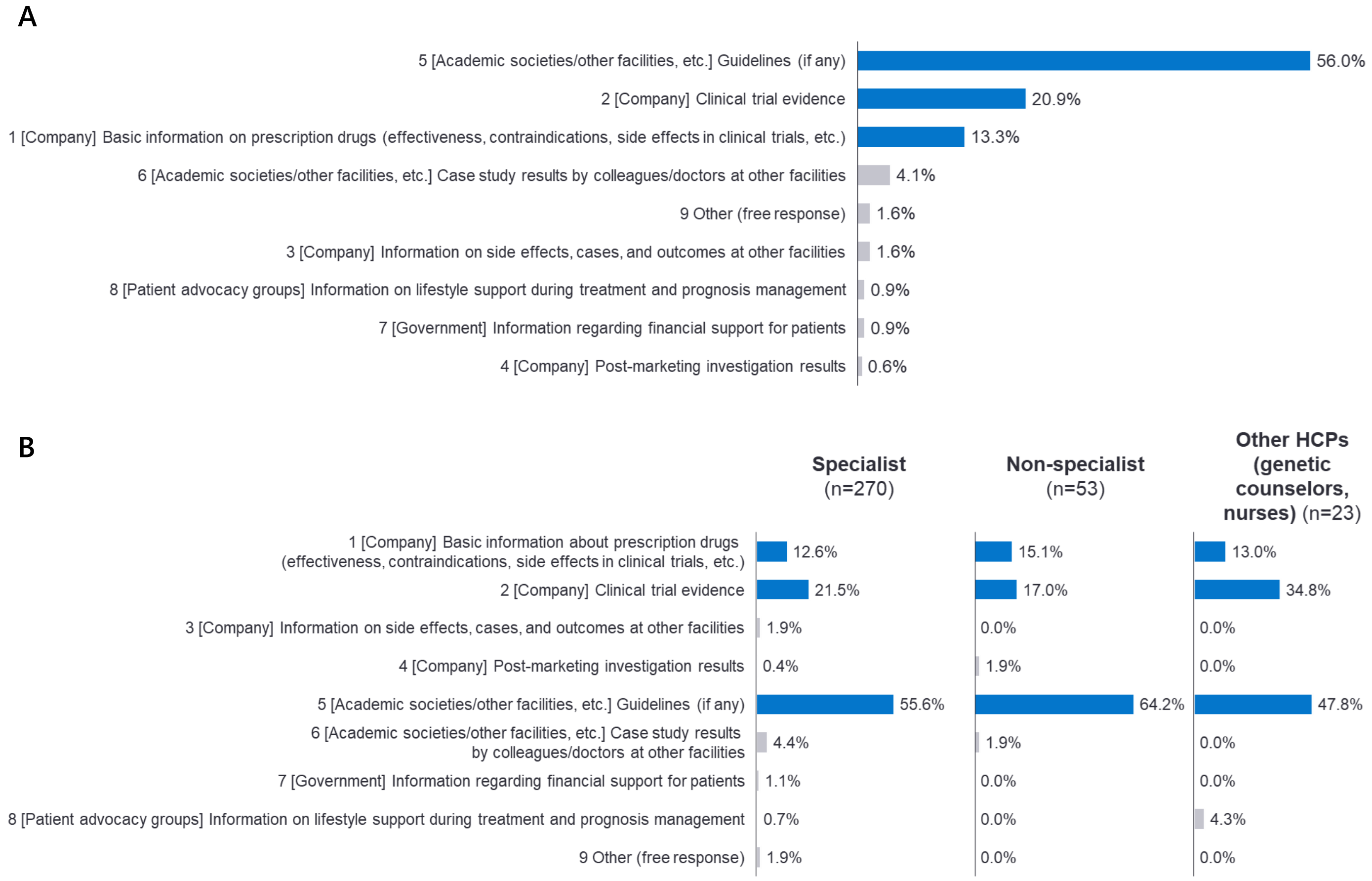
“ ***Patients feel burdened by the tests****, and it is difficult to motivate them to undergo the tests. In fact, when patients undergo the tests, they must pay for the treatment themselves, which places a heavy financial and mental burden on them.*
(Other HCPs (genetic counselors and nurses) / Department of Clinical Genetics and Gene Therapy)

“ *Even if you consult with IRUD, it takes a year for the test results to come out, so as a genetic counselor, I have seen the distress that patients are experiencing. The testing company and others are dealing with the situation carefully, but from the patient's perspective, more timely action is needed.*
(Other HCPs (genetic counselors, nurses) / Department of Clinical Genetics and Gene Therapy)

“ *For doctors who have just started working with rare diseases,* ***not only ideals and motivation are important, but also incentives related to remuneration, time and workload****. What young doctors today are looking for is to work efficiently and fairly as specialists, and degrees and titles themselves are not very motivating.*
(Clinical researcher (development) / neuromuscular disease)

4.1.5 Challenges in treatment and prognosis management

Figure 4.1.5-1: Types of information collected and used in treatment and prognosis management –
Top selection result：
A all segments ・B by occupation

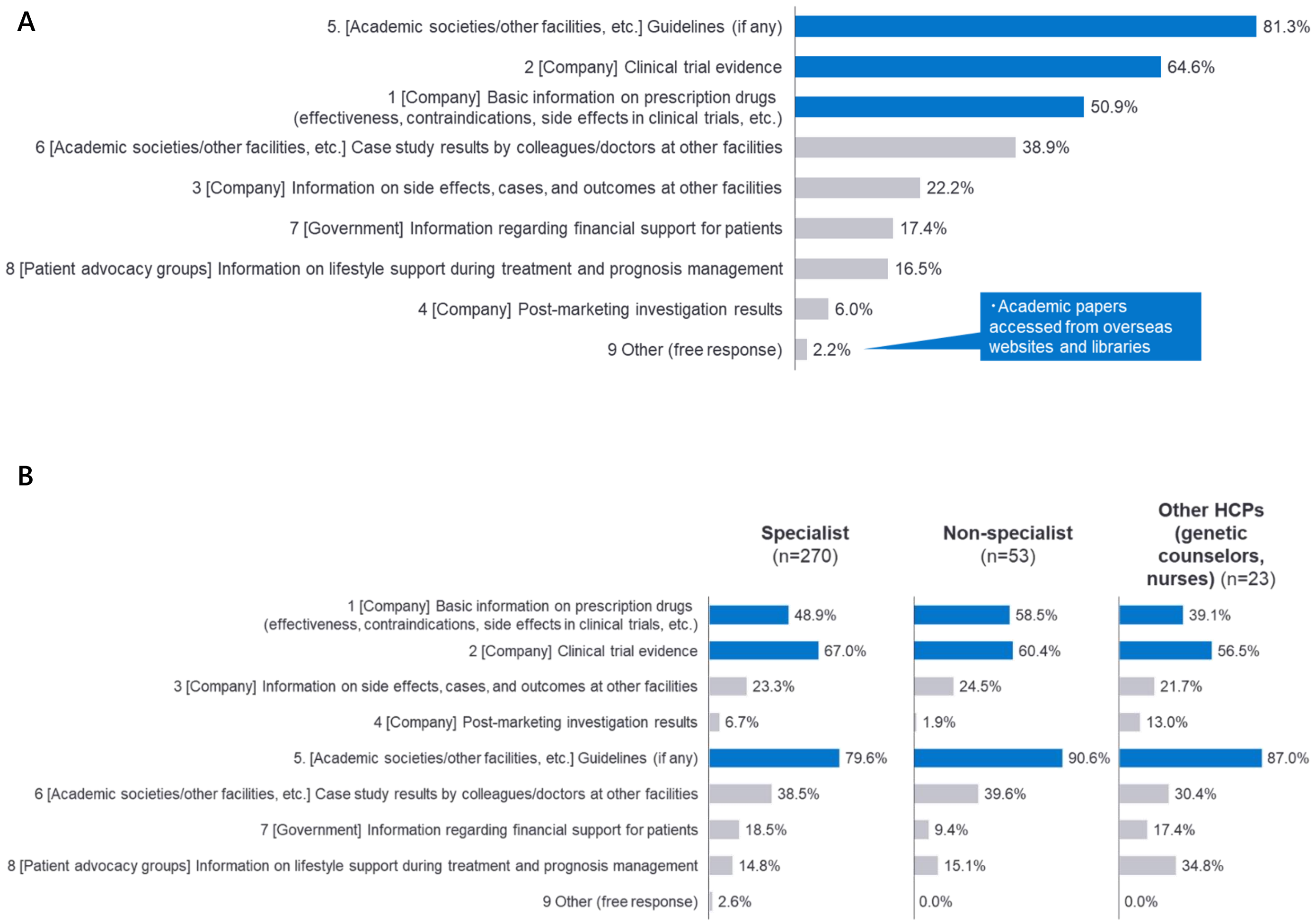


■Survey: Web survey
■Question: Please answer up to the top three types of information that you consider important to collect and use in treatment and prognosis management (ranking format)
■Subjects: 316 specialists, non-specialists, and other HCPs (genetic counselors and nurses)

Figure 4.1.5-2: Types of information collected and used in treatment and prognosis management –

Top 3 Selection Results：

A all segments ・B by occupation



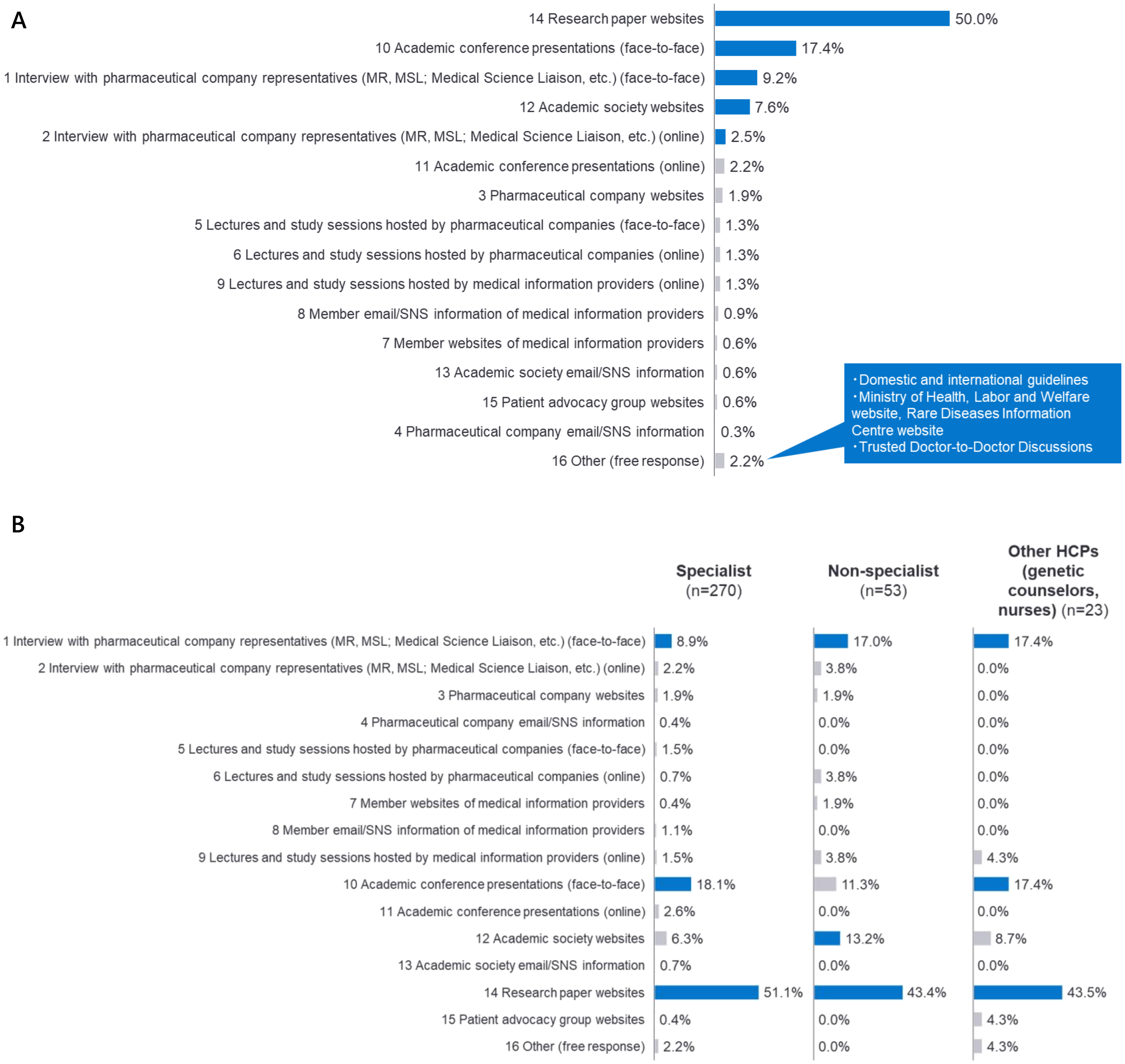
・Academic papers accessed from overseas websites and libraries

■Survey: Web survey

■Question: Please answer the top three types of information that you consider important to collect and use in treatment and prognosis management (ranking format)

■Subjects: 316 specialists, non-specialists, and other HCPs (genetic counselors and nurses)

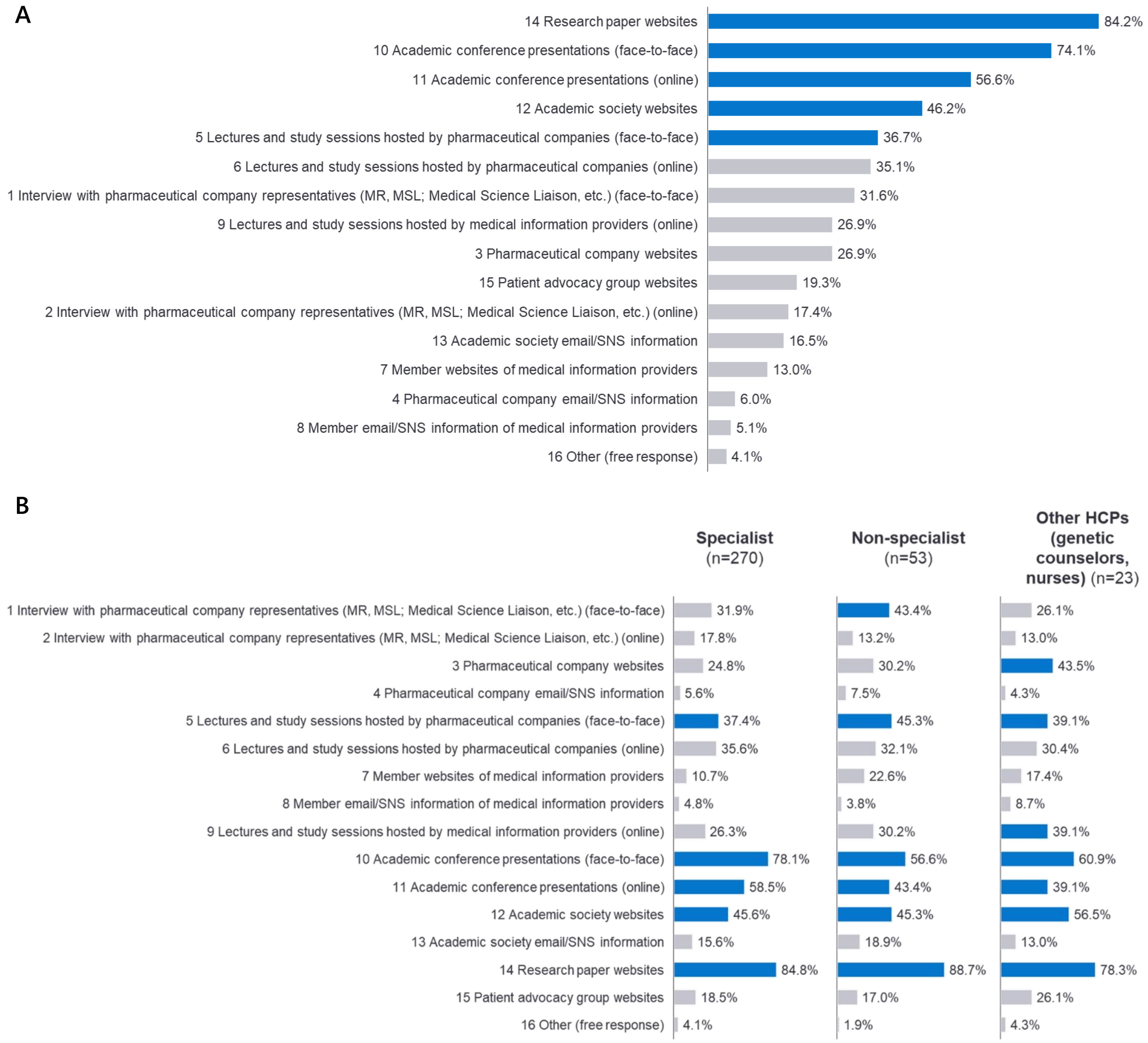
Figure 4.1.5-3: Sources of information collected and utilized in treatment and prognosis management – Top selection results：
A all segments ・B by occupation



・Domestic and international guidelines
・Ministry of Health, Labor and Welfare website, Rare Diseases Information Centre website
・Trusted Doctor-to-Doctor Discussions

■Survey: Web survey
■Question: Please select your top 5 preferred sources (media/channels) of information to be collected and utilized in treatment and prognosis management (ranked)
■Subjects: 316 specialists, non-specialists, and other HCPs (genetic counselors and nurses)

Figure 4.1.5-4: Sources of information collected and utilized in treatment and prognosis management – Top 3 selection results：
A all segments ・B by occupation

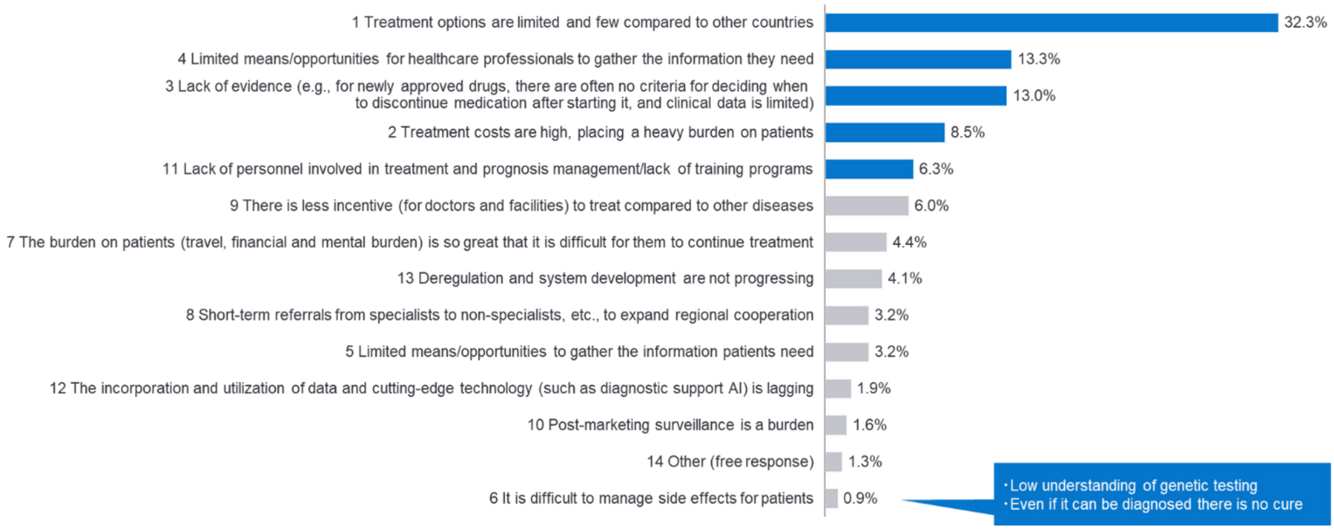


■Survey: Web survey
■Question: Please select your top 5 preferred sources (channels) of information to be collected and utilized in treatment and prognosis management (ranked)
■Subjects: 316 specialists, non-specialists, and other HCPs (genetic counselors and nurses)

Figure 4.1.5-5: Challenges in treatment and prognosis management – Top selection results:

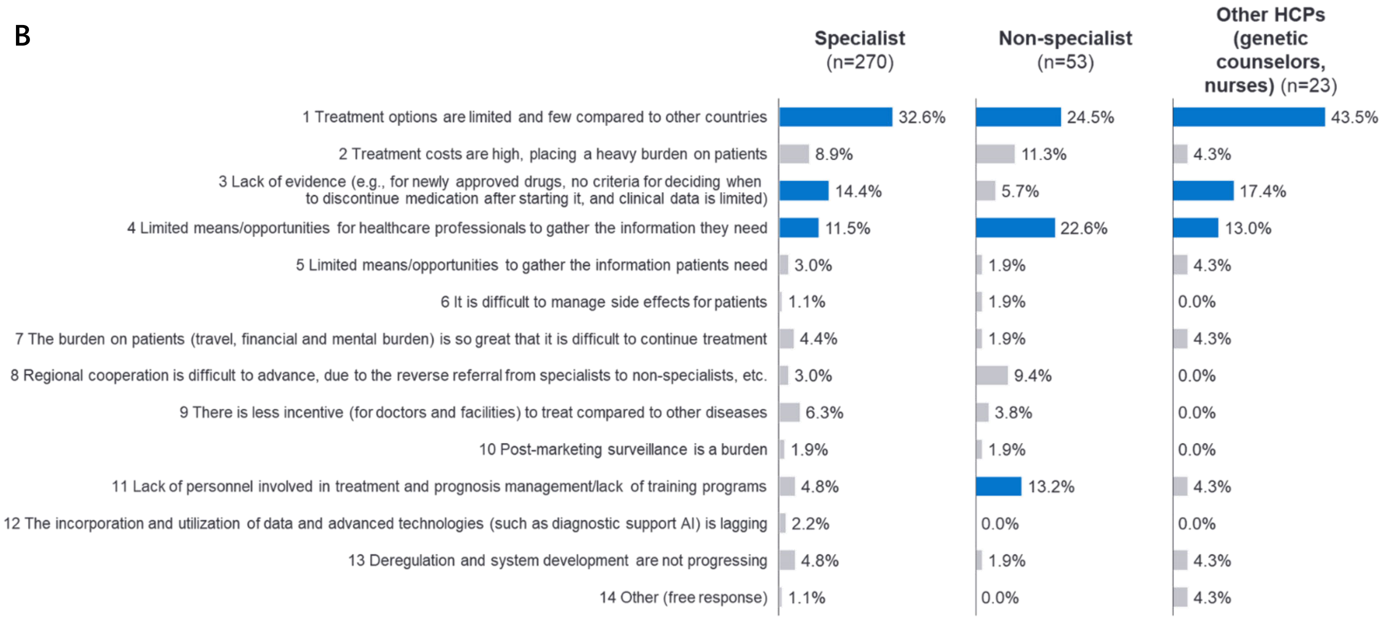
A all segments ・B by occupation

A



・Low understanding of genetic testing
・Even if it can be diagnosed there is no cure

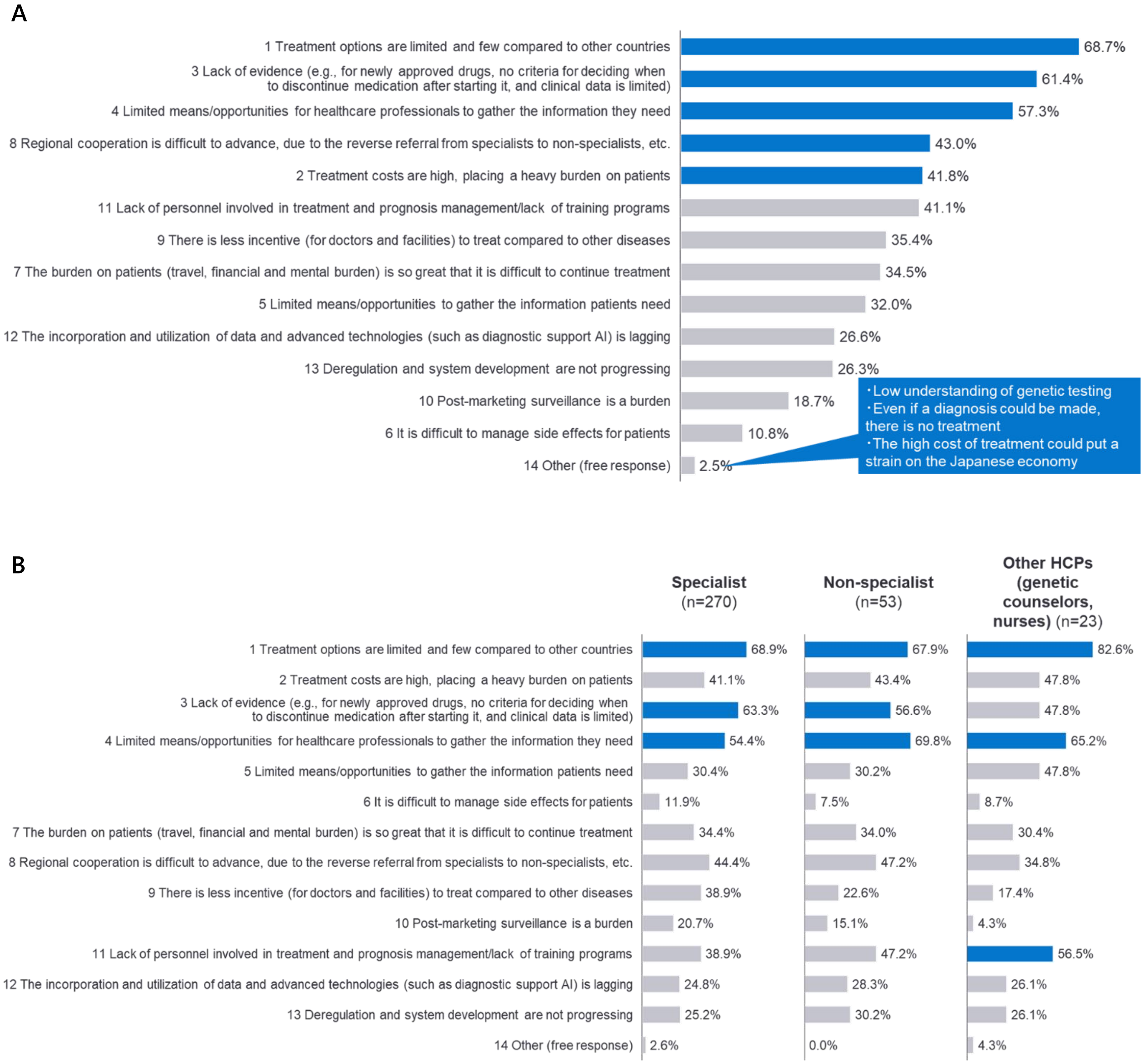
B



■Survey: Web survey
■Question: Please select the top 5 challenges you feel are most important in terms of treatment and prognosis management (ranking format)
■Subjects: 316 specialists, non-specialists, and other HCPs (genetic counselors and nurses)

Figure 4.1.5-6: Challenges in treatment and prognosis management – Top 3 selection results：

A all segments ・B by occupation



- Low understanding of genetic testing
- Even if a diagnosis could be made, there is no treatment
- The high cost of treatment could put a strain on the Japanese economy

■Survey: Web survey
■Question: Please select the top 5 challenges you feel are most challenging regarding treatment and prognosis management (ranking format)
■Subjects: 316 specialists, non-specialists, and other HCPs (genetic counselors and nurses)

> “*Because patient test data is personal information, it is not shared between facilities, and **accessible actual clinical data is limited**. A process and infrastructure are needed to determine diagnostic and treatment plans for specific patients based on shared evidence.*
> (Clinical researcher (development) / Endocrinology and Metabolic Disease)

“ *I feel that **there are many areas where disease-specific guidelines have not yet been established.***
(Clinical researcher (development) / Immunodeficiency disease)

“ *When a specialist refers a patient to a non-specialist, the **non-specialist may not accept the patient if the disease is highly specialized**. Pediatric diseases tend to be highly specialized, so many people find it difficult to deal with such referrals.*
(Non-specialist / Pediatrics)

“ ***There is a lack of knowledge about rare diseases even among healthcare professionals**, which means that they are unable to provide guidance and advice to patients in a timely manner. As a result, cases are referred to the genetics department, which can place a heavy burden on certain individuals, so it is necessary to raise the level of knowledge among healthcare professionals, including doctors. At our hospital, a limited number of genetic counselors oversee all inquiries, but it seems that many of the inquiries are ones that doctors could have answered.*
(Other HCPs (genetic counselors, nurses) / Clinical genetics, genetics department)

“ ***The high cost of rare disease treatment is an issue.** If the disease is designated as intractable, the government provides subsidies, but it seems like a difficult topic when considering medical economics. I also feel that commuting to distant university hospitals is a burden on patients. Even if patients are examined at large hospitals, they return to their hometowns, so we need to strengthen cooperation systems, such as sharing information with local medical institutions that can provide treatment.*
(Other HCPs (genetic counselors and nurses) / Department of Clinical Genetics and Gene Therapy)

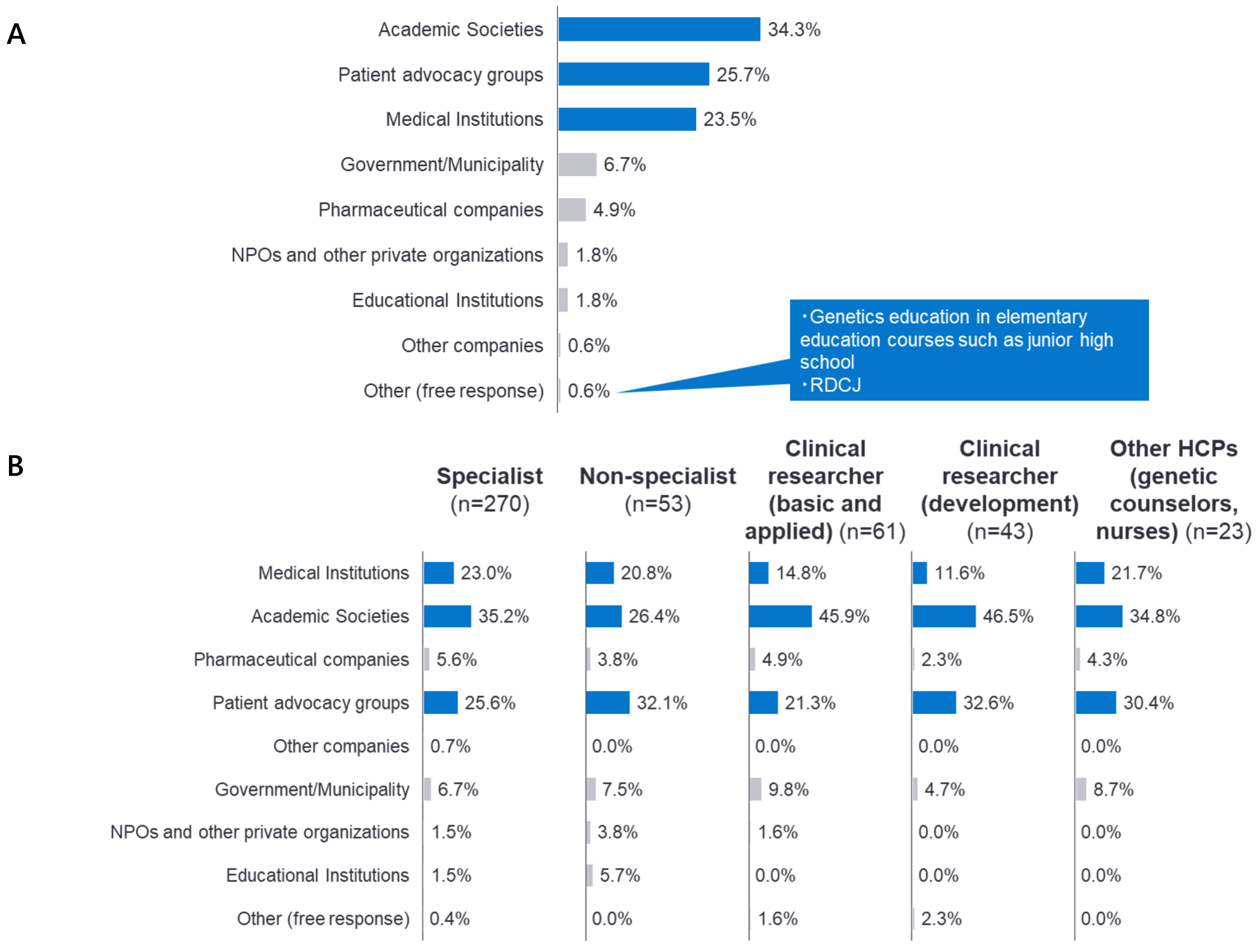
“ *When gathering information for treatment, most literature is in English, so the language barrier is a hurdle, making it **difficult to gather information and gain knowledge**, and ultimately increasing the workload of healthcare professionals.*
(Specialist / Pediatrics)

“ *I feel that **efforts regarding diagnostic and therapeutic drugs are lagging other countries**. When I was having trouble dealing with a patient, I had the opportunity to contact a doctor in the United States directly, and in the United States, I was able to obtain new medicines and treat the patient quickly. In Japan, too, in research and development and clinical practice, I feel that a scheme is needed to smoothly incorporate new technologies and treatments once a certain period has been completed, based on the premise that rare diseases have a different criticality than other diseases.*
(Specialist / Pediatrics)

4.1.6 Challenges in disease awareness activities

Figure 4.1.6-1: Effective organization for disease awareness activities (for patients and their families)

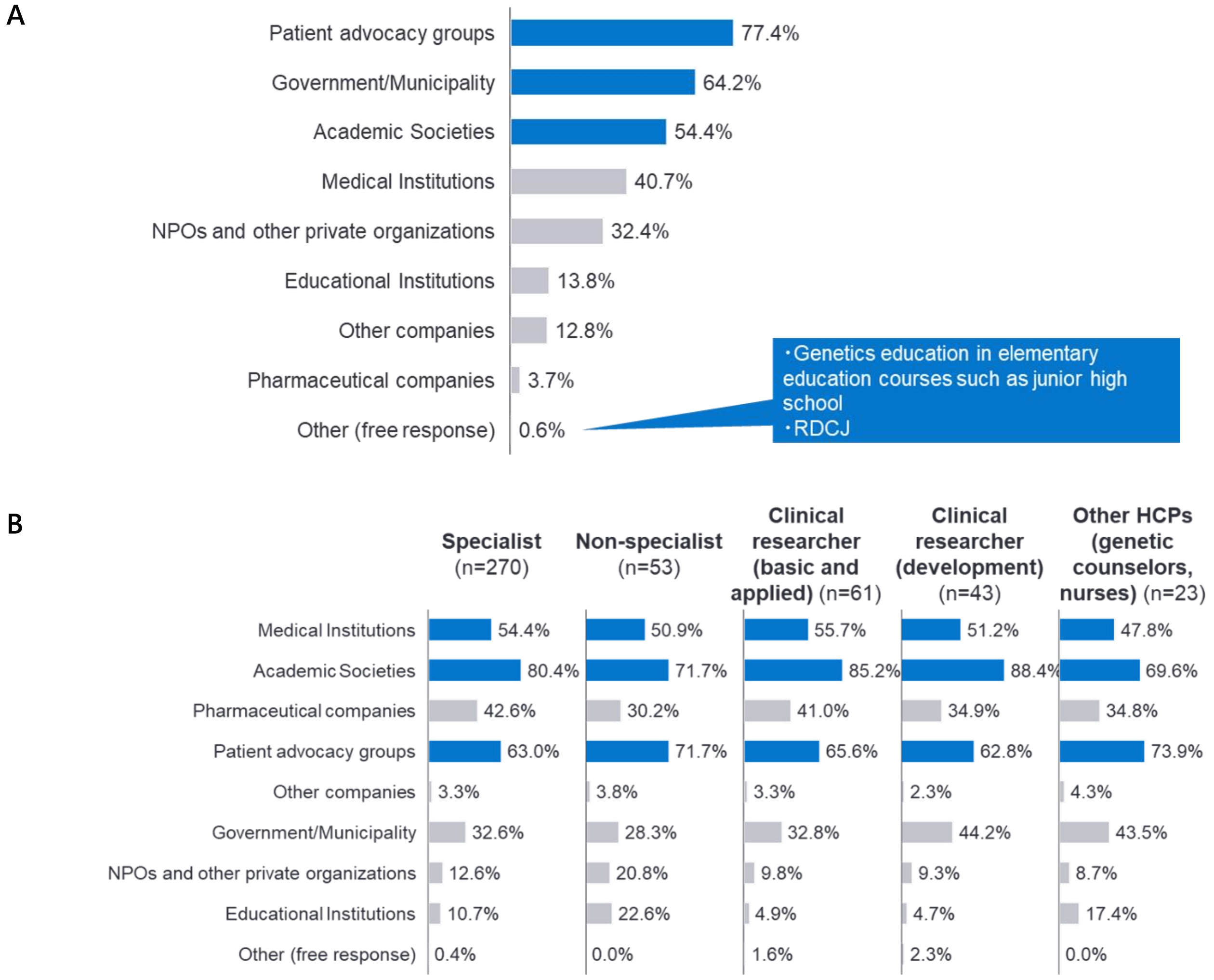
– Top selection results：A all segments ・B by occupation



・Genetics education in elementary education courses such as junior high school
・RDCJ

■Survey: Web survey
■Question: Please answer three options that you feel are effective in raising awareness about rare diseases (for patients and their families) (ranking format)
■Subjects: 327 specialists, non-specialists, clinical researchers (basic and applied), clinical researchers (development) and other HCPs (genetic counselors and nurses)

Figure 4.1.6-2: Effective organization for disease awareness activities (for patients and their families) – Top 3 selection results：
A all segments ・B by occupation

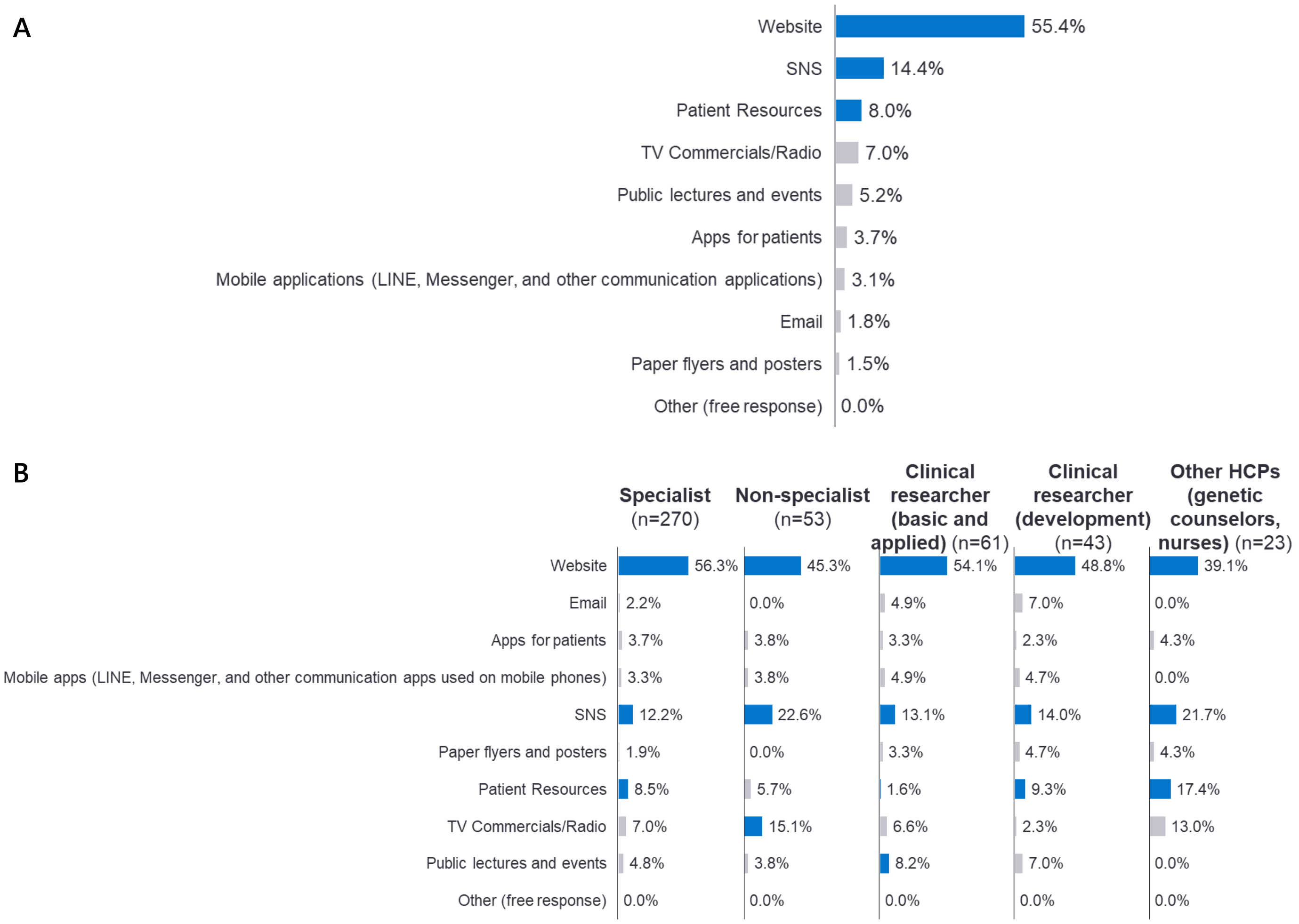


・Genetics education in elementary education courses such as junior high school
・RDCJ

■Survey: Web survey
■Question: Please select three options that you feel are effective in raising awareness about rare diseases (for patients and their families) (ranking format)
■Subjects: 327 specialists, non-specialists, clinical researchers (basic and applied), clinical researchers (development) and other HCPs (genetic counselors and nurses)

Figure 4.1.6-3: Effective media/channels for disease awareness activities (for patients and their families) – Top selection results：

A all segments ・B by occupation

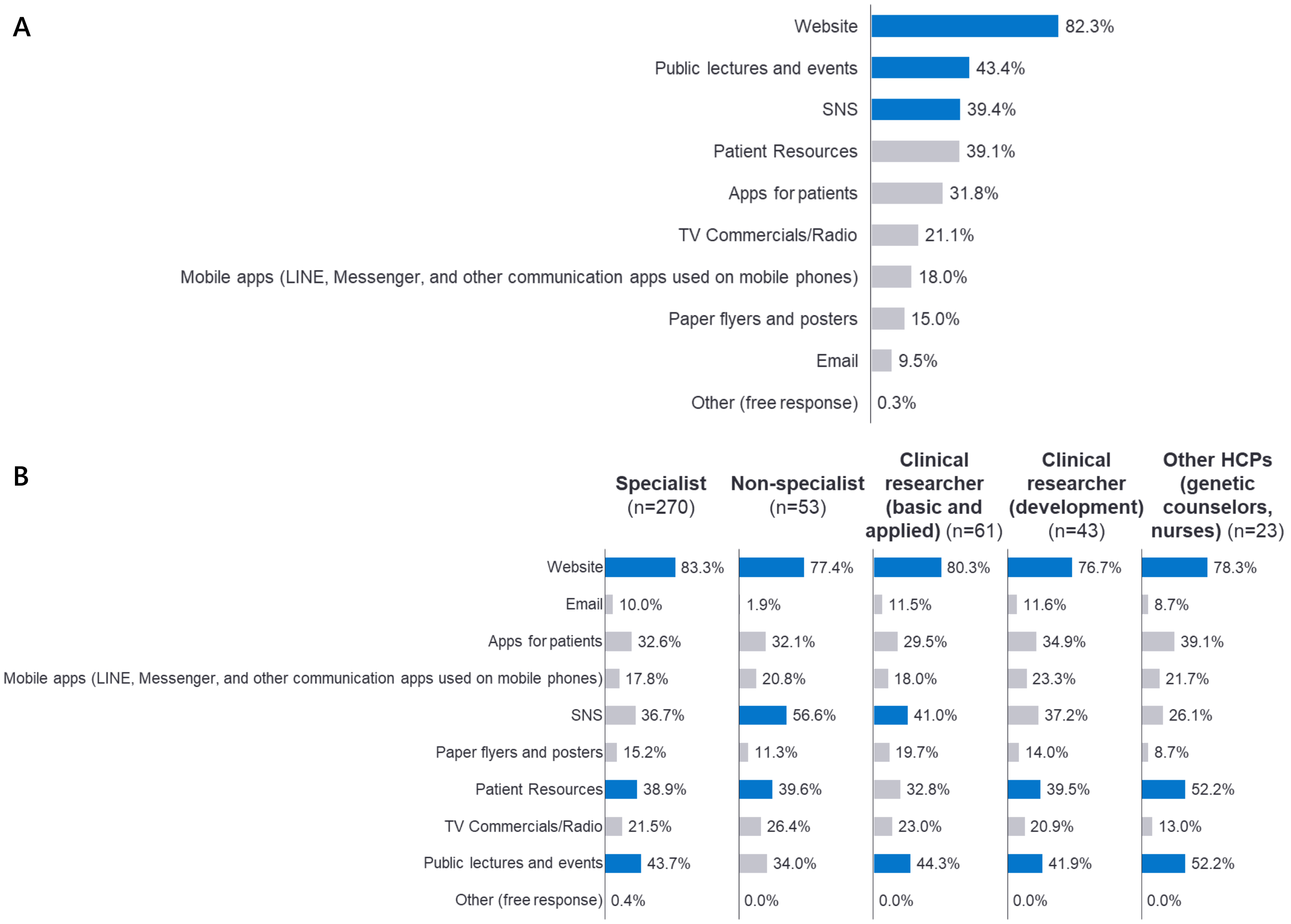


■Survey: Web survey

■Question: Please choose the three most effective media/channels for raising awareness about rare diseases (for patients and their families) (ranked)

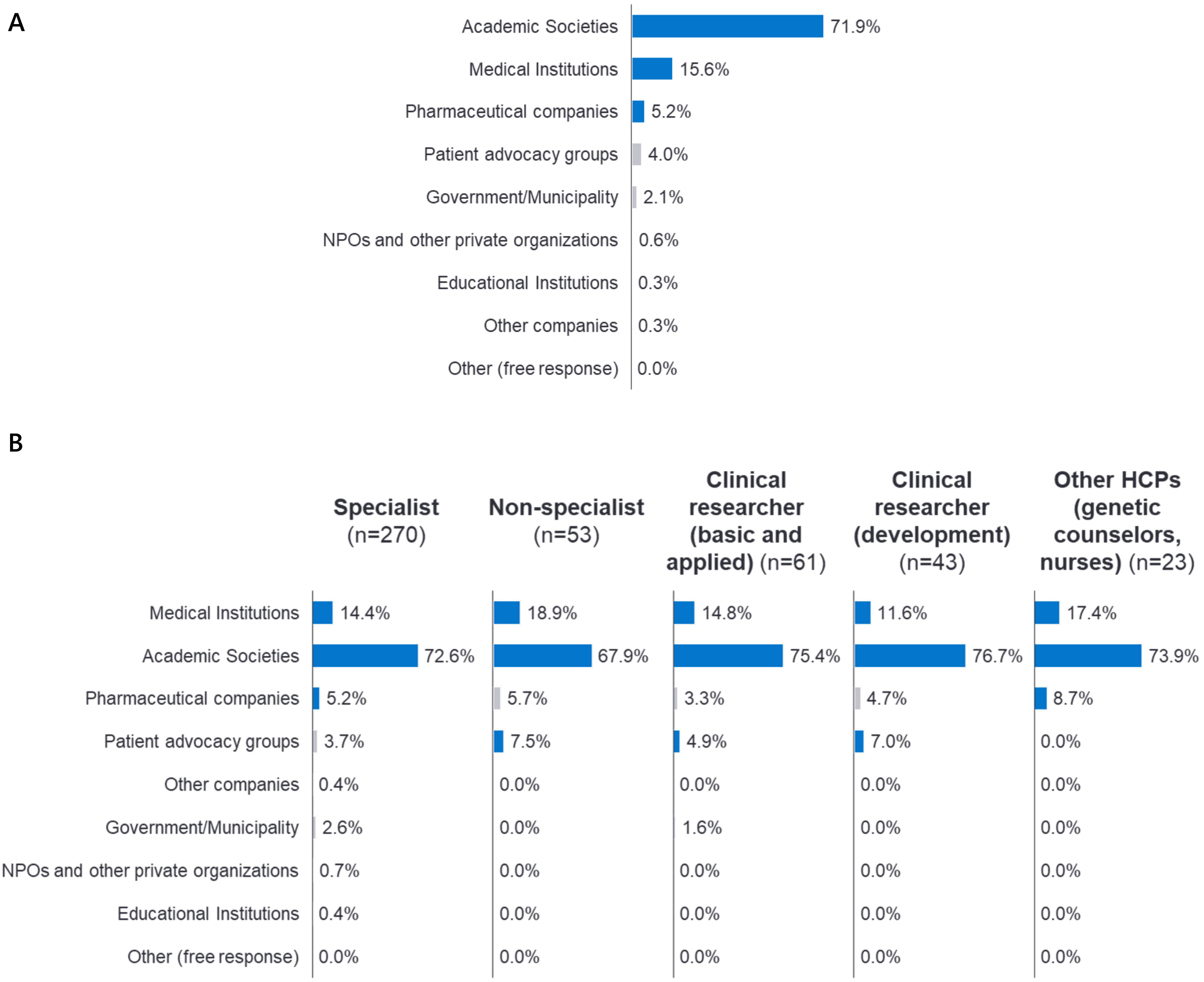
■Subjects: 327 specialists, non-specialists, clinical researchers (basic and applied), clinical researchers (development) and other HCPs (genetic counselors and nurses)

Figure 4.1.6-4: Effective media/channels for disease awareness activities (for patients and their families) – Top 3 selection results：
A all segments ・B by occupation



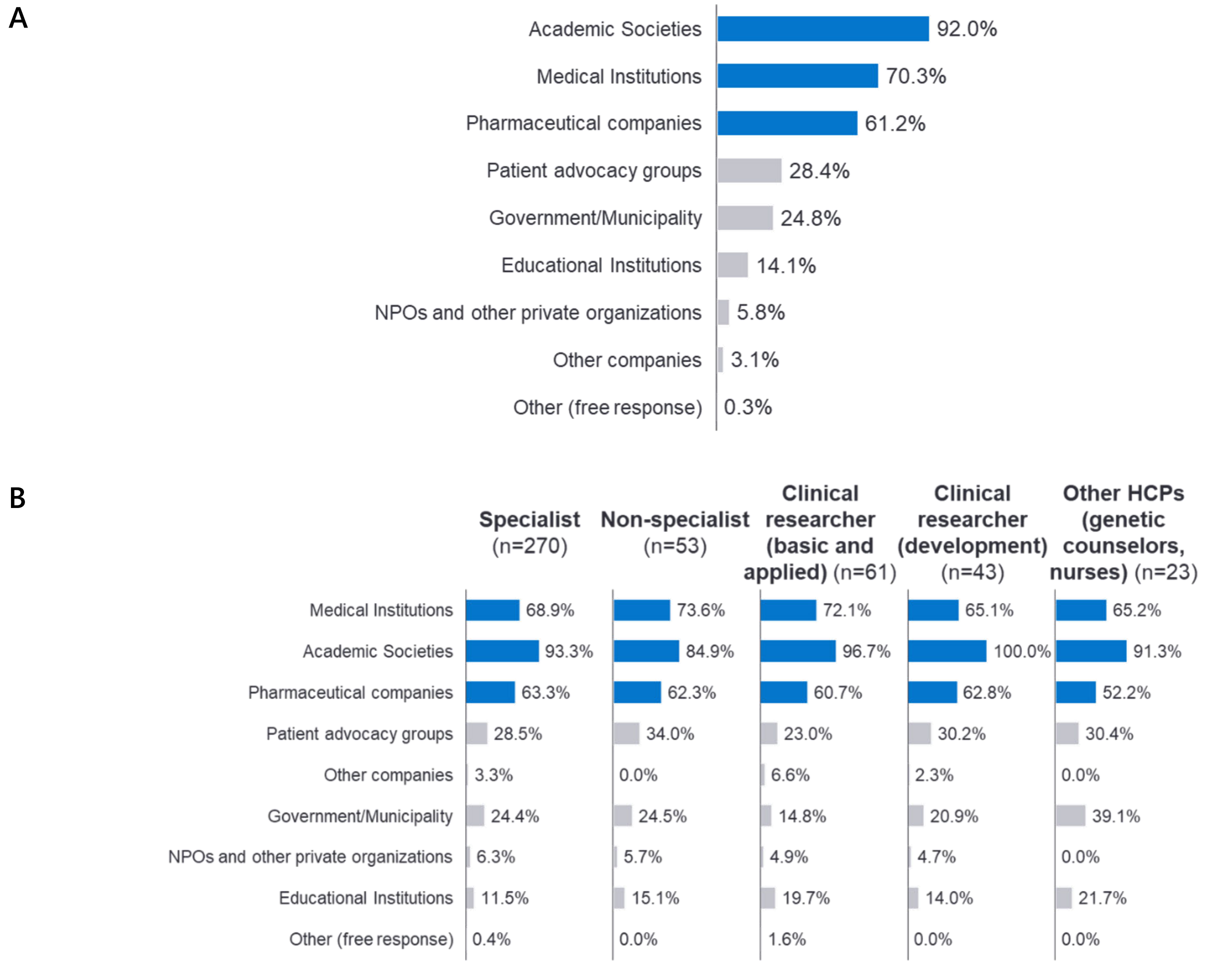
■Survey: Web survey
■Question: Please choose the three most effective media/channels for raising awareness about rare diseases (for patients and their families) (ranked)
■Subjects: 327 specialists, non-specialists, clinical researchers (basic and applied), clinical researchers (development) and other HCPs (genetic counselors and nurses)

Figure 4.1.6-5: Effective organization for disease awareness activities (for healthcare professionals)
– Top selection results：
A all segments ・B by occupation



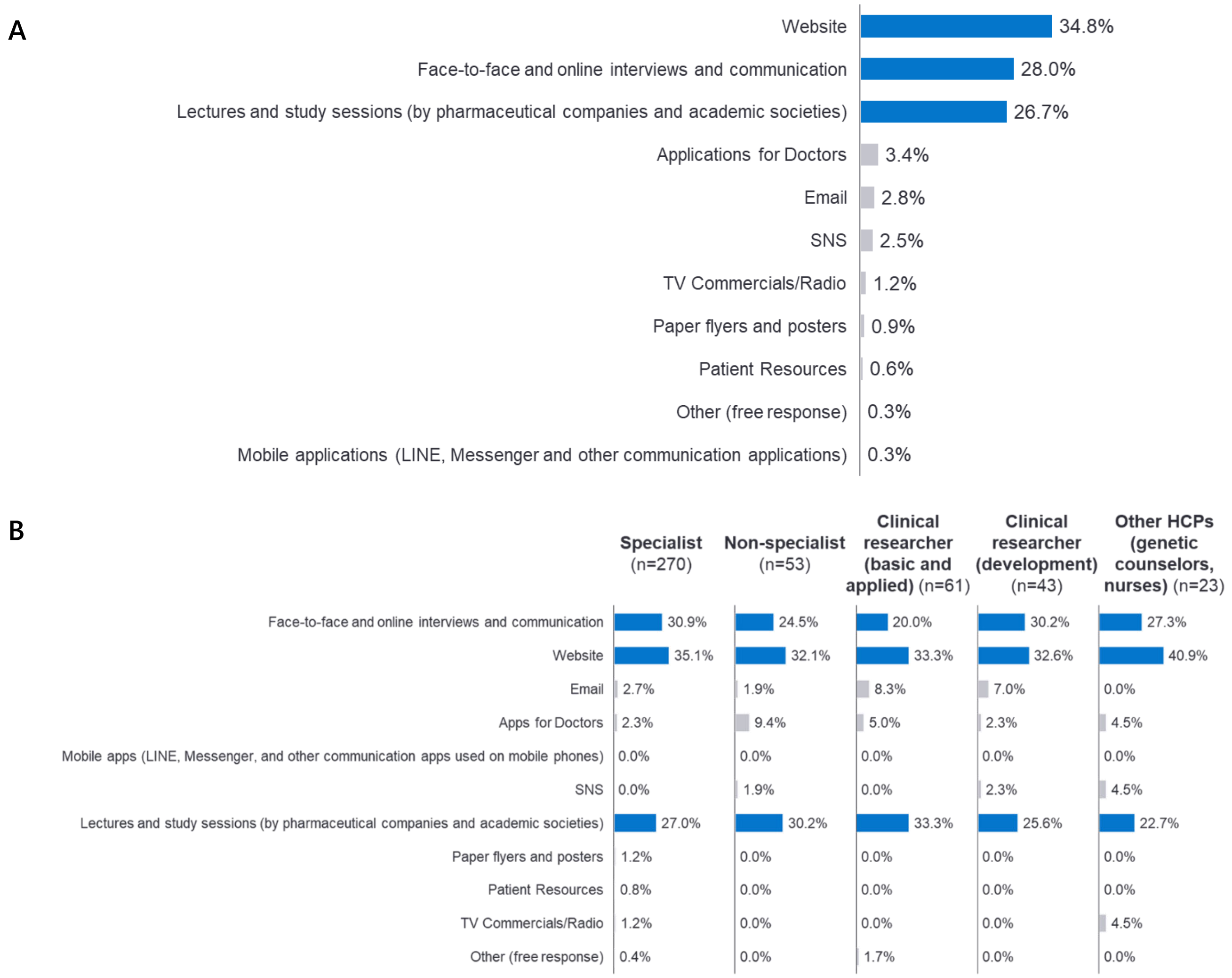
■Survey: Web survey
■Question: Please select three options that you feel are effective in raising awareness of rare diseases (for healthcare professionals) (ranking format)
■Subjects: 327 specialists, non-specialists, clinical researchers (basic and applied), clinical researchers (development) and other HCPs (genetic counselors and nurses)

Figure 4.1.6-6: Effective organization for disease awareness activities (for healthcare professionals) – Top 3 selection results：
A all segments ・B by occupation



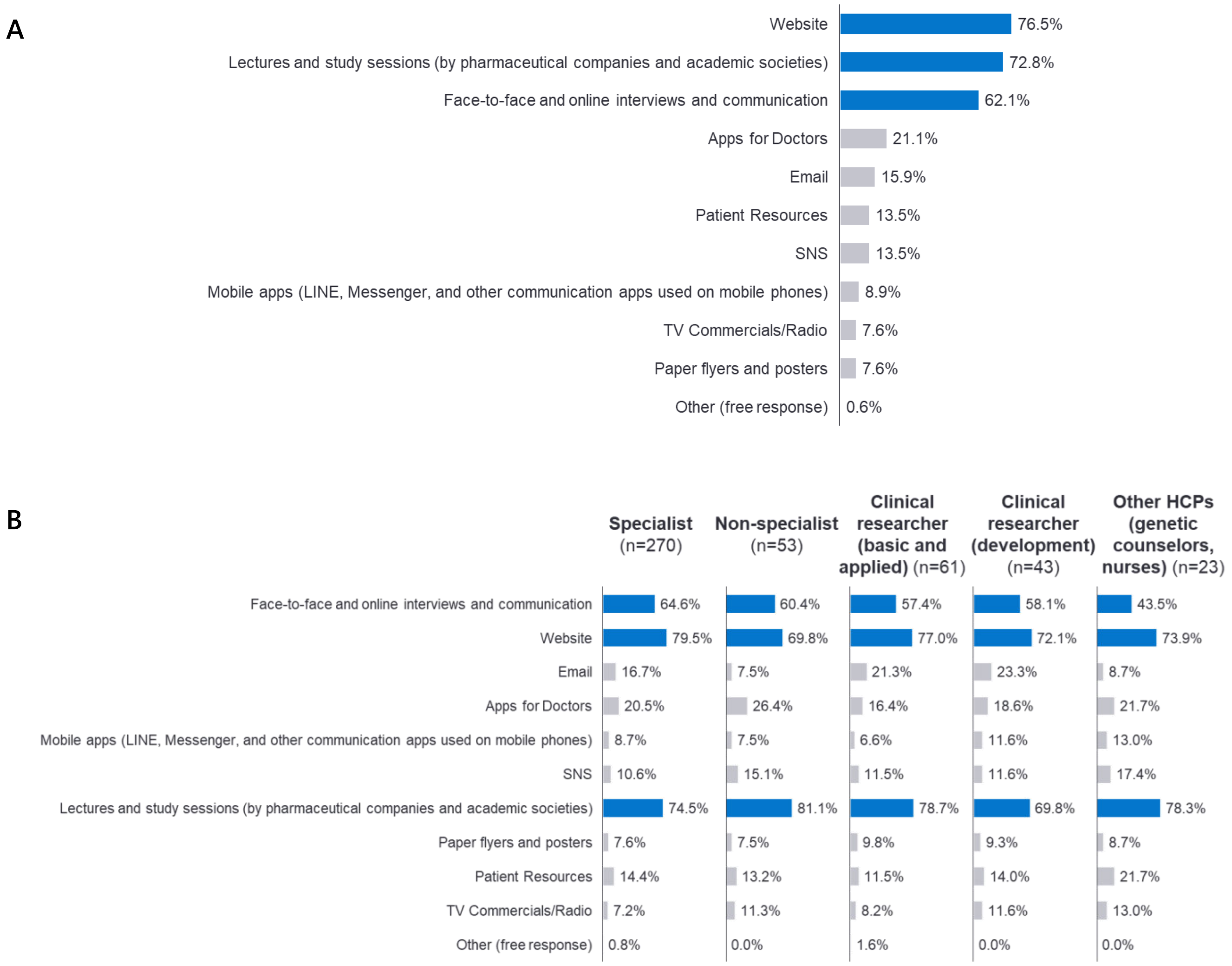
■Survey: Web survey
■Question: Please select three options that you feel are effective in raising awareness of rare diseases (for healthcare professionals) (ranking format)
■Subjects: 327 specialists, non-specialists, clinical researchers (basic and applied), clinical researchers (development) and other HCPs (genetic counselors and nurses)

Figure 4.1.6-7: Effective media/channels for disease awareness activities (for healthcare professionals) – Top selection results：
A all segments ・B by occupation



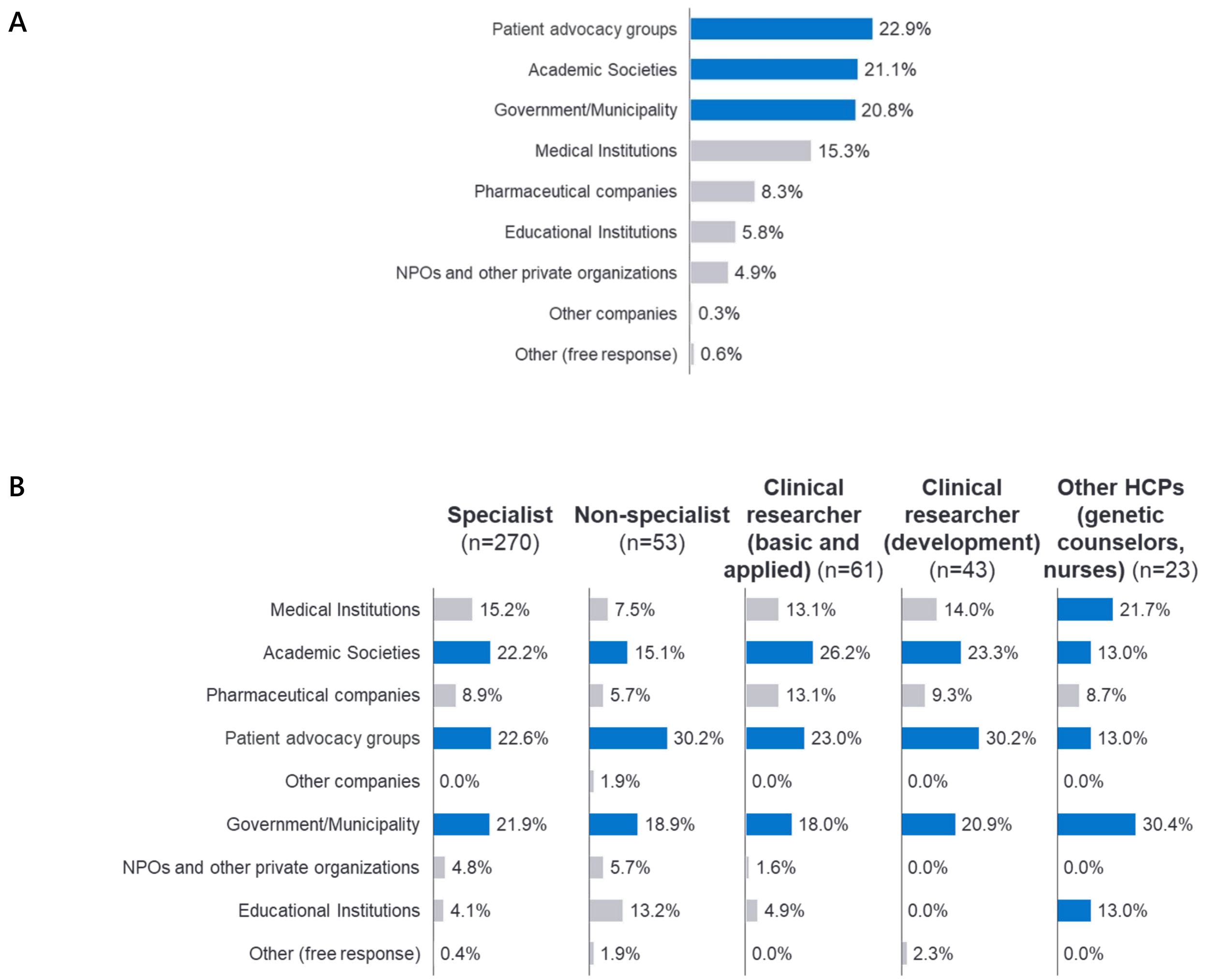
■Survey: Web survey
■Question: Please choose the three most effective media/channels for raising awareness of rare diseases (for healthcare professionals) (ranked)
■Subjects: 327 specialists, non-specialists, clinical researchers (basic and applied), clinical researchers (development) and other HCPs (genetic counselors and nurses)

Figure 4.1.6-8: Effective media/channels for disease awareness activities (for healthcare professionals) – Top 3 selection results：
A all segments ・B by occupation



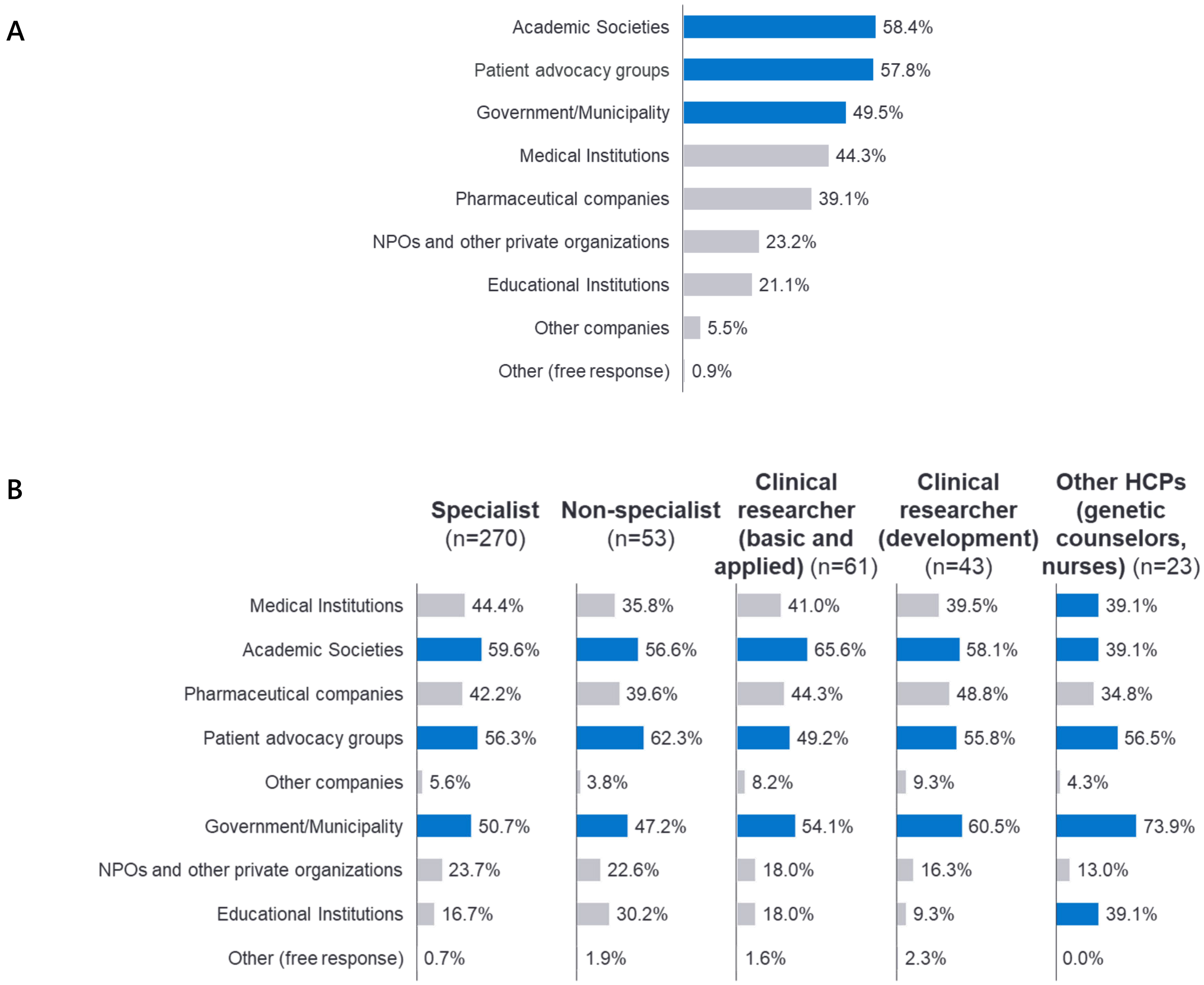
■Survey: Web survey
■Question: Please choose the three most effective media/channels for raising awareness of rare diseases (for healthcare professionals) (ranked)
■Subjects: 327 specialists, non-specialists, clinical researchers (basic and applied), clinical researchers (development) and other HCPs (genetic counselors and nurses)

Figure 4.1.6-9: Effective organization for disease awareness activities (for the public) – Top selection results：
A all segments ・B by occupation



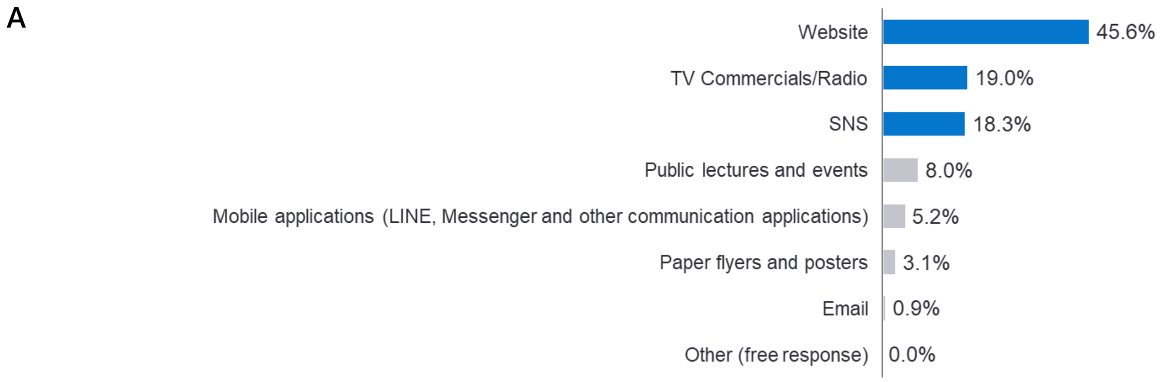
■Survey: Web survey
■Question: Please select three options that you feel are effective in raising awareness of rare diseases (for the general public) (ranking format)
■Subjects: 327 specialists, non-specialists, clinical researchers (basic and applied), clinical researchers (development) and other HCPs (genetic counselors and nurses)

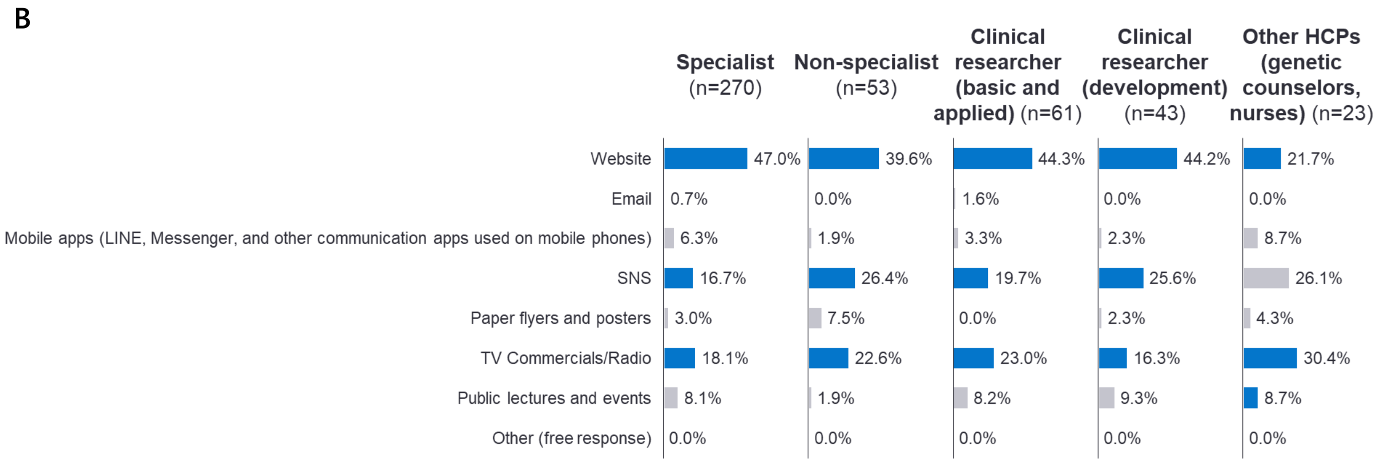
Figure 4.1.6-10: Effective organization for disease awareness activities (for the public) – Top 3 selection results：A all segments ・B by occupation



■Survey: Web survey
■Question: Please select three options that you feel are effective in raising awareness of rare diseases (for the public) (ranking format)
■Subjects: 327 specialists, non-specialists, clinical researchers (basic and applied), clinical researchers (development) and other HCPs (genetic counselors and nurses)

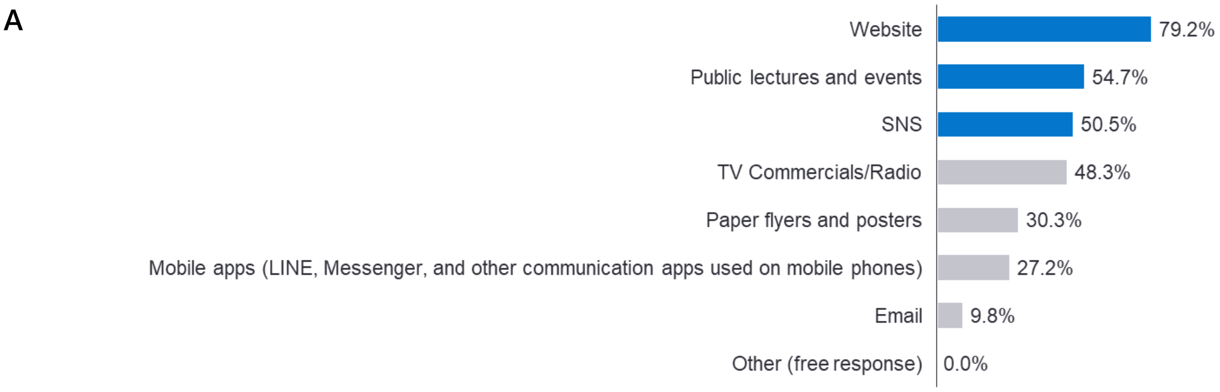
Figure 4.1.6-11: Effective media/channels for disease awareness activities (for the public) – Top selection result：A all segments ・B by occupation

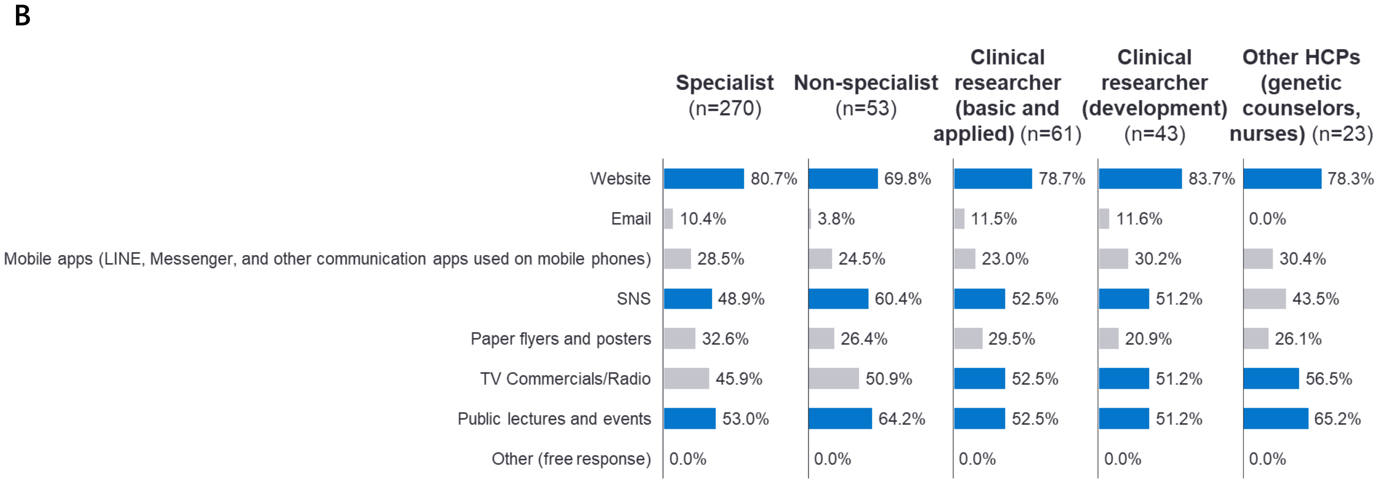




■Survey: Web survey
■Question: Please select the three most effective media/channels for raising awareness of rare diseases (for the public) (ranked)
■Subjects: 327 specialists, non-specialists, clinical researchers (basic and applied), clinical researchers (development) and other HCPs (genetic counselors and nurses)

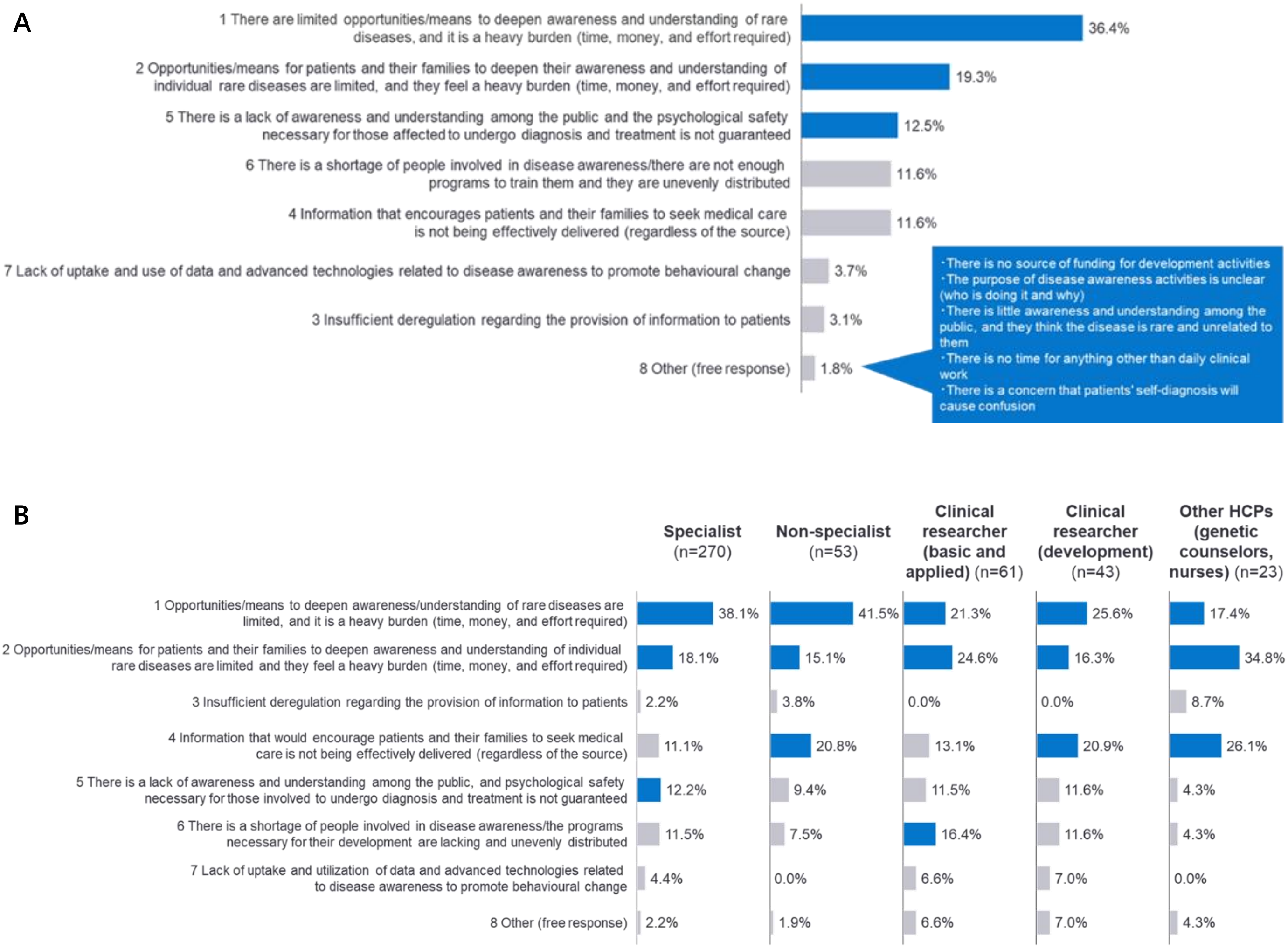
Figure 4.1.6-12: Effective media/channels for disease awareness activities (for the public) – Top 3 selection results：
A all segments ・B by occupation





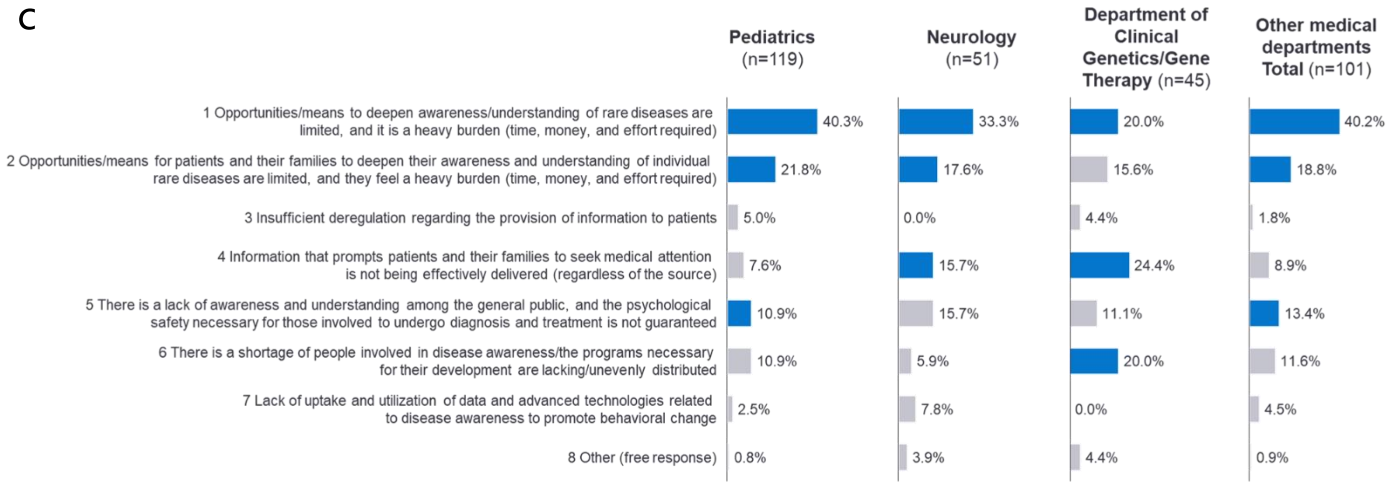
■Survey: Web survey
■Question: Please select the three media/channels that you think are effective for in-depth awareness activities (for the general public) on rare diseases (ranked)
■Subjects: 327 specialists, non-specialists, clinical researchers (basic and applied), clinical researchers (development) and other HCPs (genetic counselors and nurses)

Figure 4.1.6-13: Challenges in disease awareness activities – Top selection results：
A all segments ・B by occupation ・C by medical department ・D by disease research area

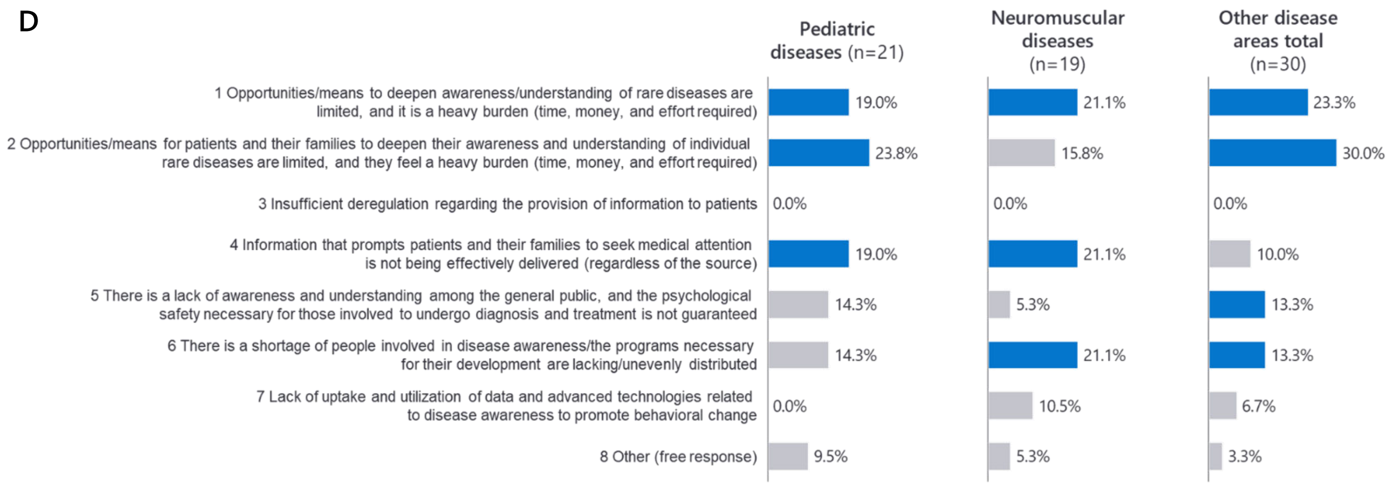


・There is no source of funding for development activities
・The purpose of disease awareness activities is unclear (who is doing it and why)
・There is little awareness and understanding among the public, and they think the disease is rare and unrelated to them
・There is no time for anything other than daily clinical work
・There is a concern that patients' self-diagnosis will cause confusion

■Survey: Web survey
■Question: Please select the three most important challenges in relation to disease awareness activities (ranked)
■Subjects: 327 specialists, non-specialists, clinical researchers (basic and applied), clinical researchers (development) and other HCPs (genetic counselors and nurses)



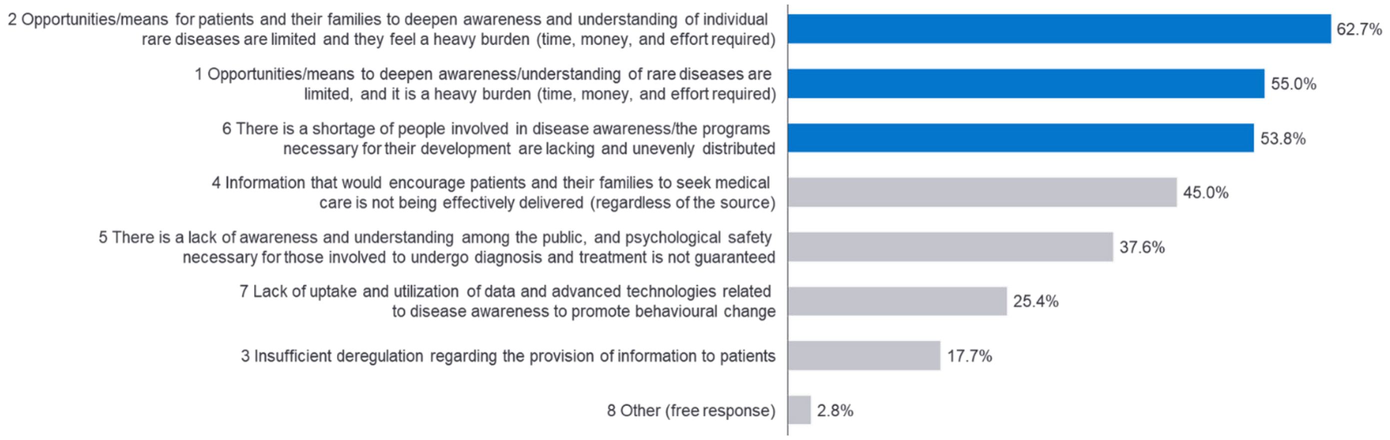
■Survey: Web survey
■Question: Please select the three most important challenges in relation to disease awareness activities (ranked)
■Subjects: 316 specialists, non-specialists, and other HCPs (genetic counselors and nurses)



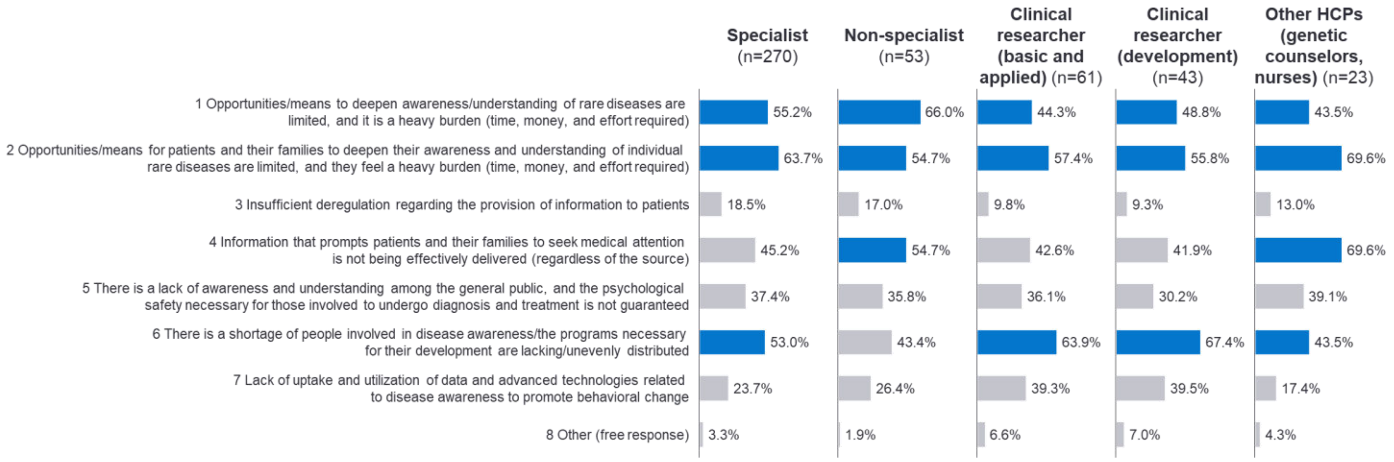
■Survey: Web survey
■Question: Please select the three most important challenges in relation to disease awareness activities (ranked)
■Subjects: 70 clinical researchers (basic and applied) and clinical researchers (development)

Figure 4.1.6-14: Challenges in disease awareness activities – Top 3 selection results：
A all segments ・B by occupation・C by medical department ・D by disease research area

A

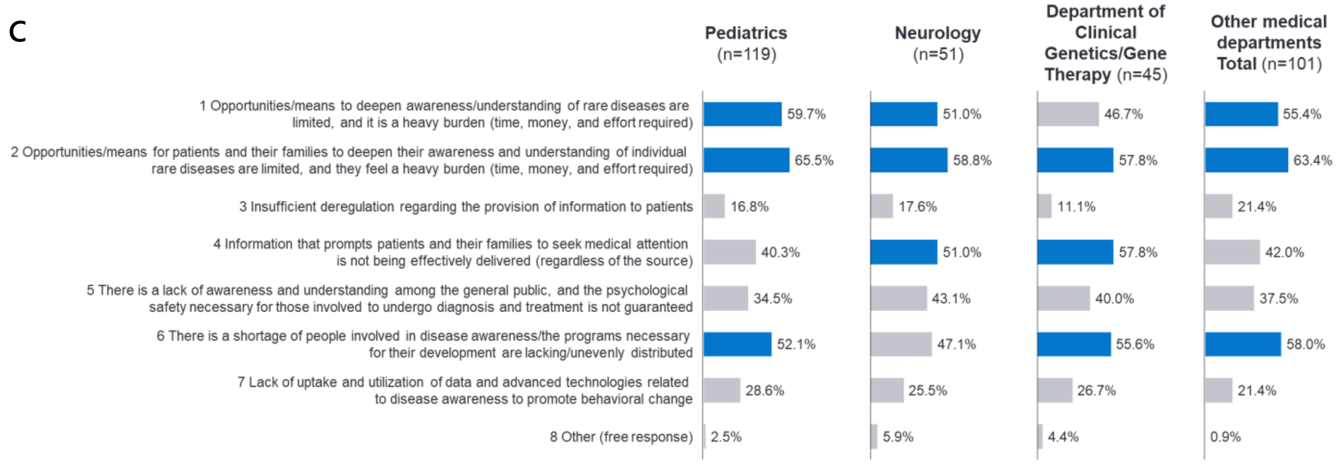


B

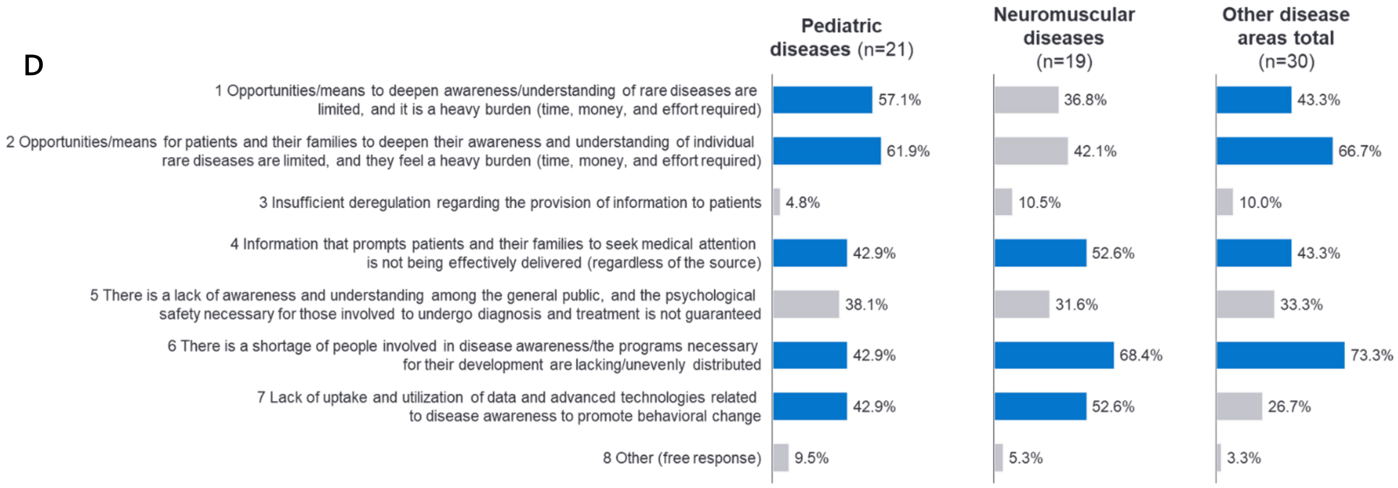


■Survey: Web survey
■Question: Please select the three most important challenges in relation to disease awareness activities (ranked)
■Subjects: 327 specialists, non-specialists, clinical researchers (basic and applied), clinical researchers (development) and other HCPs (genetic counselors and nurses)

C



■Survey: Web survey
■Question: Please select the three most important challenges in relation to disease awareness activities (ranked)
■Subjects: 316 specialists, non-specialists, and other HCPs (genetic counselors and nurses)



■ Survey: Web survey
■ Question: Please select the three most important challenges in relation to disease awareness activities (ranked)
■ Subjects: 70 clinical researchers (basic and applied) and clinical researchers (development)

“ *Due to the* ***uneven distribution of human resources*** *and bases involved in rare diseases, there are some regions where opportunities to learn about rare diseases are not provided, especially in the training curriculum for medical interns. In such cases, there are no rare disease role models during the career development stage, so young doctors lose the opportunity to become interested in/motivated by rare diseases. In such a situation, the result is that human resources involved in rare diseases are not developed.*
It is also ***difficult for patients to gather the information they need****. Because patients do not know the tools to use to gather information or the optimal way to search, the hurdle of gathering information is even higher than for healthcare professionals. Not being able to obtain or understand information causes anxiety in patients, so it is felt to be an issue.*
(Specialist / Pediatrics)

“ ***There is a lack of accurate and up-to-date information provided to patients and their families****, and many patients do not visit the hospital in the first place because their parents are not aware that there are treatments available for their child's developmental delays. Many patients and families of pediatric patients do not recognize or understand the disease, testing methods, or benefits of diagnosis before the patient even seeks a diagnosis.*
(Non-specialist / Pediatrics)

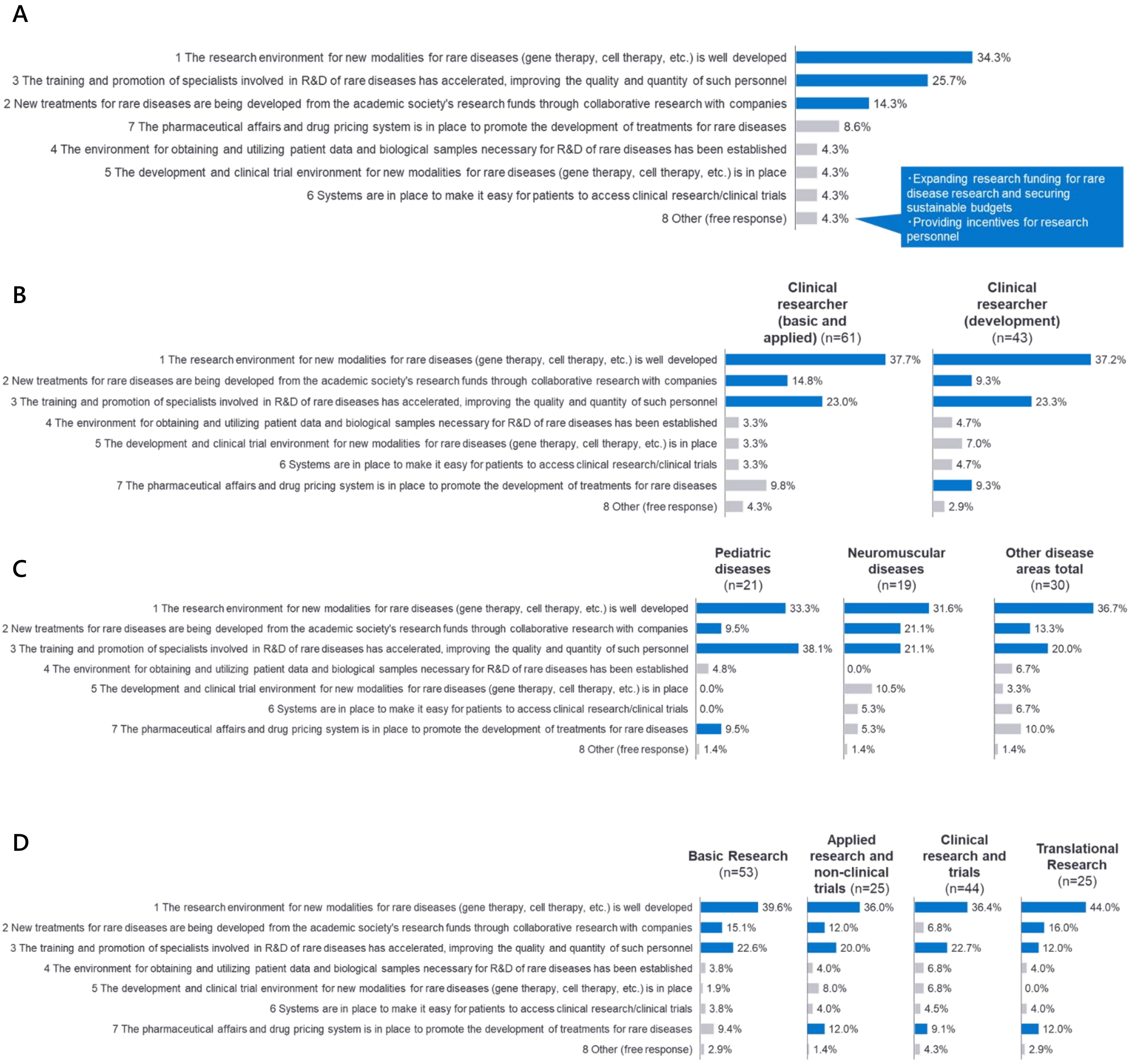
“ *As discussed at the Genomic Medicine Promotion conference, in Japan,* ***even within the family, there is prejudice in society where conflict exists, and people feel discriminatory attitudes even if they don't tell others about it****. To solve these problems, it is necessary to foster awareness that 'everyone has genetic changes' through genetic education for children, and to create a system that makes it easy for anyone to go out into society.*
(Other HCPs (genetic counselors, nurses) / Department of Clinical Genetics and Gene Therapy)

“*I feel that information regarding diagnosis made through genetic testing that directly leads to treatment has not yet been sufficiently constructed and disseminated to healthcare professionals.*
(Clinical researcher (development) / Endocrinology and Metabolic Disease)

“***It is difficult to convey correct information to patients, considering the differences in their positions and levels of understanding.*** *Prior knowledge, expectations, and enthusiasm vary from person to person, and there is a risk that healthcare professionals will be misled if they communicate without sufficient knowledge.*
(Specialist / Pediatrics)

4.2.1 Ideal state in research, development and clinical practice

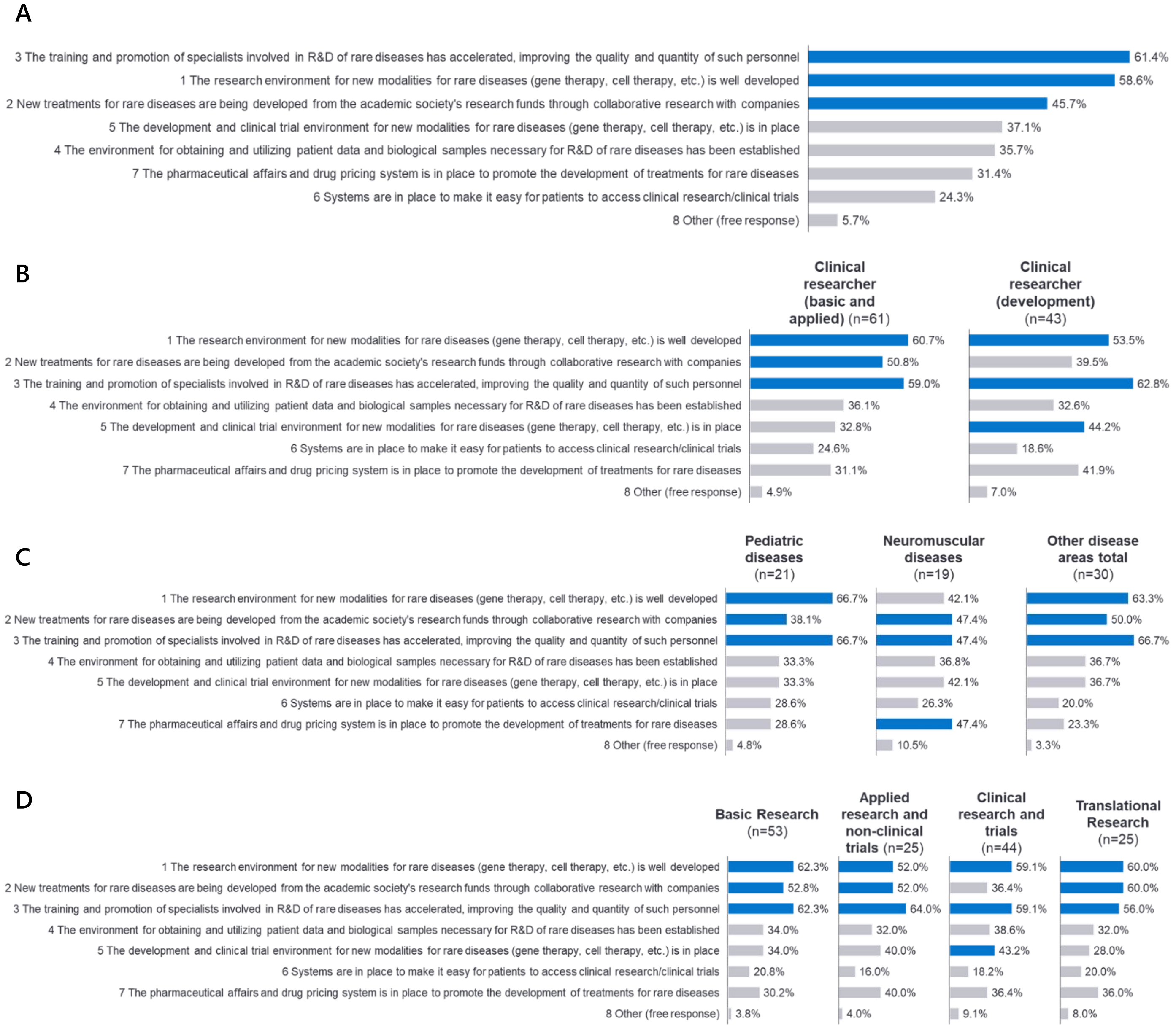
Figure 4.2.1-1: What research and development should be like – Top selection result：
A all segments ・B by occupation ・C by disease research area・ D by specialty



・Expanding research funding for rare disease research and securing sustainable budgets
・Providing incentives for research personnel

■Survey: Web survey
■Question: Based on the research and development challenges you have answered so far, please choose the top three that you agree are the way things should be (ranked)
■Subjects: 70 clinical researchers (basic and applied) and clinical researchers (development)

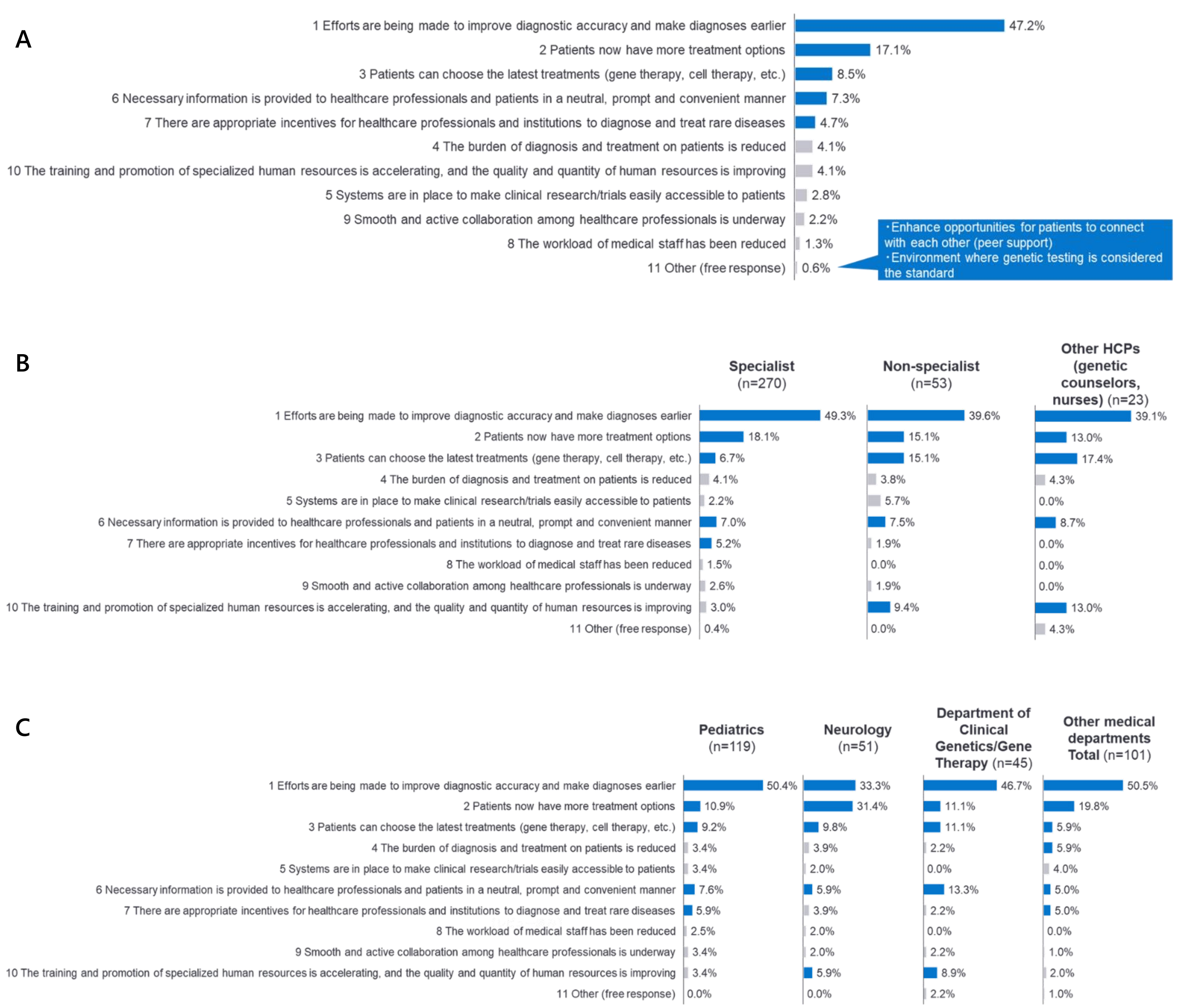
Figure 4.2.1-2: What research and development should be like – Top 3 selection results：
A all segments ・B by occupation ・C by disease research area・ D by specialty



■Survey: Web survey
■Question: Based on the research and development challenges you have answered so far, please choose the top three that you agree are the way things should be (ranked)
■Subjects: 70 clinical researchers (basic and applied) and clinical researchers (development)

Figure 4.2.1-3: What should happen in clinical practice – Top selection result:

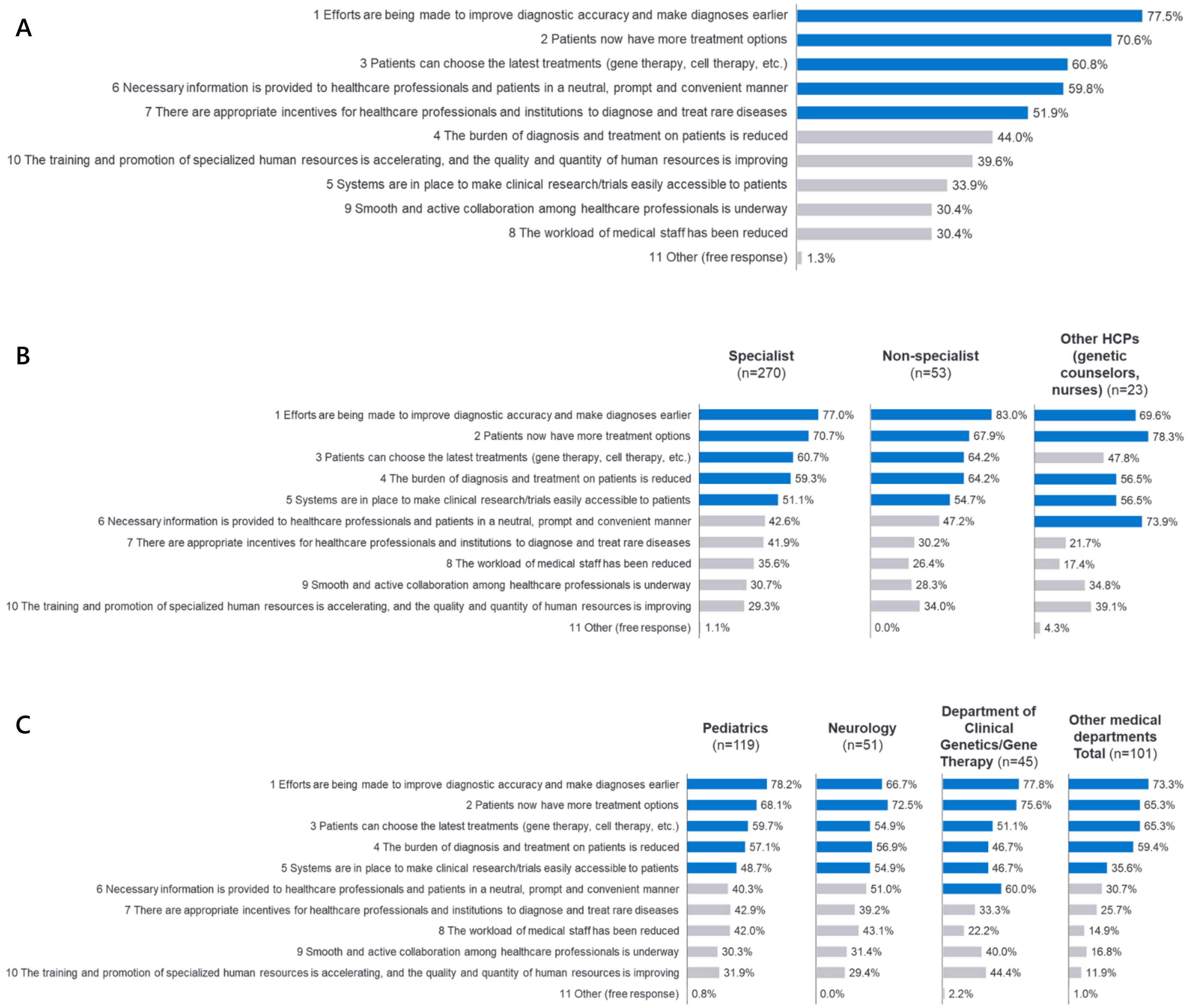
A all segments ・B by occupation ・C by medical department



■Survey: Web survey
■Question: Based on the challenges you have answered so far regarding clinical practice, please select the top 5 that you agree are the way things should be (ranked)
■Subjects: 316 specialists, non-specialists, and other HCPs (genetic counselors and nurses)

Figure 4.2.1-4: What should happen in clinical practice – Top 3 selection results：

A all segments ・B by occupation ・C by medical department



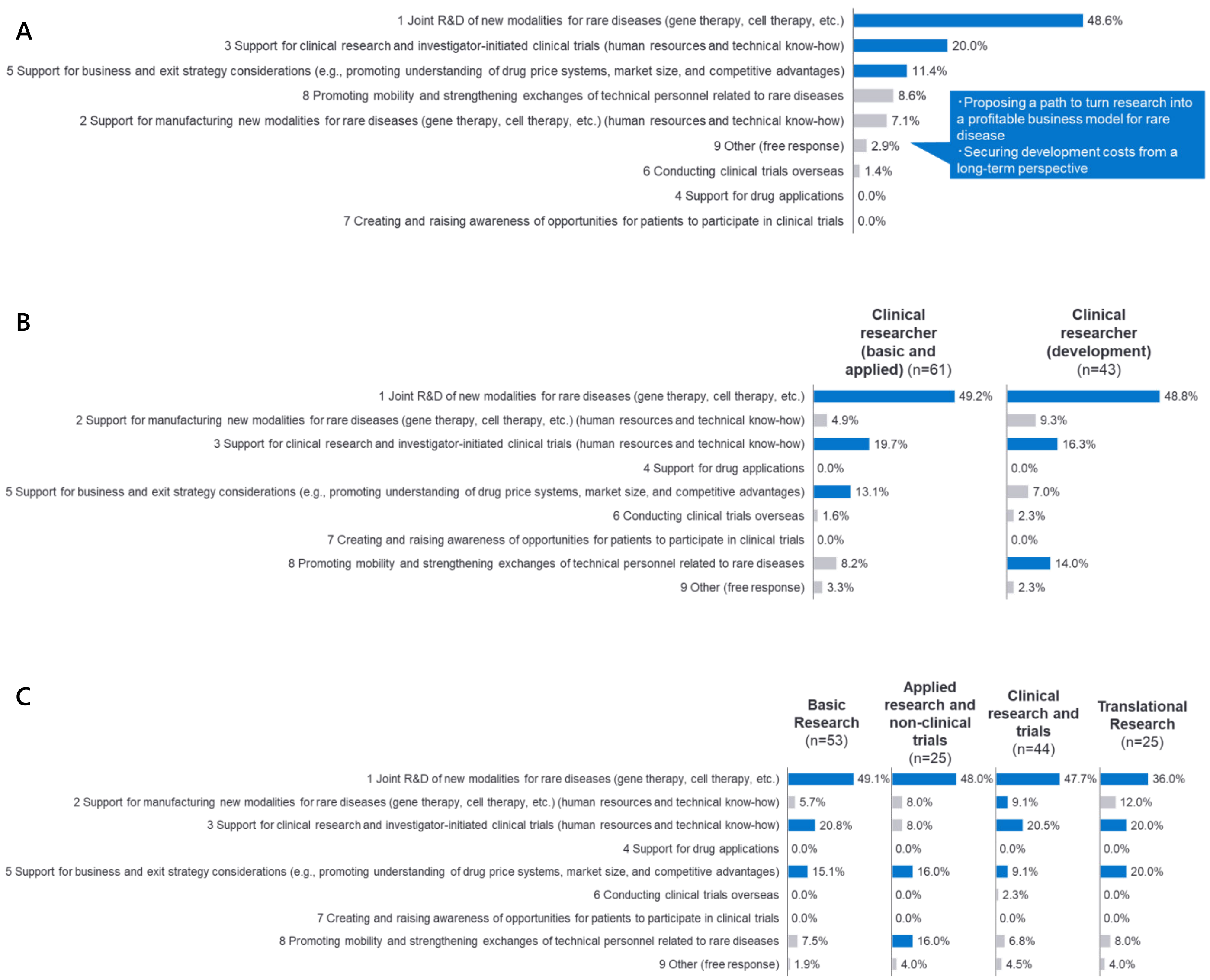
■Survey: Web survey

■Question: Based on the challenges you have answered so far regarding clinical practice, please select the top 5 that you agree are the way things should be (ranked)

■Subjects: 316 specialists, non-specialists, and other HCPs (genetic counselors and nurses)

4.2.2 Expectations for the pharmaceutical industry

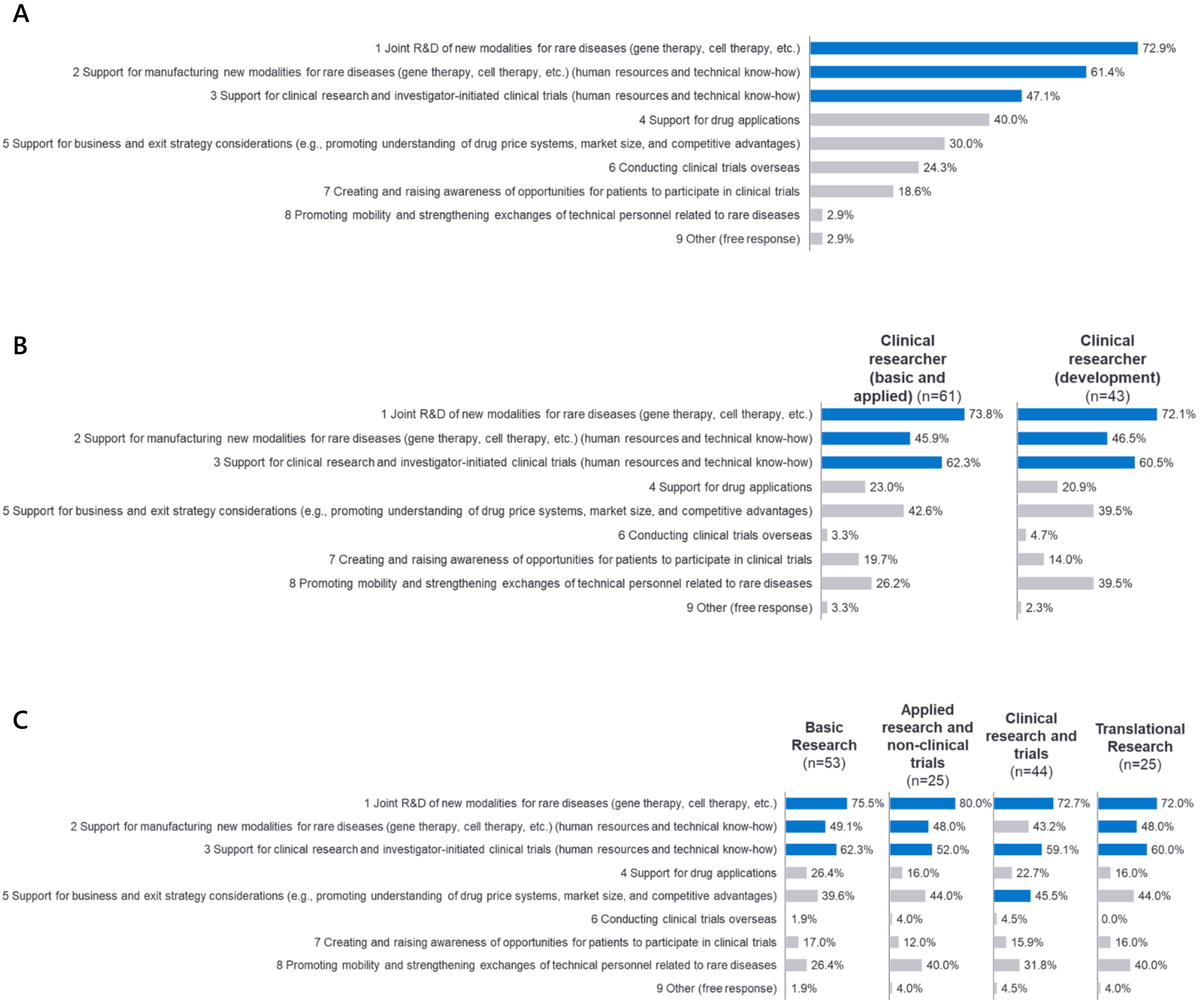
Figure 4.2.2-1: Expectations for the pharmaceutical industry in research and development – Top selection result：A all segments ・B by occupation ・C by specialty



・Proposing a path to turn research into a profitable business model for rare disease
・Securing development costs from a long-term perspective

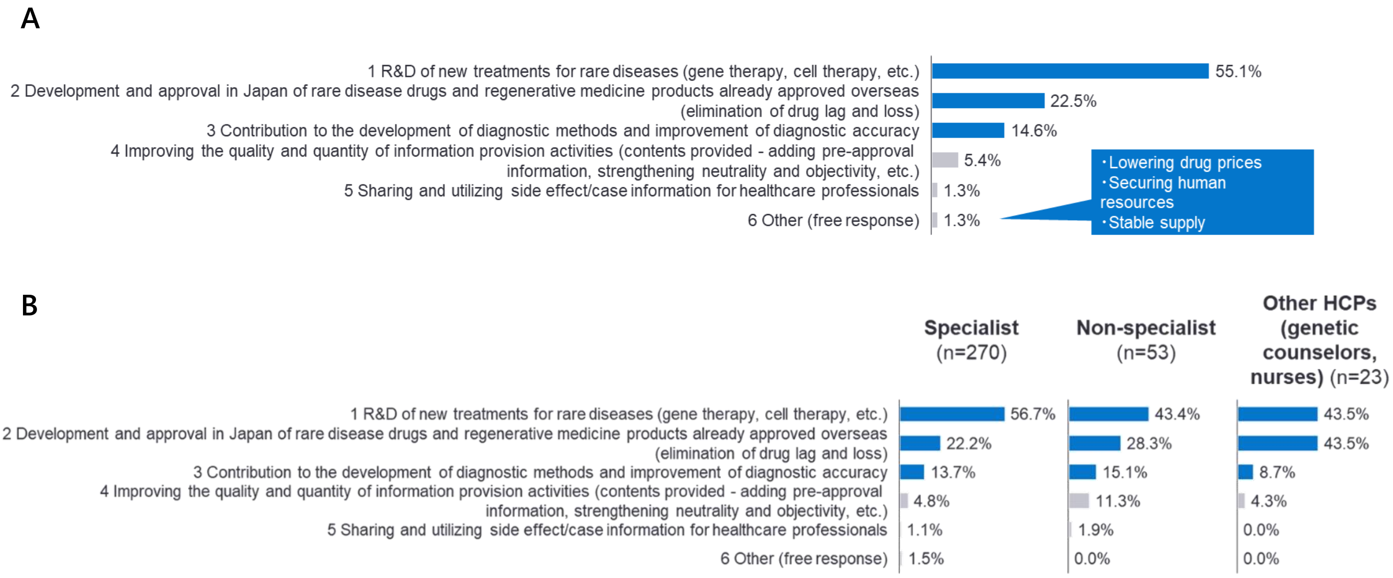
■Survey: Web survey
■Question: Please list your top three expectations for the pharmaceutical industry to achieve the ideal state (ranked)
■Subjects: 70 clinical researchers (basic and applied) and clinical researchers (development)

Figure 4.2.2-2: Expectations for the pharmaceutical industry in research and development – Top 3 selection results：
A all segments ・B by occupation ・C by specialty



■Survey: Web survey
■Question: Please list your top three expectations for the pharmaceutical industry to achieve the ideal state (ranked)
■Subjects: 70 clinical researchers (basic and applied) and clinical researchers (development)

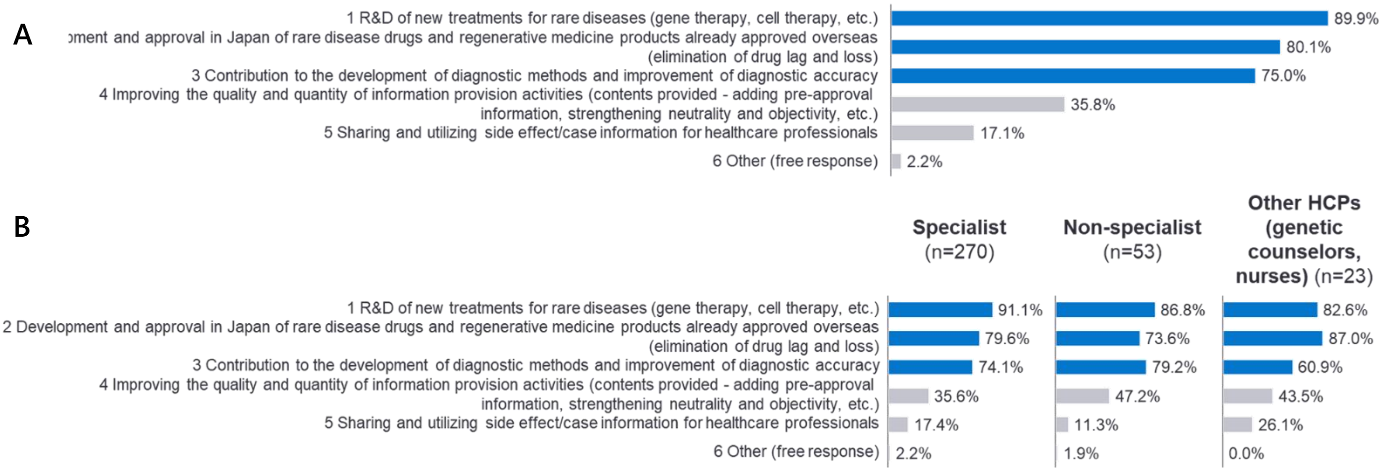
Figure 4.2.2-3: Expectations for the pharmaceutical industry in clinical practice – Top selection result：A all segments ･B by occupation



・Lowering drug prices
・Securing human resources
・Stable supply

■Survey: Web survey
■Question: Please list your top three expectations for the pharmaceutical industry to achieve the ideal state (ranked)
■Subjects: 316 specialists, non-specialists, and other HCPs (genetic counselors and nurses)

Figure 4.2.2-4: Expectations for the pharmaceutical industry in clinical practice – Top 3 selection result：A all segments ･B by occupation



■Survey: Web survey
■Question: Please list your top three expectations for the pharmaceutical industry to achieve the ideal state (ranked)
■Subjects: 316 specialists, non-specialists, and other HCPs (genetic counselors and nurses)

We hope to be able to communicate clinical trial information to patients and healthcare professionals in an easy-to-understand manner, and to ***develop new drugs****.*
(Specialist / Pediatrics)

In the past, when I recommended testing for a patient who may have a genetic disease, the patient told me, 'I don't want to get tested if there is no treatment,' so I have high hopes for pharmaceutical companies to ***develop treatments and reduce drug waste****. I think that knowing that treatments exist will encourage patients to go to the hospital and face their disease.*
(Non-specialist / Neurology)

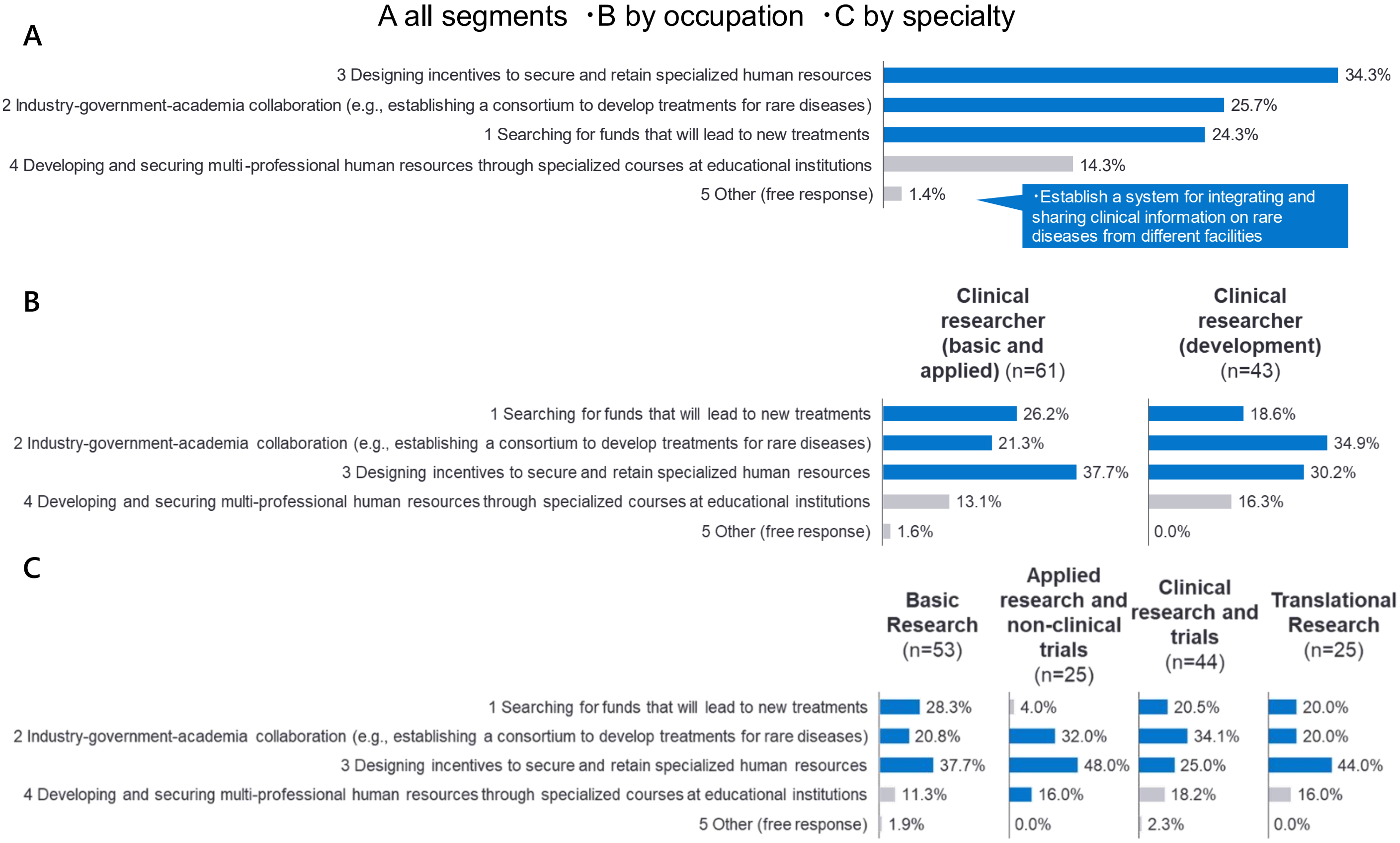
There are university institutions that conduct research like that conducted by companies, so I think it would be good if there were more opportunities for joint research between academia and pharmaceutical companies. Also, when academia tries to contact pharmaceutical companies, they don't know the companies' areas of focus or expertise, so they don't know who to contact, and the procedures are complicated, so ***I would like companies to disclose information about themselves to academia and clarify who to contact****.*
(Non-specialist / Pediatrics)

I hope to create collaborative opportunities with academia and build closer ties. I believe that an environment in which academia, pharmaceutical companies, and patient groups can naturally interact daily, such as by ***sending researchers from pharmaceutical companies to academia or creating joint research spaces****, will become a very important ecosystem for the development of new drugs.*
(Clinical researcher (basic and applied) / neuromuscular disease)

I would like to see more efforts put into ***drug discovery and testing/diagnosis accuracy improvement for rare diseases****. Specifically, I would like to see the progress of rare disease research made visible, with continued updates and improved accessibility, a system built for the accumulation of genetic analysis data in Japan, and the development of domestically produced drugs.*
(Clinical researcher (basic and applied) / neuromuscular disease)

4.2.3 Expectations for academic societies

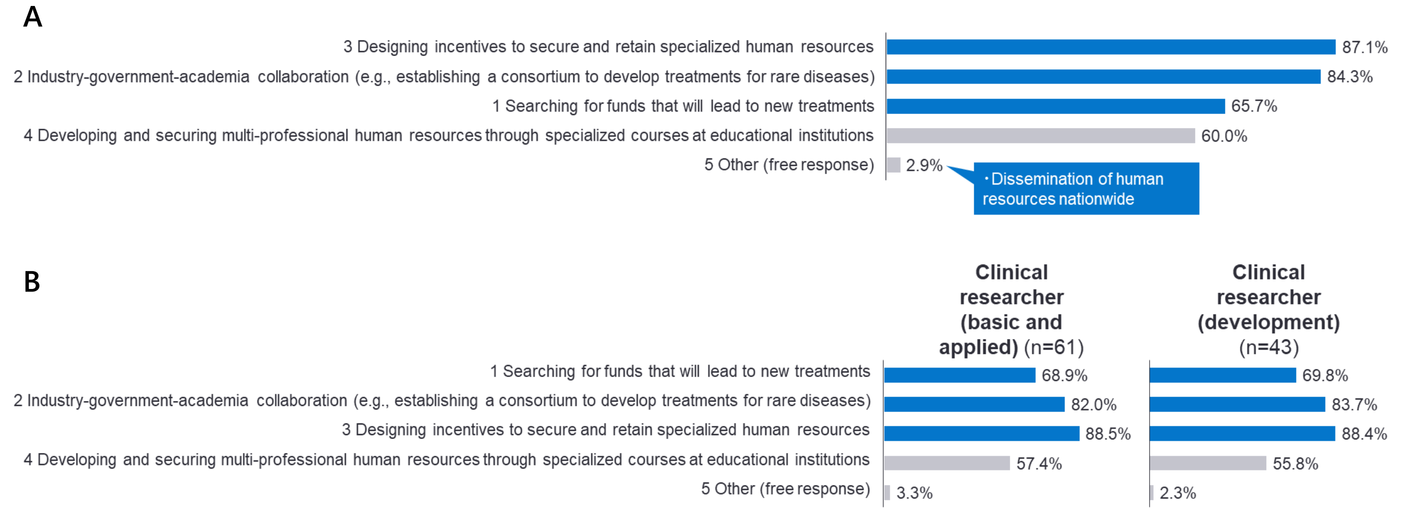
Figure 4.2.3-1: Expectations for academic societies in research and development – Top selection result：
A all segments ・B by occupation ・C by specialty



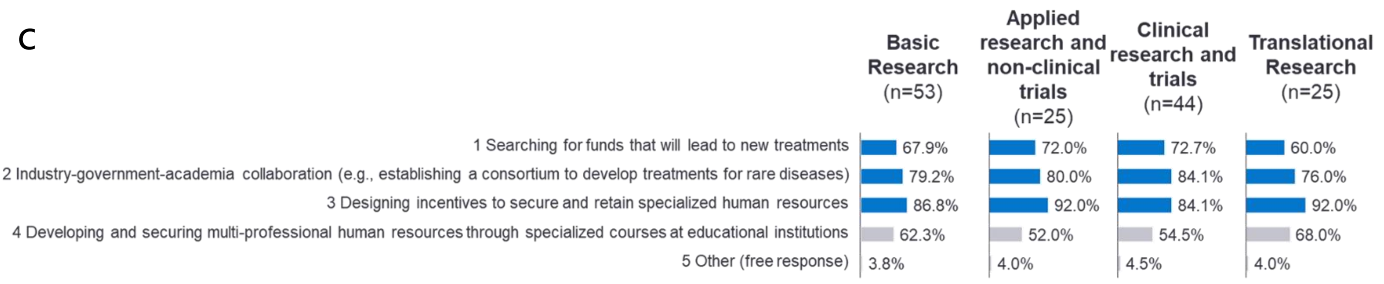
・Establish a system for integrating and sharing clinical information on rare diseases from different facilities

■Survey: Web survey
■Question: Please list your top three expectations of the academic society to realize your ideal state (ranked)
■Subjects: 70 clinical researchers (basic and applied) and clinical researchers (development)

Figure 4.2.3-2: Expectations for academic societies in research and development – Top 3 selection result：
A all segments ・B by occupation ・C by specialty



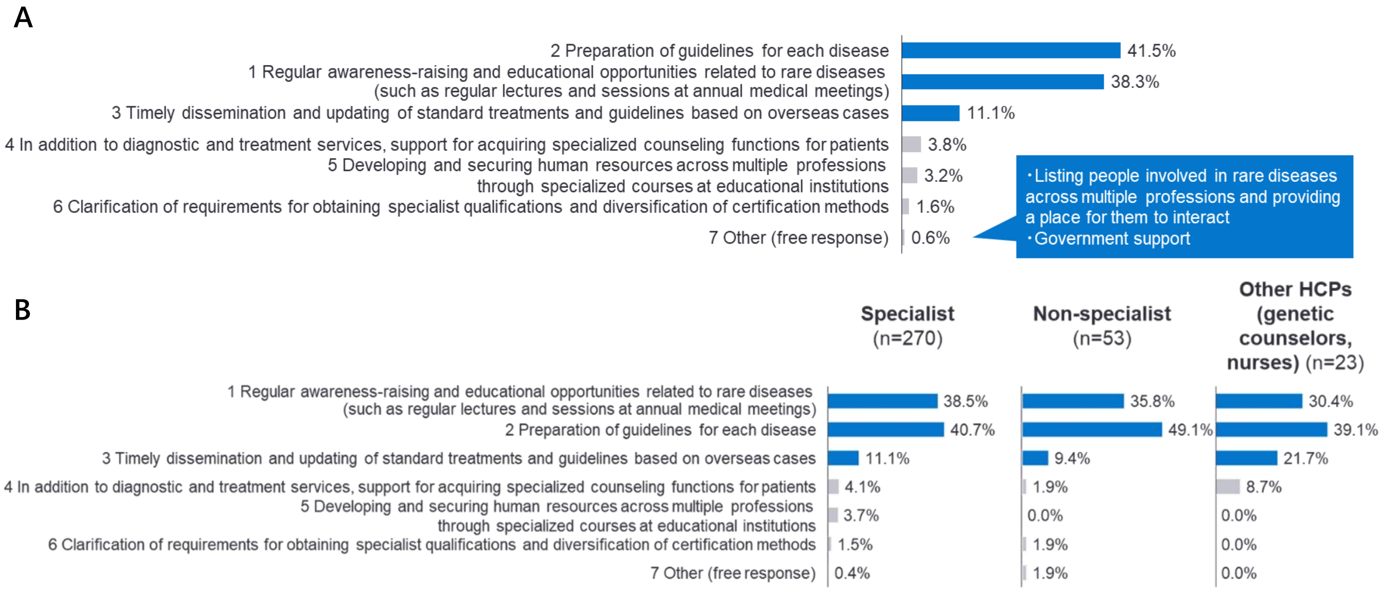
・Dissemination of human resources nationwide



■Survey: Web survey
■Question: Please list your top three expectations of the academic society to realize your ideal state (ranked)
■Subjects: 70 clinical researchers (basic and applied) and clinical researchers (development)

Figure 4.2.3-3: Expectations for academic societies in clinical practice – Top selection result：

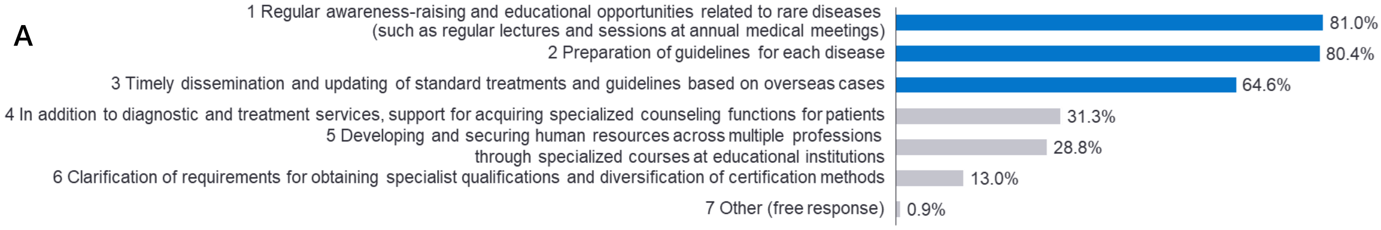
A all segments ・B by occupation

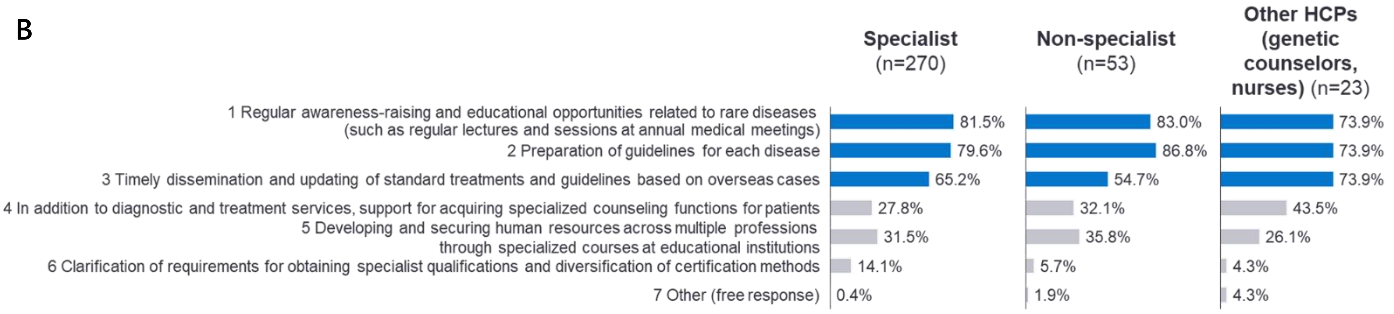


■Survey: Web survey
■Question: Please list top three expectations of the Society to realize the ideal state (ranked)
■Subjects: 316 specialists, non-specialists, and other HCPs (genetic counselors and nurses)

Figure 4.2.3-4: Expectations for academic societies in clinical practice – Top 3 selection result：

A all segments ・B by occupation





- ■Survey: Web survey
- ■Question: Please list top three expectations of the Society to realize the ideal state (ranked)
- ■Subjects: 316 specialists, non-specialists, and other HCPs (genetic counselors and nurses)

“ *Many of the patients who come to our hospital are referred from other hospitals, but some doctors at other hospitals are unsure of which patients they can refer to their hospital, so we feel it is necessary to strengthen awareness of the referral criteria to each facility. We should* ***clarify which facilities have what kind of expertise and the referral criteria to specialized facilities for each disease and accelerate cooperation between medical institutions****.*
(Specialist / Pediatrics)

“ *It is hoped that the academic society will strengthen its presence in rare disease awareness activities for patients and healthcare professionals. In addition, as the number of cases of people moving from universities to pharmaceutical companies is increasing, to* ***create a workplace where specialized human resources can do what they really want and where treatment is guaranteed****, it is necessary to operate a financially independent organization, such as covering research expenses from investment fund profits.*
(Specialist / Collagen Disease)

“ *To create an environment where referral sources can refer patients with peace of mind in a timely manner, we hope to see* ***coordination regarding the mechanism for referrals from non-specialists to specialists****.*
(Non-specialist / Neurology)

“ *I would like them to collect and disseminate accurate information regarding treatment and diagnostic needs.*
(Non-specialist / Pediatrics)

“ ***I want the government to convey voices that individual patients cannot convey*** *as a unified academic opinion backed by academia. I believe that academic societies can complement the correctness and volume of individual patients' voices. The Internet has improved access to information, but it has also led to the spread of incorrect information and different interpretations. Therefore, I want academic societies to* ***clearly communicate what information is correct and what is incorrect.***
(Clinical researcher (development) / All other hereditary disease)

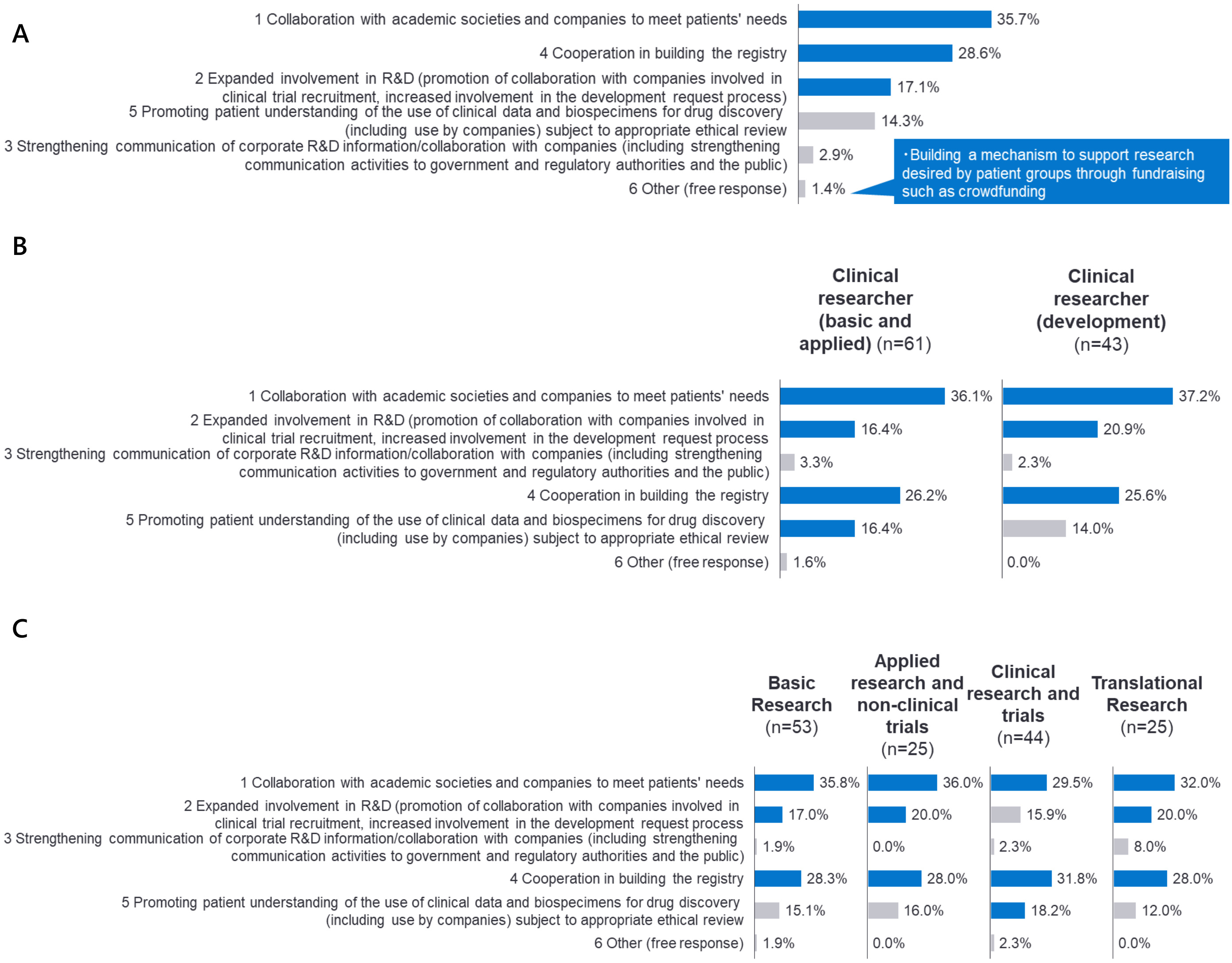
“ *From the perspective of human resource development, it can take four to five years to obtain specialist certification within an academic society, and since young people who are interested in rare diseases are particularly valuable, a more flexible system design, such as a grading system, may be necessary to broaden the base of expertise.*

(Clinical researcher (basic and applied) / Other hereditary disease)

4.2.4 Expectations for patient advocacy groups

Figure 4.2.4-1: Expectations for academic societies in clinical practice – Top selection result：

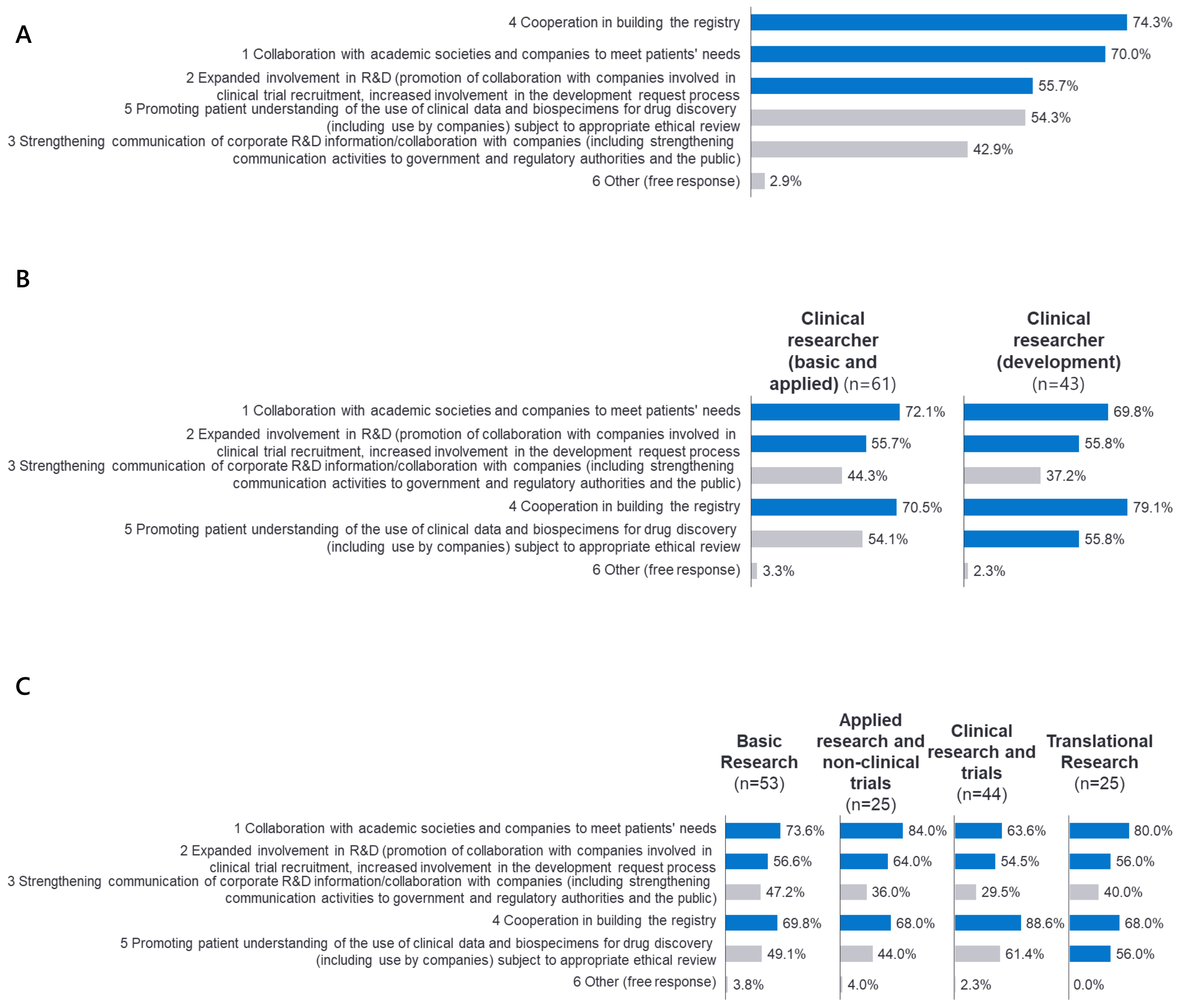
A all segments ・B by occupation ・C by specialty



- ■Survey: Web survey
- ■Question: Please list top three expectations of patient organizations to achieve your vision (ranked)
- ■Subjects: 70 clinical researchers (basic and applied) and clinical researchers (development)

Figure 4.2.4-2: Expectations for academic societies in clinical practice – Top 3 selection result：

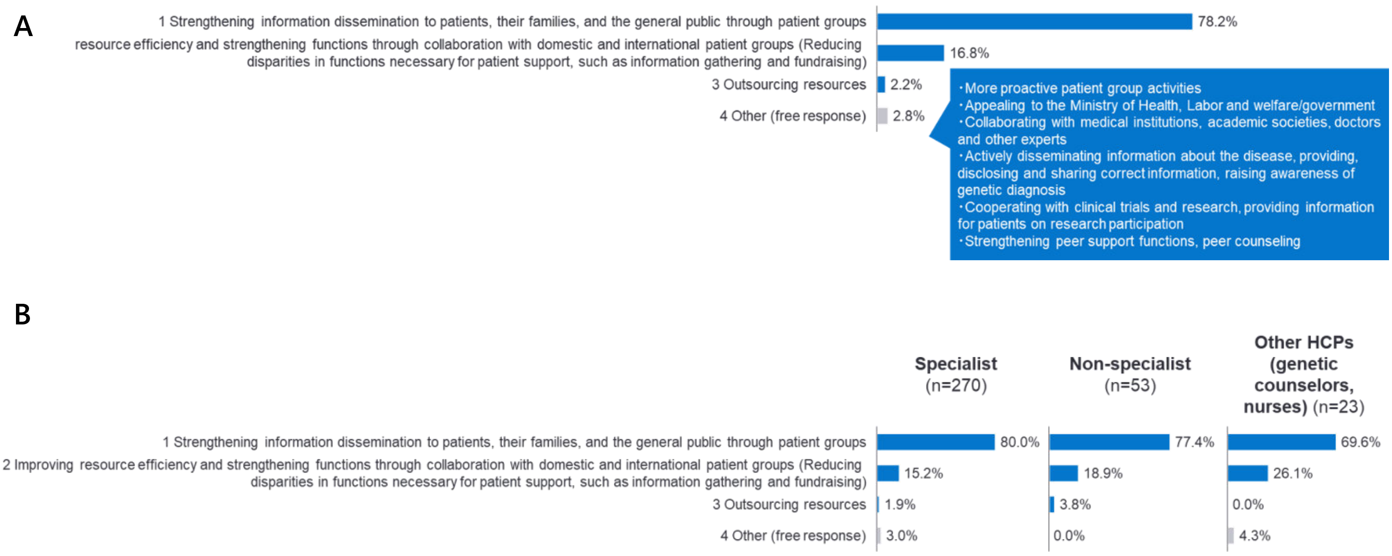
A all segments ・B by occupation ・C by specialty



■Survey: Web survey
■Question: Please list top three expectations of patient organizations to achieve your vision (ranked)
■Subjects: 70 clinical researchers (basic and applied) and clinical researchers (development)

Figure 4.2.4-3: Expectations for patient groups in clinical practice – Top selection result：

A all segments ・B by occupation

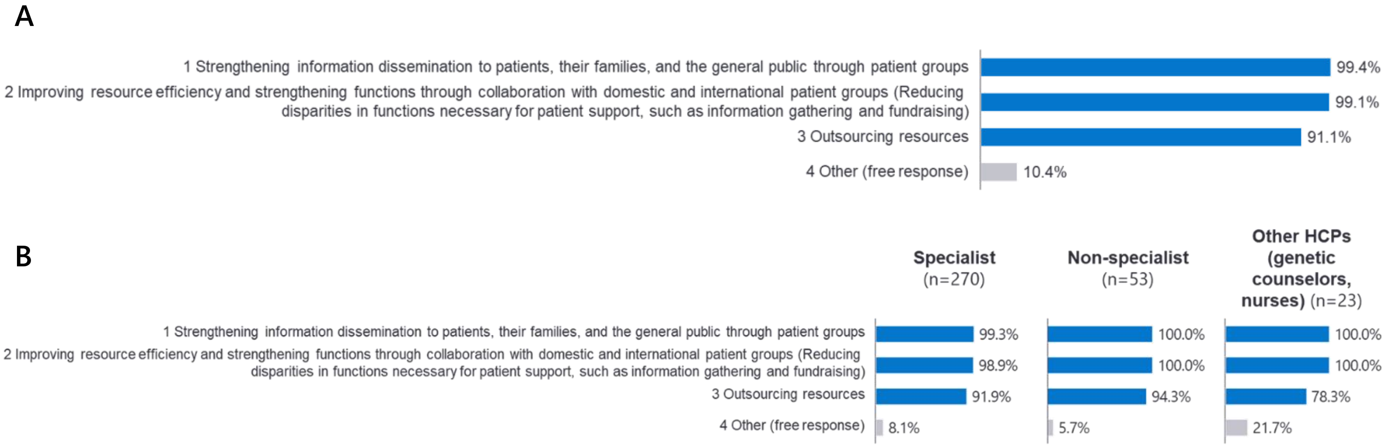


・More proactive patient group activities
・Appealing to the Ministry of Health, Labor and welfare/government
・Collaborating with medical institutions, academic societies, doctors and other experts
・Actively disseminating information about the disease, providing, disclosing and sharing correct information, raising awareness of genetic diagnosis
・Cooperating with clinical trials and research, providing information for patients on research participation
・Strengthening peer support functions, peer counseling

■Survey: Web survey
■Question: Please list top three expectations of patient organizations to achieve your vision (ranked)
■Subjects: 316 specialists, non-specialists, and other HCPs (genetic counselors and nurses)

Figure 4.2.4-4: Expectations for patient groups in clinical practice – Top 3 selection result：

A all segments ・B by occupation



■Survey: Web survey
■Question: Please list top three expectations of patient organizations to achieve your vision (ranked)
■Subjects: 316 specialists, non-specialists, and other HCPs (genetic counselors and nurses)

“ *Currently, the activities of each patient group vary, but we would like patient groups to* ***improve peer support for patients and their families who cannot be supported by HCPs****, regardless of the disease.*
(Specialist / Pediatrics)

“ *I would like them to* ***collect and disseminate information*** *about patients' needs regarding medical expenses, such as raising disease awareness and lowering drug prices.*
(Specialist / Collagen Disease)

“ *We believe that the existence/activities of patient groups have a major impact after a definitive diagnosis, so we hope that they will provide support to patients in how to* ***deal with the disease and in their daily lives****, mainly in prognosis management.*
(Non-specialist / Neurology)

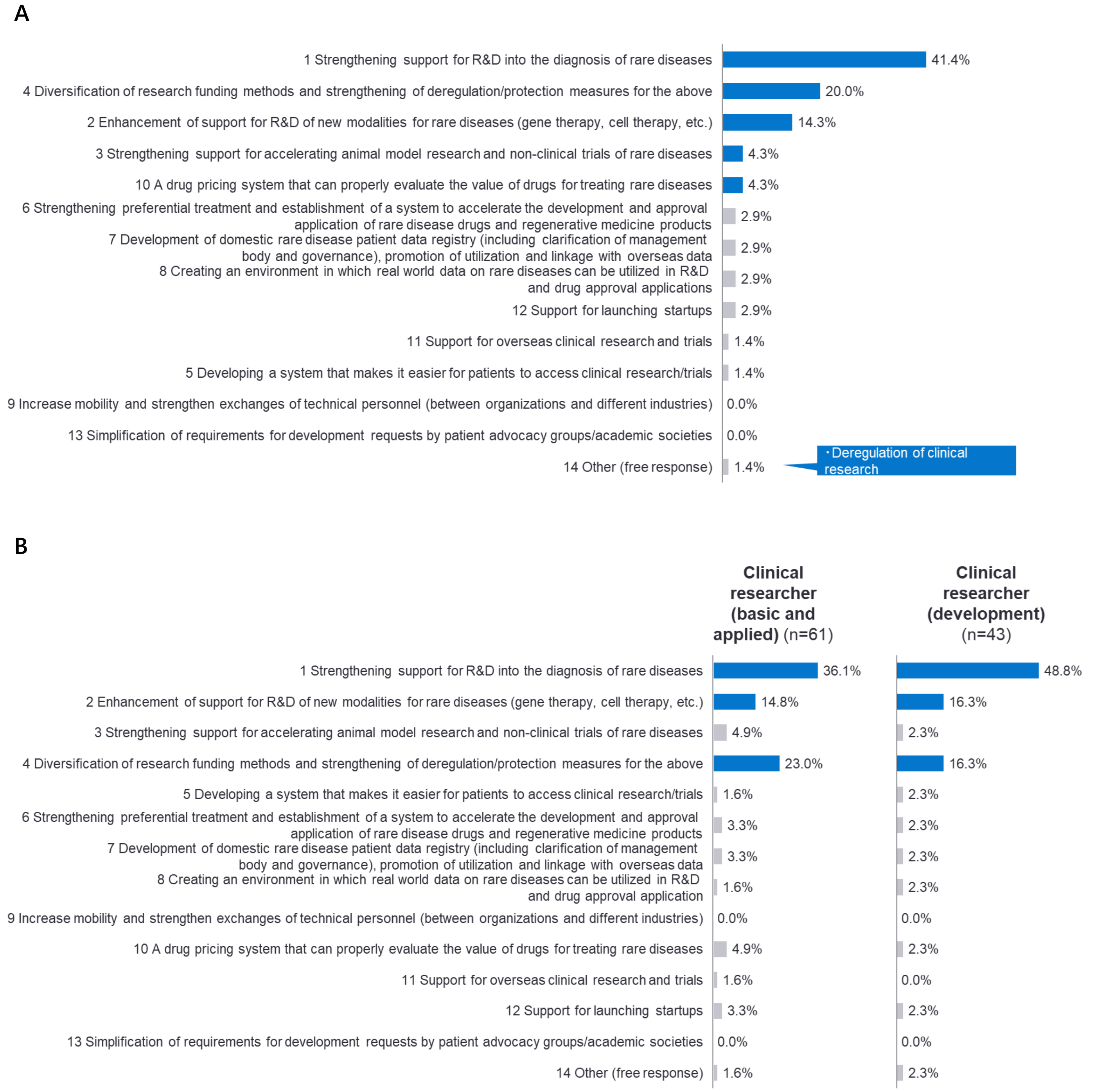
“ *I want them to* ***improve their fundraising ability and medical literacy*** *to have a voice and initiative. Patient groups in Japan do not have the means to mobilize people, such as funds, so they inevitably have a weak voice. If* ***patient groups can provide funds for research and development****, they should be able to have a voice and initiative, just like patient groups in the West, where fundraising activities such as charities are active.*
(Basic and applied research doctor / neuromuscular disease)

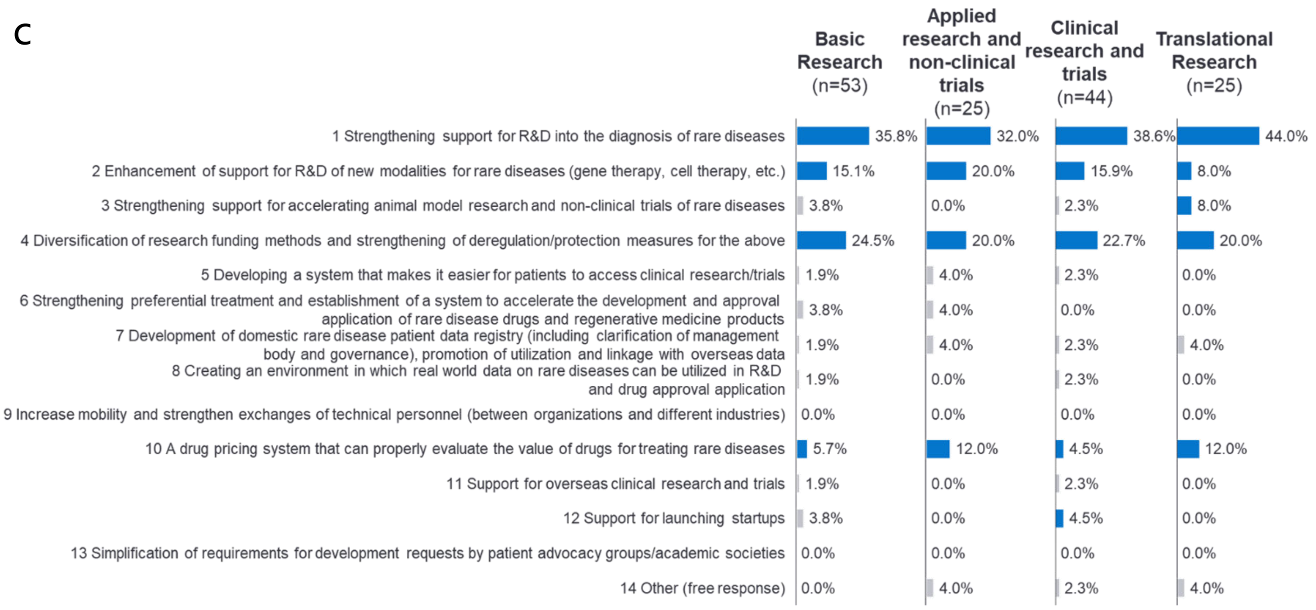
“ *I would like them to* ***strengthen their activities to make themselves known****, such as media exposure, activities in the field of education, and crowdfunding. I think that by involving more stakeholders, creating contact points, and deepening mutual understanding, their activities will accelerate.*
(Clinical researcher (basic and applied) / Other hereditary disease)

“ *As a patient advocacy group, I would like you to be* ***actively involved in research and development and clinical activities****. For example, I feel that the current registry registration is not in a state where it is easy for patients to register, and since there is no foundation for patient data, further development is not possible, so I would like patient advocacy groups to actively cooperate.*
(Clinical researcher (development) / All other hereditary disease)

4.2.5 Expectations for government and regulatory authorities

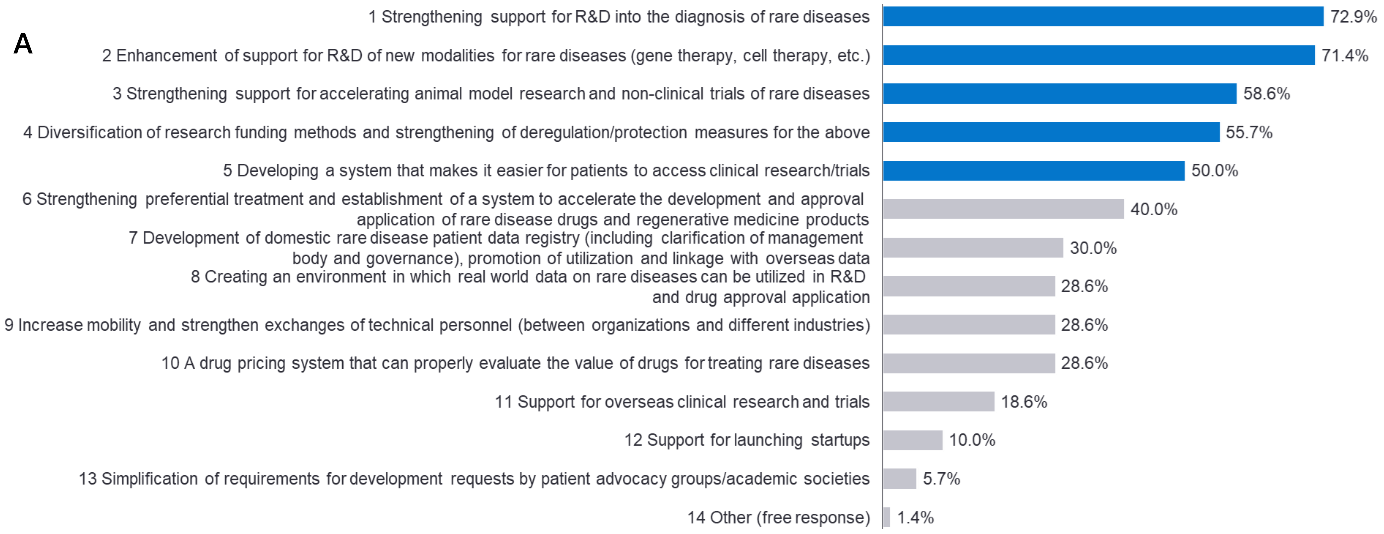
Figure 4.2.5-1: Expectations for administrative and regulatory authorities in research and development – Top selection result：
A all segments ・B by occupation ・C by specialty

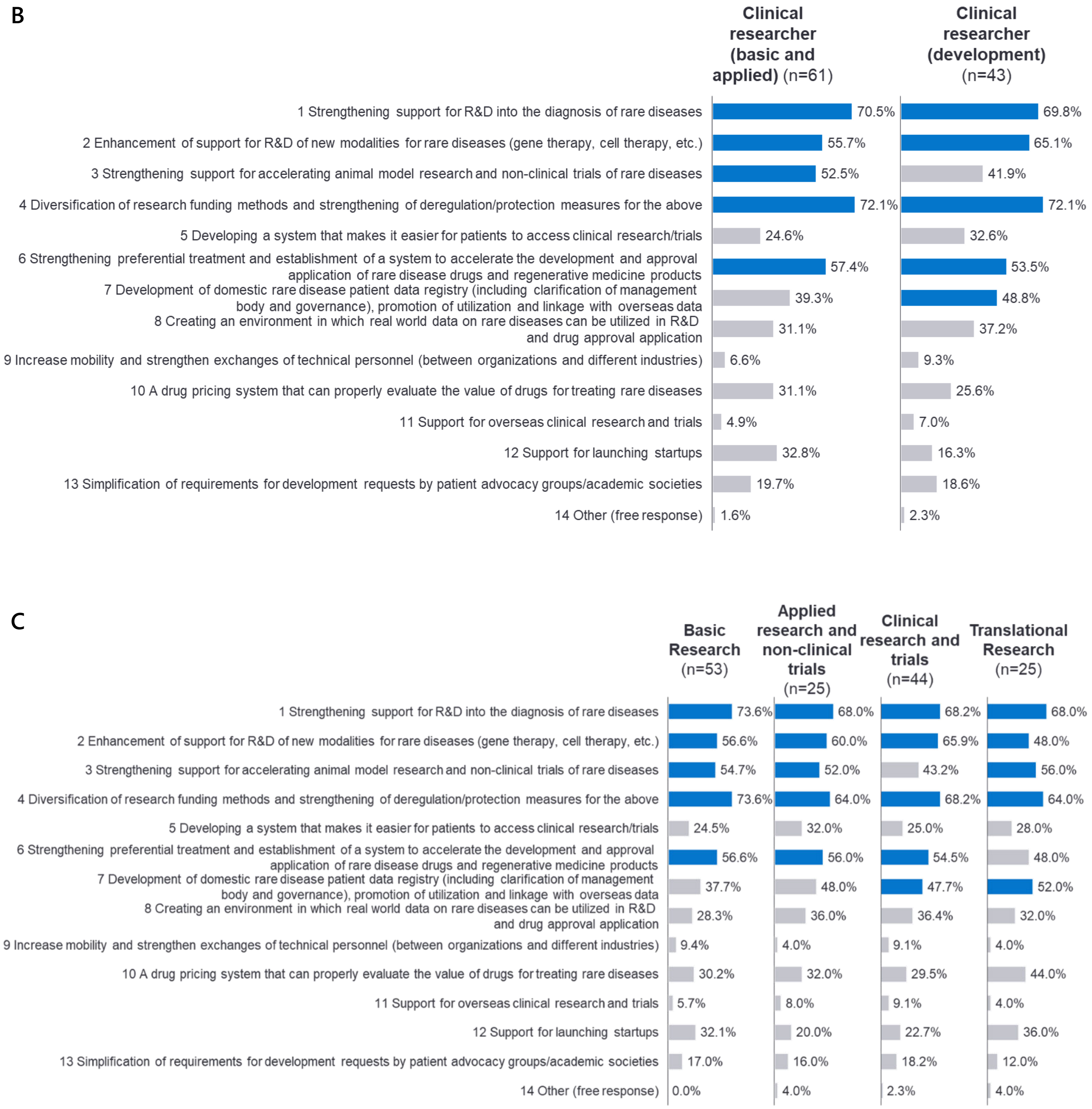




■Survey: Web survey
■Question: Please select top five expectations of government and regulatory authorities to achieve the ideal state (ranking format)
■Subjects: 70 clinical researchers (basic and applied) and clinical researchers (development)

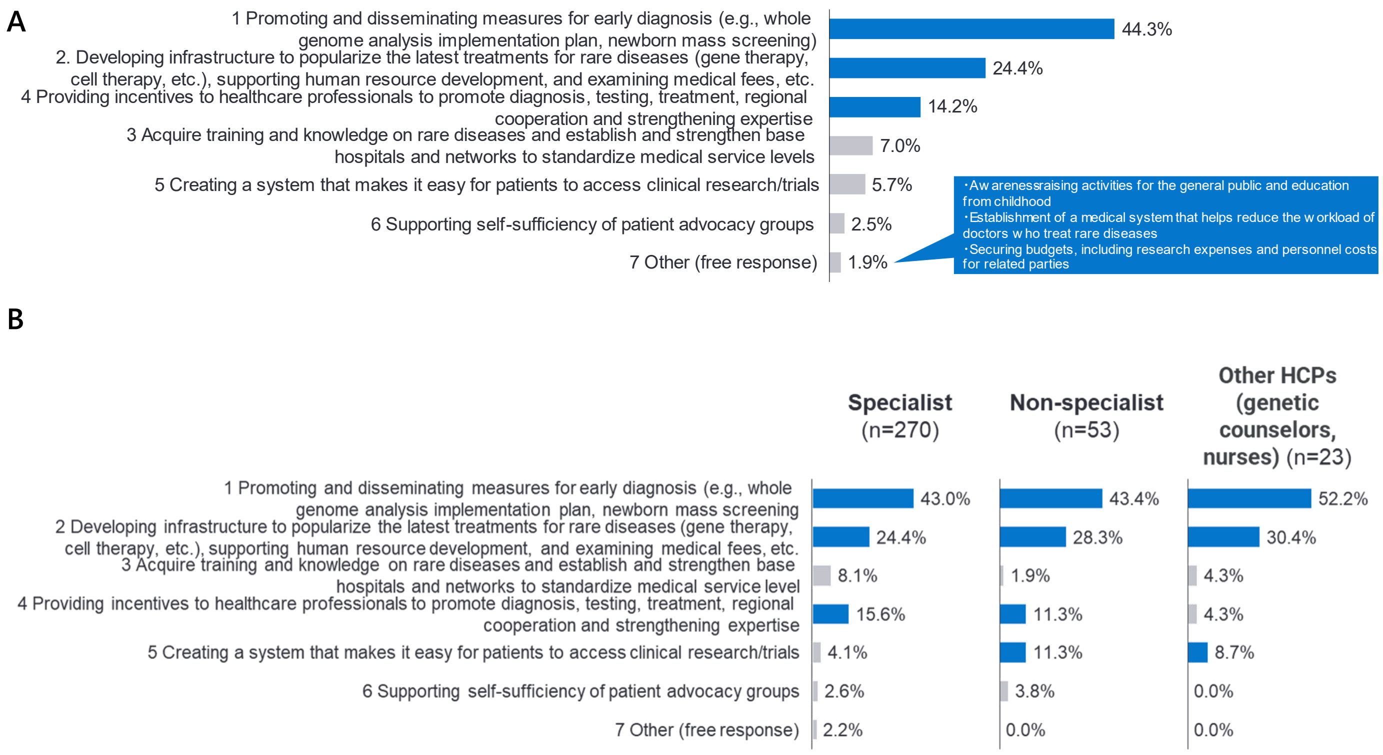
Figure 4.2.5-2: Expectations for administrative and regulatory authorities in research and development – Top 3 selection result：
A all segments ・B by occupation ・C by specialty





■Survey: Web survey
■Question: Please select top five expectations of government and regulatory authorities to achieve the ideal state (ranking format)
■Subjects: 70 clinical researchers (basic and applied) and clinical researchers (development)

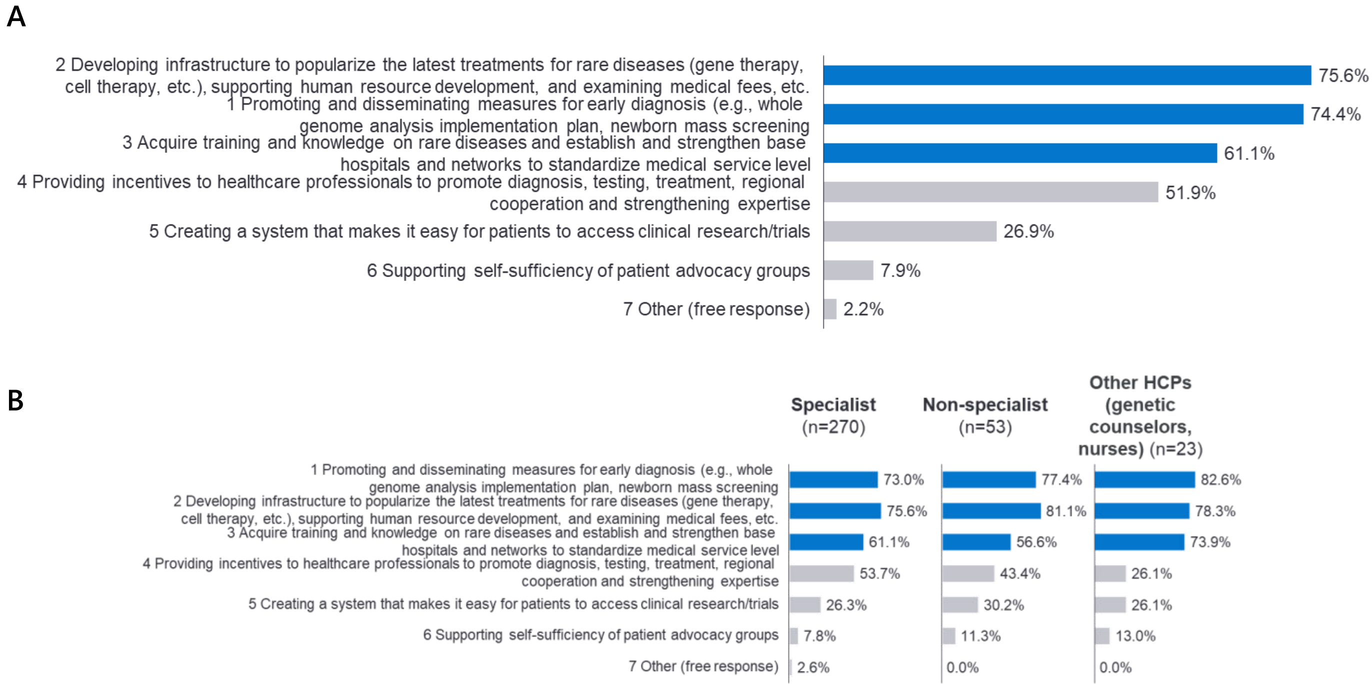
Figure 4.2.5-3: Expectations for government and regulatory authorities in clinical practice – Top selection result：A all segments ・B by occupation



・Awarenessraising activities for the general public and education from childhood
・Establishment of a medical system that helps reduce the workload of doctors who treat rare diseases
・Securing budgets, including research expenses and personnel costs for related parties

■Survey: Web survey
■Question: Please list top three expectations of government and regulatory authorities to achieve your ideal state (ranked)
■Subjects: 316 specialists, non-specialists, and other HCPs (genetic counselors and nurses)

Figure 4.2.5-4: Expectations for government and regulatory authorities in clinical practice – Top 3 selection result：A all segments ・B by occupation



■Survey: Web survey
■Question: Please list top three expectations of government and regulatory authorities to achieve your ideal state (ranked)
■Subjects: 316 specialists, non-specialists, and other HCPs (genetic counselors and nurses)

“ *In clinical trials, information about clinical trials is not fully communicated to subjects, making it extremely difficult to recruit subjects who meet the conditions. If* ***clinical trial information were centrally collected*** *and there was a system that allowed users to narrow down clinical trial information and subject information that meets the conditions, it would be convenient for both healthcare professionals and subjects.*
(Specialist / Pediatrics)

“ *I hope that income restrictions for patients who are eligible for subsidies (such as subsidies for families raising children with disabilities) will be lifted. Also, among the registered designated intractable diseases, there are diseases that qualify as designated intractable diseases based on the disease name alone, and diseases that are determined based on the disease name and severity, so in the latter case, there are patients who do not qualify as designated intractable diseases even though they need to go to the hospital regularly. I feel that there is currently a lack of support for such patients, so I would like to see* ***the number of designated intractable diseases eligible for subsidies expanded****.*
(Specialist / Pediatrics)

“ *To resolve the shortage of human resources at medical institutions, we need to see* ***incentives designed within academia*** *through personnel evaluation and rules on part-time work, such as securing the necessary budget for hiring and training full-time personnel and making it easier to startup ventures on campus.*
(Specialist / Collagen Disease)

“ *I would like to see deregulation of fundraising for each stakeholder involved in research and development, and the establishment of a* ***drug pricing system that makes it easier for companies to recoup their investments****. Rather than focusing on a set system, I would like to see ideas about how to achieve goals and how the government can be involved in helping to achieve those goals.*
(Clinical researcher (basic and applied) / neuromuscular disease)

“ *I hope that we can work to create a society in which ‘patients can access the information and medicines they need’ and ‘patients and their families can live the same lives as healthy people’ through a* ***significant increase in budgets and personnel, relaxation of the pharmaceutical industry's restrictions on the provision of information****, and a review of how genetic and diversity education is taught in primary education.*
(Clinical researcher (development) / All other hereditary disease)

“ *I hope to see deregulation that will make it easier for industry-government-academia collaboration to proceed, genetic education for younger generations to help create a* ***society free of prejudice and friction****, and the establishment of mechanisms and systems that will allow people in rural areas to receive healthcare such as testing and examinations for rare diseases.*
(Other HCPs (Genetic counselors and nurses) / Department of Clinical Genetics and Gene Therapy)

“ *Unless society increases its tolerance for failure, pharmaceutical companies will not be able to take on new* *challenges. Since these are medicines for Japanese people, we need* ***deregulation to accelerate drug discovery*** *and encourage foreign capital restrictions and a return to Japan, so that domestic pharmaceutical companies can invest in medicines that accurately meet domestic needs.*

(Clinical researcher (basic and applied) / All other hereditary disease)

Regarding use of this publication

- To maintain neutrality, this report uses survey data conducted by IRUD, RDCJ, and the Japan Pharmaceutical Manufacturers Association (JPMA) on behalf of a third-party organization (EY Strategy & Consulting Co., Ltd.)
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