

PATIENT PRIORITIES

PATIENTS AND CAREGIVERS SETTING THE AGENDA

Jaishri Blakeley, MD

Johns Hopkins Comprehensive
Neurofibromatosis Center

SETTING THE STAGE

As part of the 2015 International Children's Tumor Foundation Meeting, closed the meeting with clinical and basic scientists with expertise within each syndromes (NF1, NF2, SWN) reviewing:

- Results from a survey of patients and caregivers through the CTF patient registry
- Literature review of publications about NF1, NF2, SWN in the last year
- Review of abstracts at the 2015 NF Conference

CTF PATIENT/FAMILY SURVEY

- Survey Monkey Questionnaire created and reviewed by panel:
 - Asking questions re: symptoms of greatest concern and priorities for research to the patients and caregivers participating in the CTF registry
- Sent to those enrolled in CTF NF Registry
 - NF1, NF2, Schwannomatosis groups each sent a unique questionnaire

NF1 RESPONDERS

Age		
Answer Options	Response Percent	Response Count
17 or younger	35.3%	205
18-20	1.4%	8
21-29	11.0%	64
30-39	16.7%	97
40-49	12.4%	72
50-59	14.5%	84
60 or older	8.8%	51

Sex		
Answer Options	Response Percent	Response Count
Female	61.6%	358
Male	38.4%	223

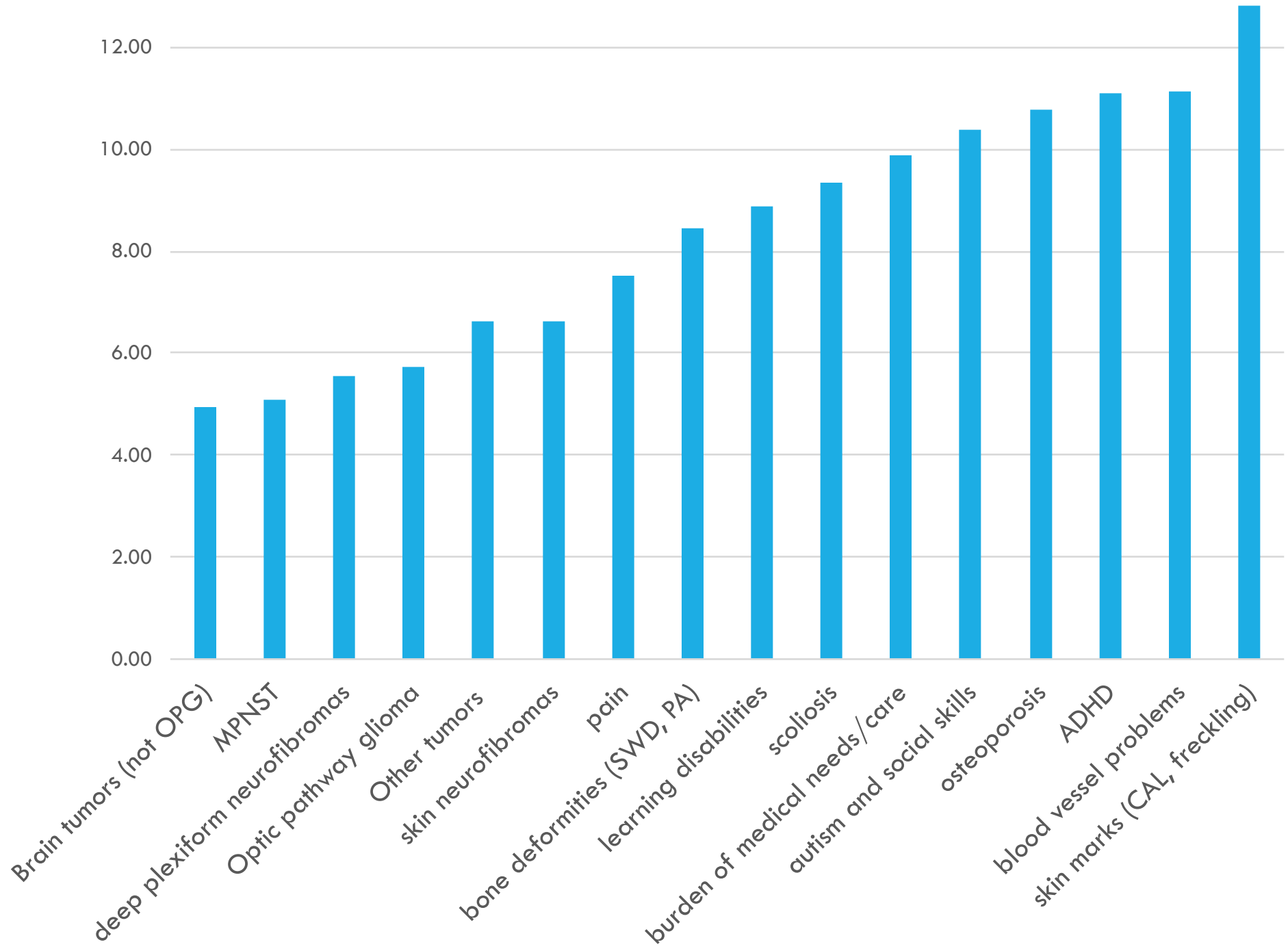
Location		
Australia	2.9%	17
Brazil	0.7%	4
Canada	6.7%	39
China	0.7%	4
France	0.2%	1
Germany	0.5%	3
India	0.5%	3
Italy	0.5%	3
Japan	0.0%	0
Mexico	0.2%	1
New Zealand	0.7%	4
Russia	1.2%	7
Spain	0.2%	1
United Kingdom	4.5%	26
United States	80.6%	468
Other (please specify)		38

QUESTIONS ASKED

Please rank what you think are the biggest problems for patients with NF1 (1 being the top priority and 16 being the least urgent priority):

Optic pathway glioma
Brain tumors not in the optic pathway
Other tumors (breast cancer, pheochromocytoma, leukemia, GIST)
Skin neurofibromas
Deep (plexiform) neurofibromas
Cancerous nerve tumors (malignant peripheral nerve sheath tumor)
Osteoporosis
Scoliosis
Bone deformities (sphenoid wing dysplasia; tibial pseudarthrosis)
Autism and social skills
Learning disabilities
ADHD
Blood vessel problems
Skin marks (café au lait spot and freckling)
Pain
Burden of medical needs and care

NF1
(N=581)
51% PATIENT
49% PARENT



- Other:
- Itching
 - Anxiety/depression
 - Speech/language

QUESTIONS ASKED

Please rank where you feel that researchers should be spending their time and efforts to help patients with NF1 (1 being the top priority and 11 being the least urgent priority):

Studying cells for clues about the disorder and its manifestations

Studying animal models for clues about the disorder and its

Studying human tissues for clues about the disorder

Researching genetic therapies

Researching drug therapies

Researching patient symptoms and natural history

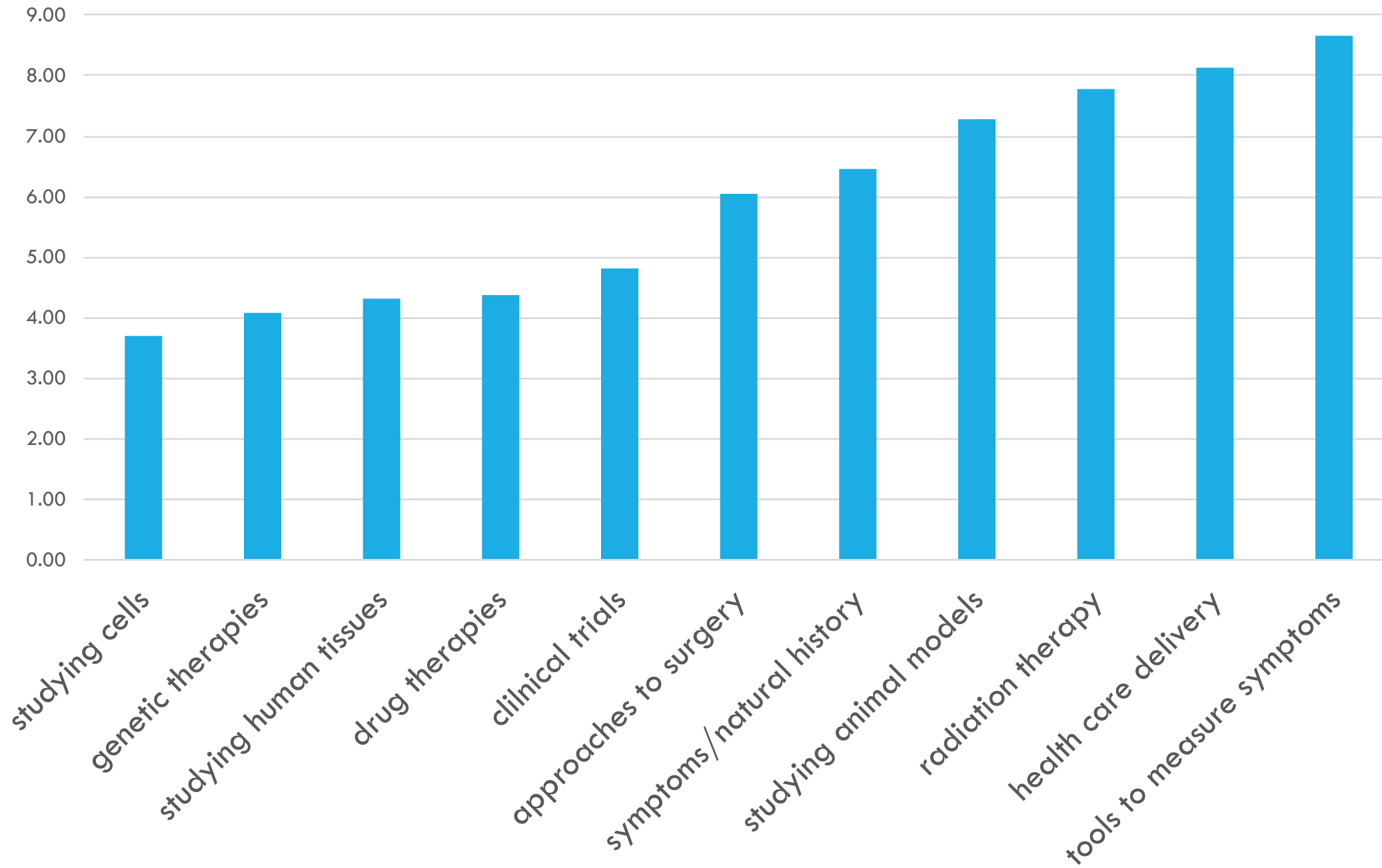
Researching approaches to surgery

Researching radiation therapy

Conducting clinical trials

Researching health care delivery

Researching tools to measure symptoms



NF1
(N=581)
51% patient
49% parent

NF2 RESPONDERS

Age		
17 or younger	10.0%	11
18-20	0.9%	1
21-29	19.1%	21
30-39	28.2%	31
40-49	13.6%	15
50-59	15.5%	17
60 or older	12.7%	14

Sex		
Female	62.7%	69
Male	37.3%	41

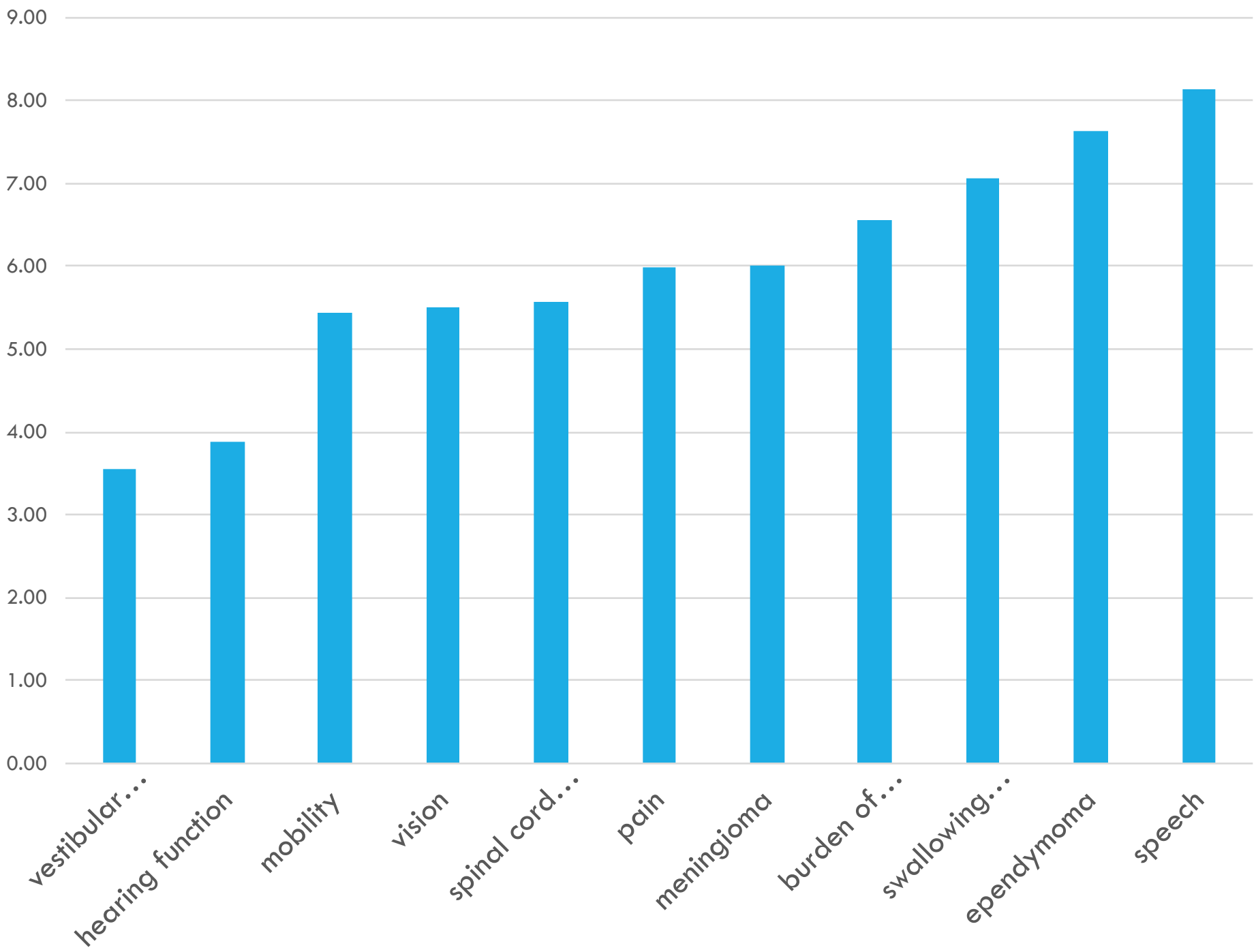
Location		
Australia	4.5%	5
Brazil	2.7%	3
Canada	7.3%	8
China	0.9%	1
France	0.0%	0
Germany	2.7%	3
India	1.8%	2
Italy	0.9%	1
Japan	0.0%	0
Mexico	0.0%	0
New Zealand	0.9%	1
Russia	0.9%	1
Spain	3.6%	4
United Kingdom	7.3%	8
United States	66.4%	73
Other (please specify)		11

QUESTIONS ASKED

In the setting of NF2, rank what you think are the biggest problems for patients (1 being the top priority and 11 being the least urgent priority):

Vestibular schwannomas
Meningioma
Ependymoma
Hearing function
Pain
Spinal cord function
Mobility
Vision
Swallowing function
Speech
Burden of medical needs and care

NF2
(N=110)
80% PATIENT
20% PARENT

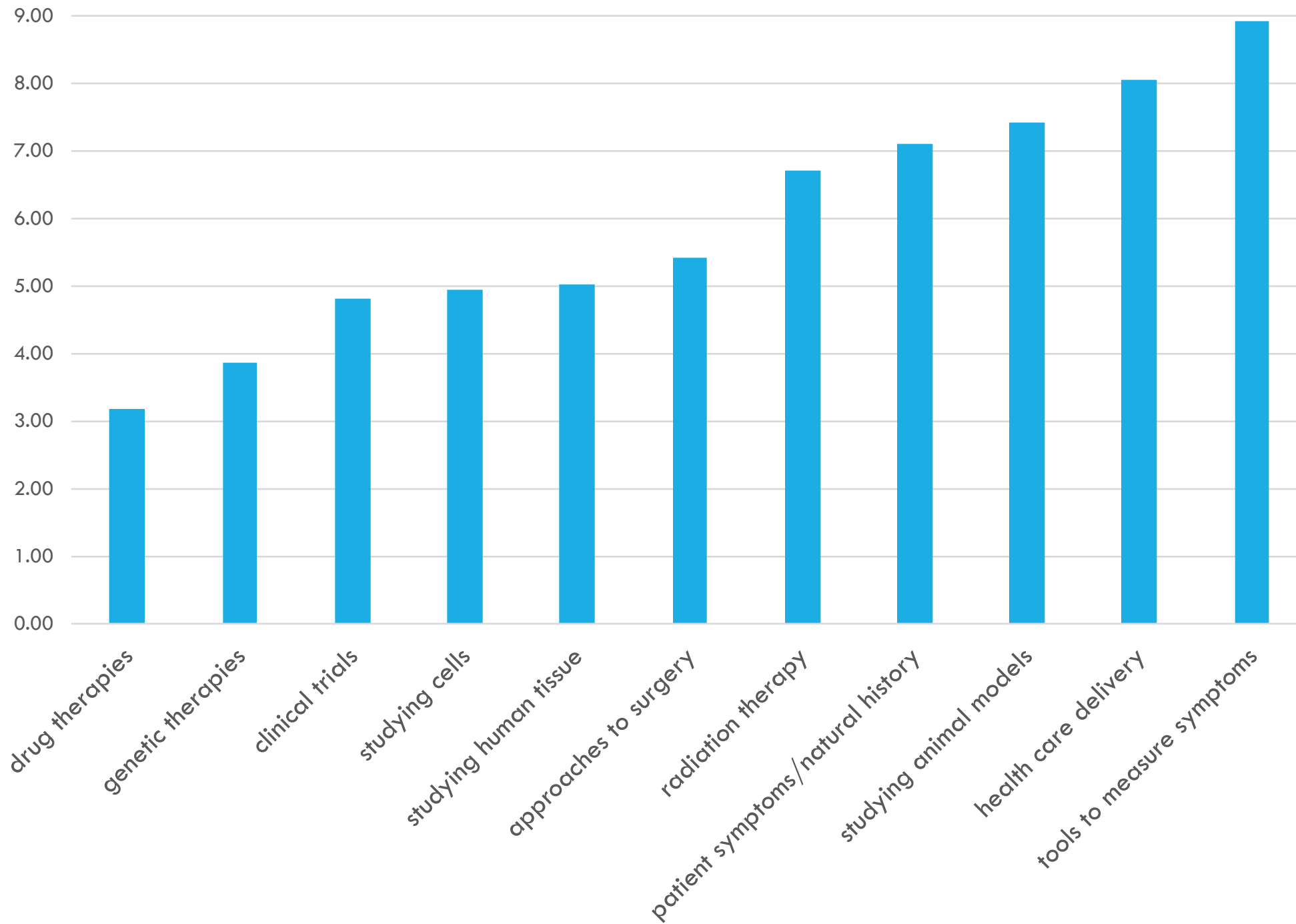


- Other:**
- Balance
 - Depression
 - Facial function/paralysis

QUESTIONS ASKED

Please rank where you feel that researchers should be spending their time and efforts to help patients with NF2 (1 being the top priority and 11 being the least urgent priority):

- Studying cells for clues about the disorder and its manifestations
- Studying animal models for clues about the disorder and its
- Studying human tissues for clues about the disorder
- Researching genetic therapies
- Researching drug therapies
- Researching patient symptoms and natural history
- Researching approaches to surgery
- Researching radiation therapy
- Conducting clinical trials
- Researching health care delivery
- Researching tools to measure symptoms



NF2
(N=110)
80% patient
20% parent

SWN RESPONDERS

Age		
17 or younger	0.0%	0
18-20	0.0%	0
21-29	7.1%	2
30-39	17.9%	5
40-49	35.7%	10
50-59	25.0%	7
60 or older	14.3%	4

Sex		
Female	67.9%	19
Male	32.1%	9

Location		
Australia	0.0%	0
Brazil	0.0%	0
Canada	7.1%	2
China	0.0%	0
France	0.0%	0
Germany	3.6%	1
India	0.0%	0
Italy	0.0%	0
Japan	0.0%	0
Mexico	0.0%	0
New Zealand	0.0%	0
Russia	0.0%	0
Spain	0.0%	0
United Kingdom	10.7%	3
United States	78.6%	22
Other (please specify)		2

QUESTIONS ASKED

In the setting of Schwannomatosis, rank what you think are the biggest problems for patients (1 being the top priority and 4 being the least urgent priority):

Schwannomas

Functional limitations

Pain

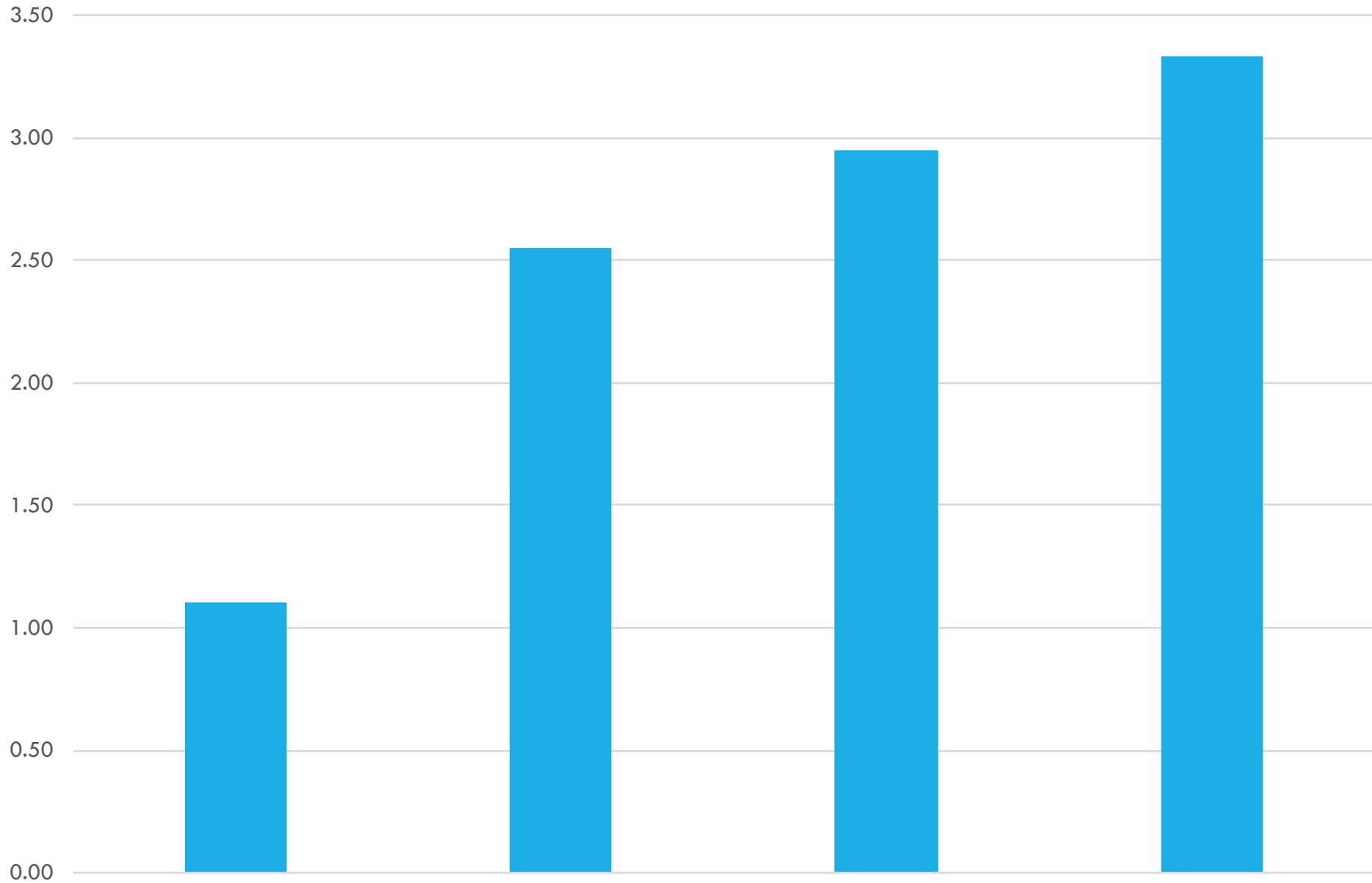
Burden of medical needs and care

SCHWANNOMATOSIS

(N=28)

96% PATIENT

4% PARENT



pain

schwannomas

functional
limitations

burden of
medical care

Other:

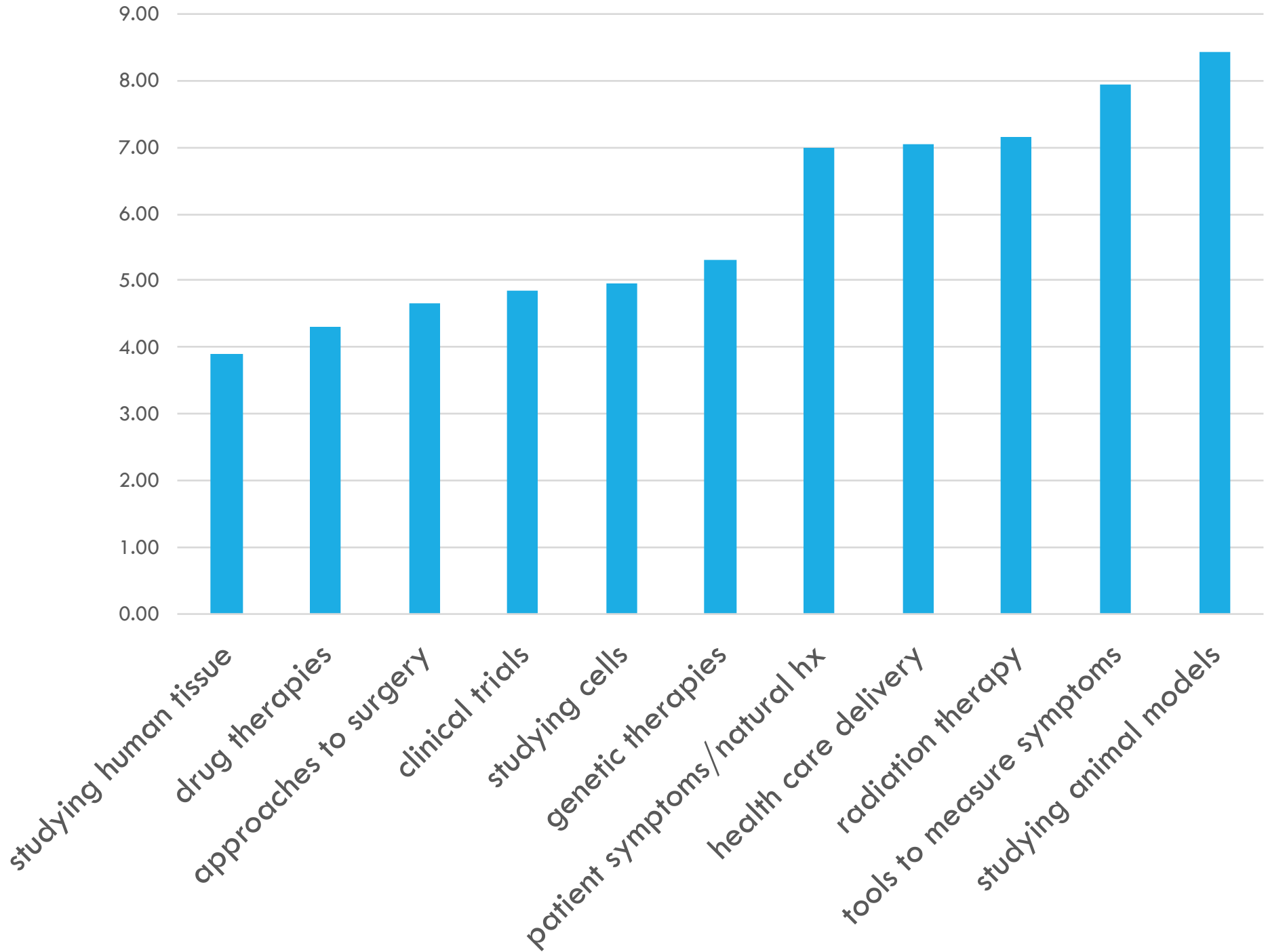
- Awareness/Education

QUESTIONS ASKED

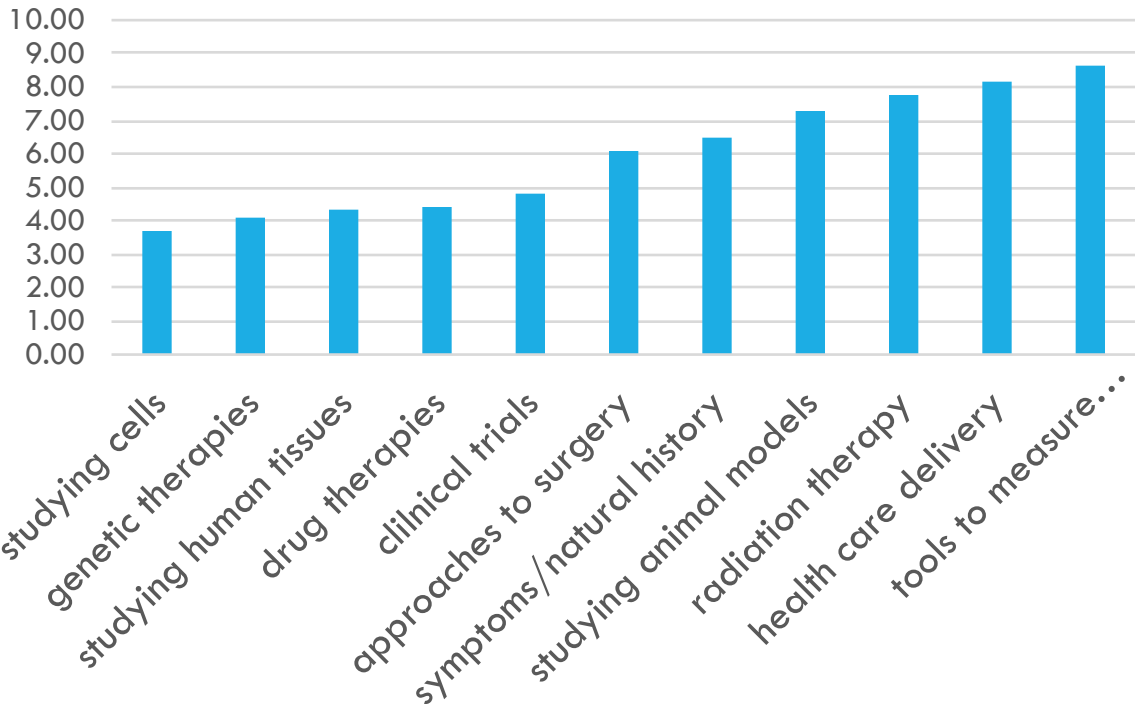
Please rank where you feel that researchers should be spending their time and efforts to help patients with SWN (1 being the top priority and 11 being the least urgent priority):

- Studying cells for clues about the disorder and its manifestations
- Studying animal models for clues about the disorder and its
- Studying human tissues for clues about the disorder
- Researching genetic therapies
- Researching drug therapies
- Researching patient symptoms and natural history
- Researching approaches to surgery
- Researching radiation therapy
- Conducting clinical trials
- Researching health care delivery
- Researching tools to measure symptoms

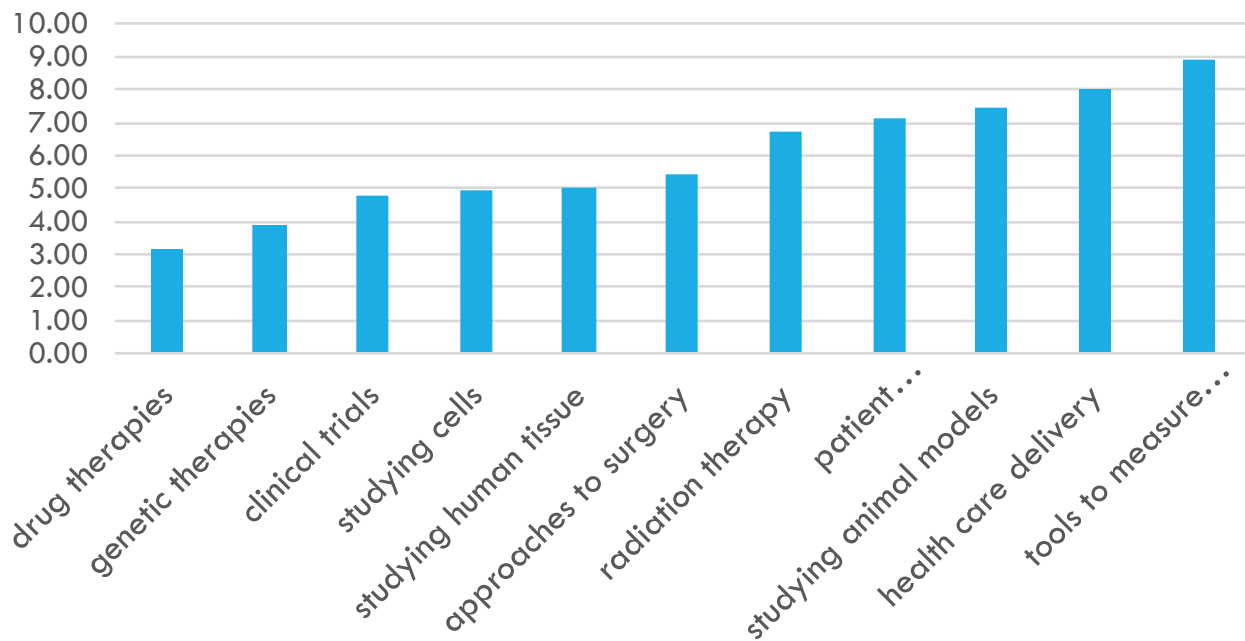
SCHWANNOMATOSIS
(N=28)
96% PATIENT
4% PARENT



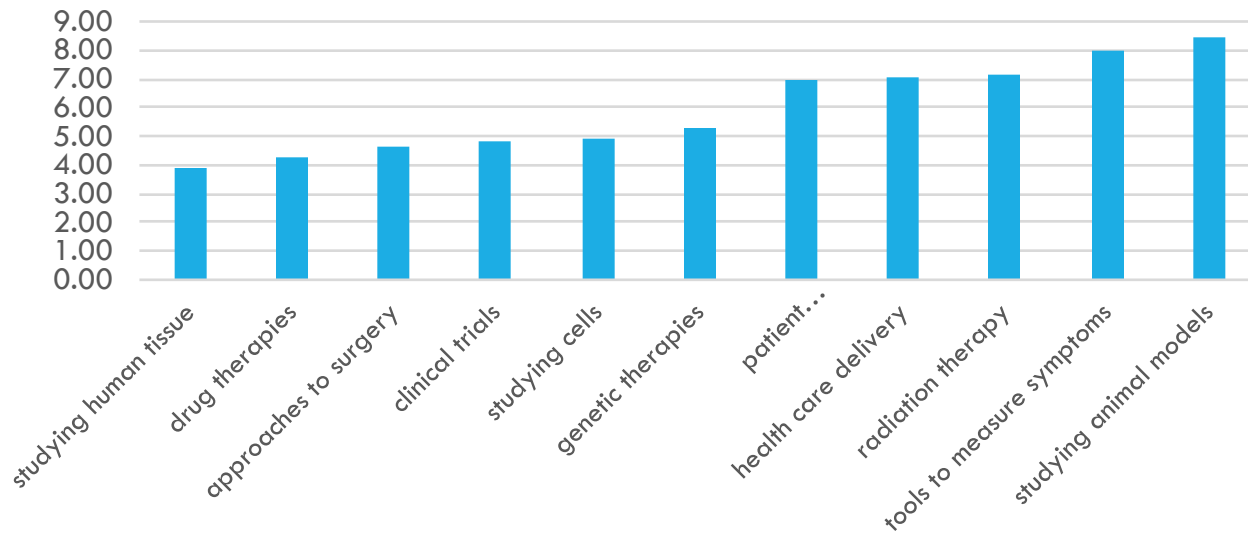
NF1



NF2



SWN



CDMRP AREAS OF EMPHASIS

Health services research

- Evidence-based clinical care pathways
- Innovative healthcare delivery systems
- Utilization of technology and informatics

Heterogeneity of neurofibromas and other NF-related tumors

Non-tumor manifestations

- Sleep, pain, hypotonia, etc. including quality of life measures, self-reported and otherwise

Novel disease and treatment response markers using genomics, epigenetics, systems biology, metabolomics or similar approaches

- Transition from benign to malignant

Nutritional, environmental, and other modifiers of NF

Post-adolescent manifestations

Preclinical efficacy studies

Target identification, drug discovery, targeted and immunotherapies



THANK YOU

Victoria Huang

Pam Knight

David Stevenson

Annette Bakker

Betty Schorry

James Gusella

Larry Sherman

David Gutmann

Gareth Evans

Allan Belzberg