

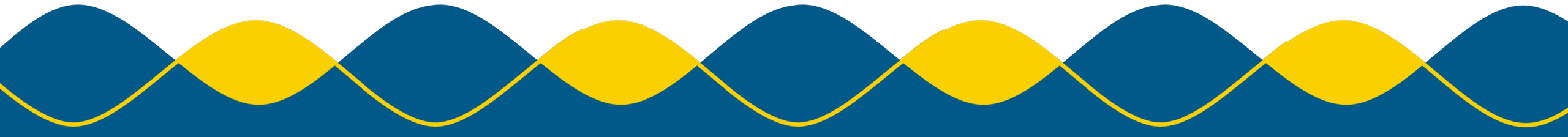
ARRE Foundation Family Survey

February – March 2021



About the survey

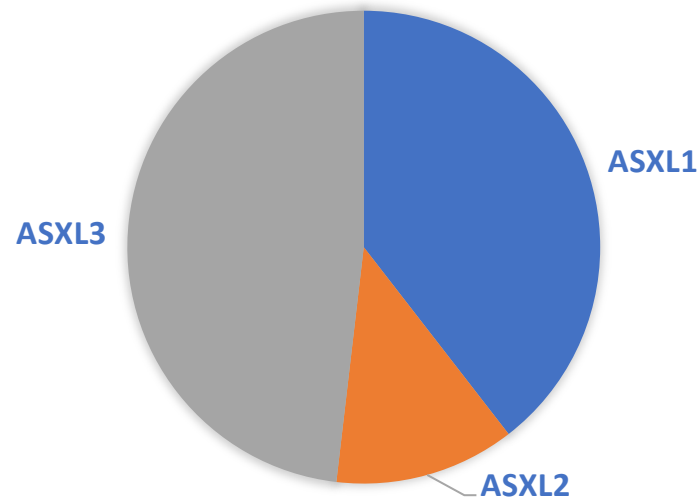
- Intended for families/caregivers only
- Open for two weeks for responses
- Promoted via:
 - ARRE Foundation Facebook page
 - ARRE Foundation email newsletter
 - Posted in each syndrome's private Facebook support group
- Translated into 4 languages
 - English, Spanish, French, German
 - *Additional requests for Italian and Portuguese*



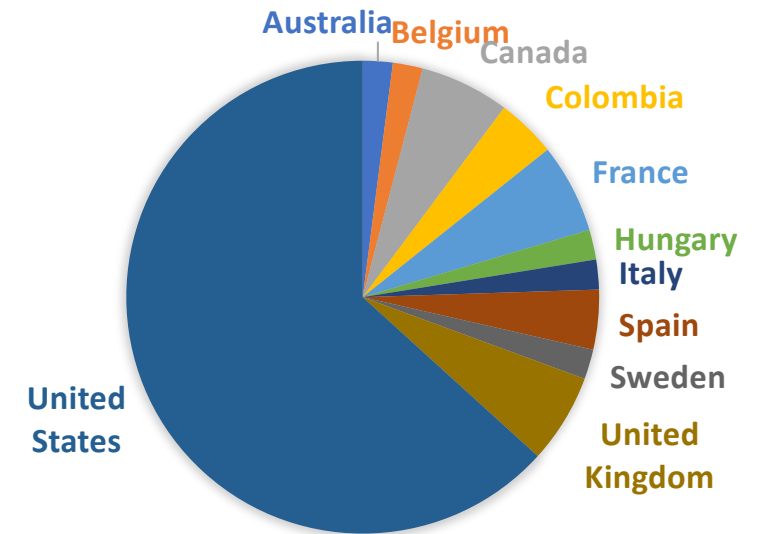
Survey demographics

- 81 survey responses
- Response by syndrome
 - 32 Bohring-Opitz (39.5%)
 - 10 Shashi-Pena (12.3%)
 - 39 Bainbridge-Ropers (48.1%)
- Response from 7 countries

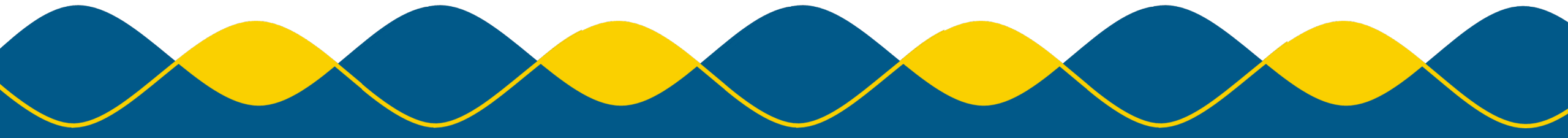
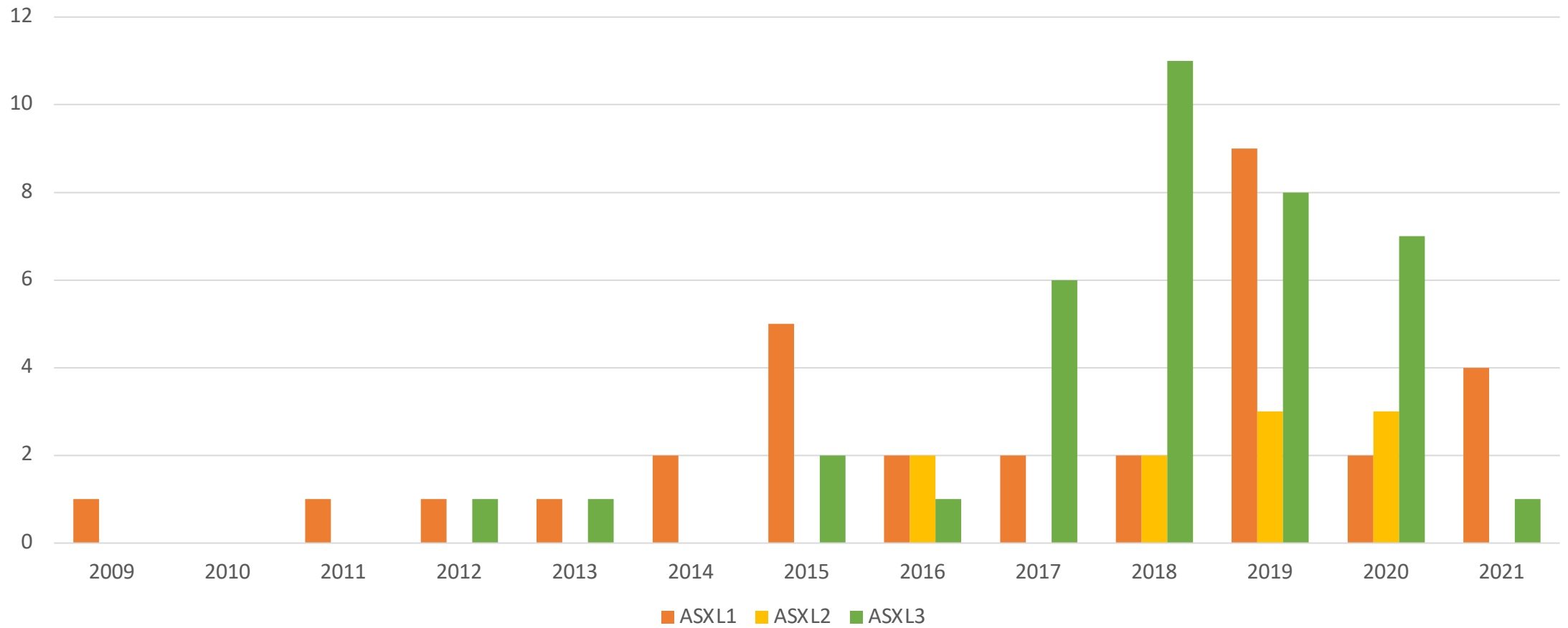
RESPONSE BY SYNDROME



RESPONSE BY COUNTRY



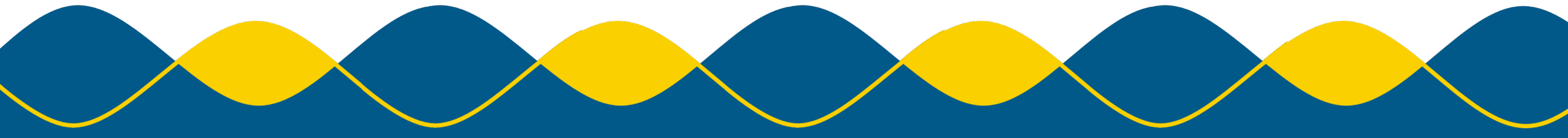
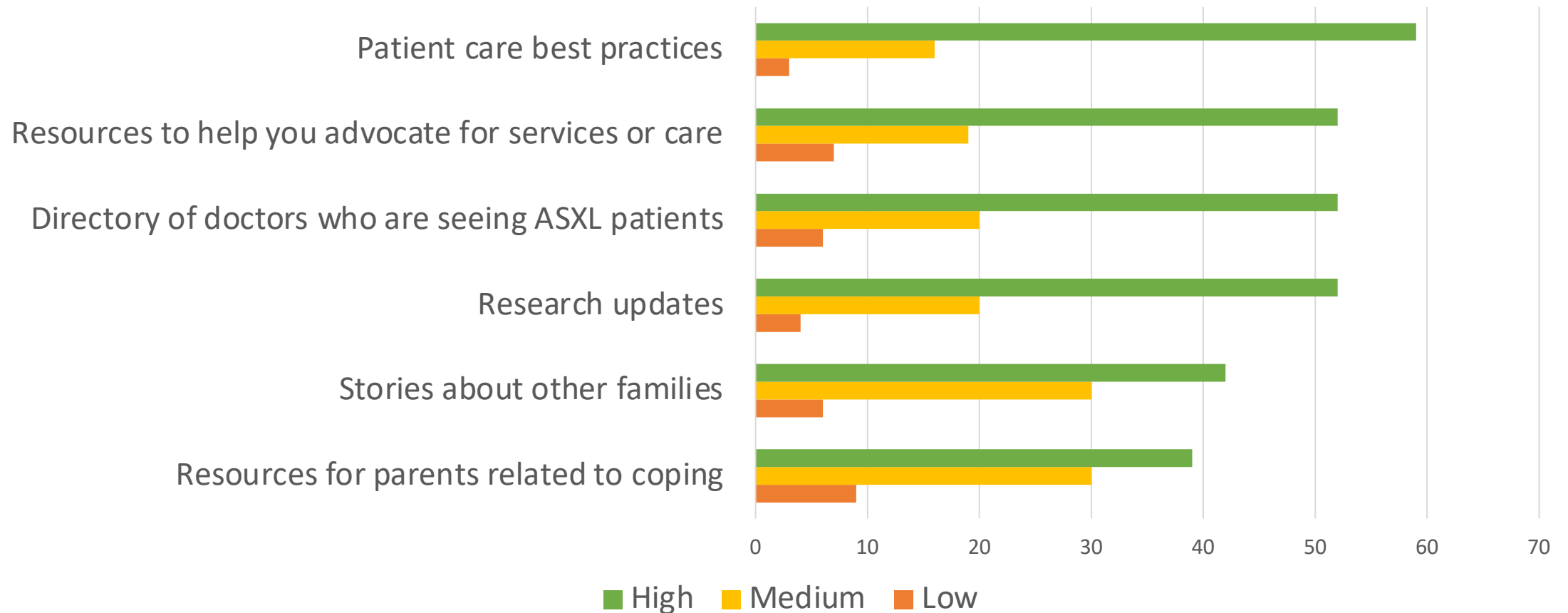
Diagnoses by year



Needs and priorities



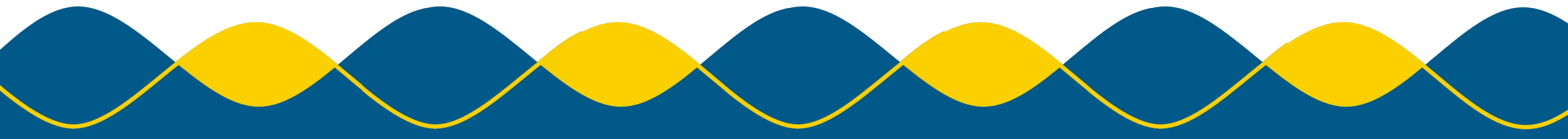
What types of information and resources are most important to you? (rate low to high)



We're planning to focus our work in several major program areas. Rank by importance to you.

by average ranking (1=highest priority, 8=lowest priority)

	Priorities	Average ranking
Highest	Increasing the number of doctors who treat ASXL syndromes	3.54
	Increasing the number of researchers studying ASXL conditions	3.57
	Providing resources for common challenges experienced by those with an ASXL syndrome	3.64
	Funding research grants	4.26
	Providing resources for newly diagnosed families	4.69
	Hosting virtual education sessions for families	5.28
	Encouraging participation in the ASXL Patient Registry and Biobank	5.32
Lowest	Hosting an annual in-person conference for families (when safe to do so)	5.49



What should we start doing?

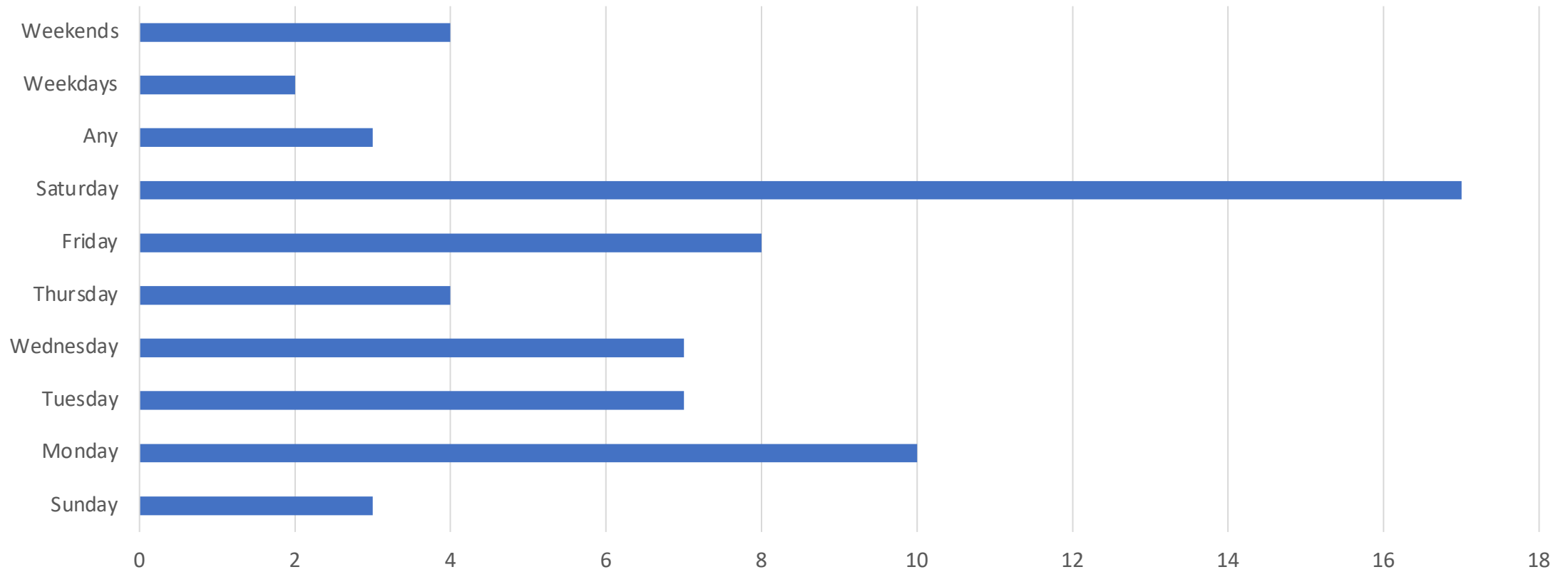
Themes	Frequency
Treatment guidelines/best practices	10
Research updates	4
Education for providers/therapists	5
Advocating for better access to genetic testing	1
Building professional network	1
Centralized source of information on website that links to other sources (Facebook, etc)	1
Gene therapy	1
Genetic research with relatives	1
Increase people studying disease	1
List of specialists	1
Simple language to explain syndrome to personal network	1
Summary of symptoms that others have	1
Support group via Zoom	1
Welcome info for new families	1

What should we stop doing?

Themes	Frequency
Nothing	13
Not sure in-person meet-up is beneficial (variations in syndromes, difficulty traveling)	2
Doctors don't need special ASXL training	1
Excited for BOS Foundation/ARRE collaboration	1

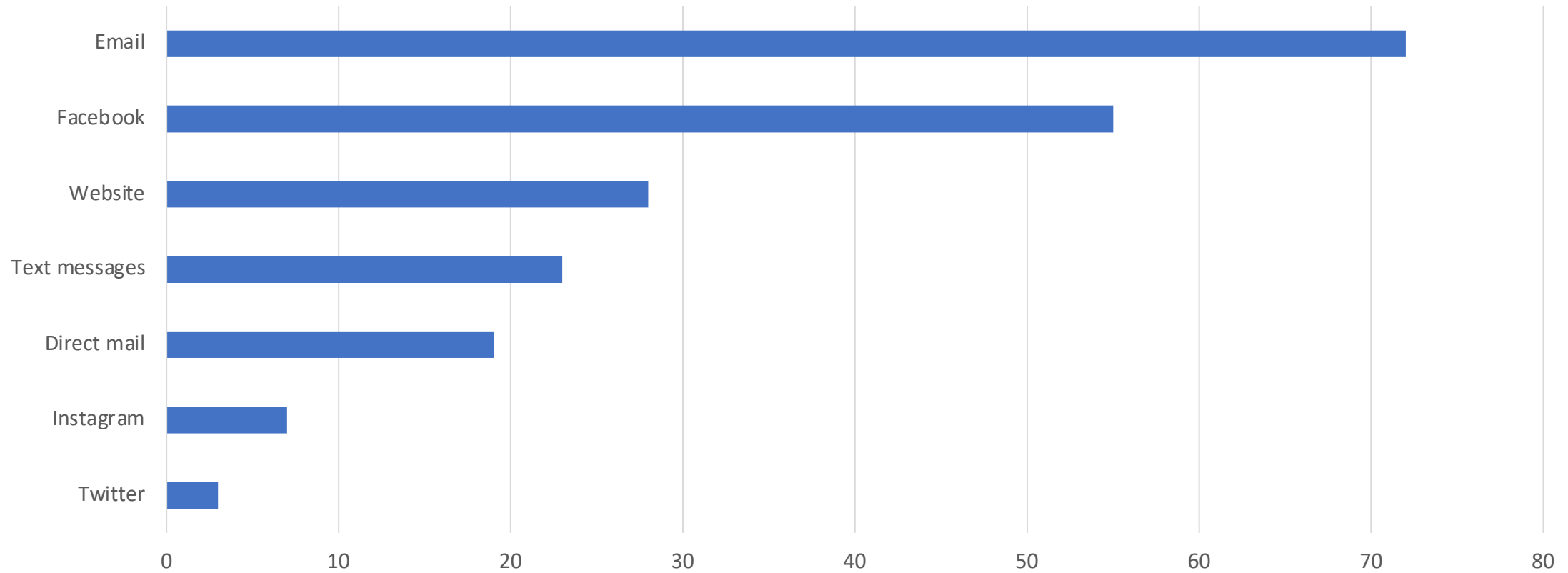


Best day for virtual event/webinar



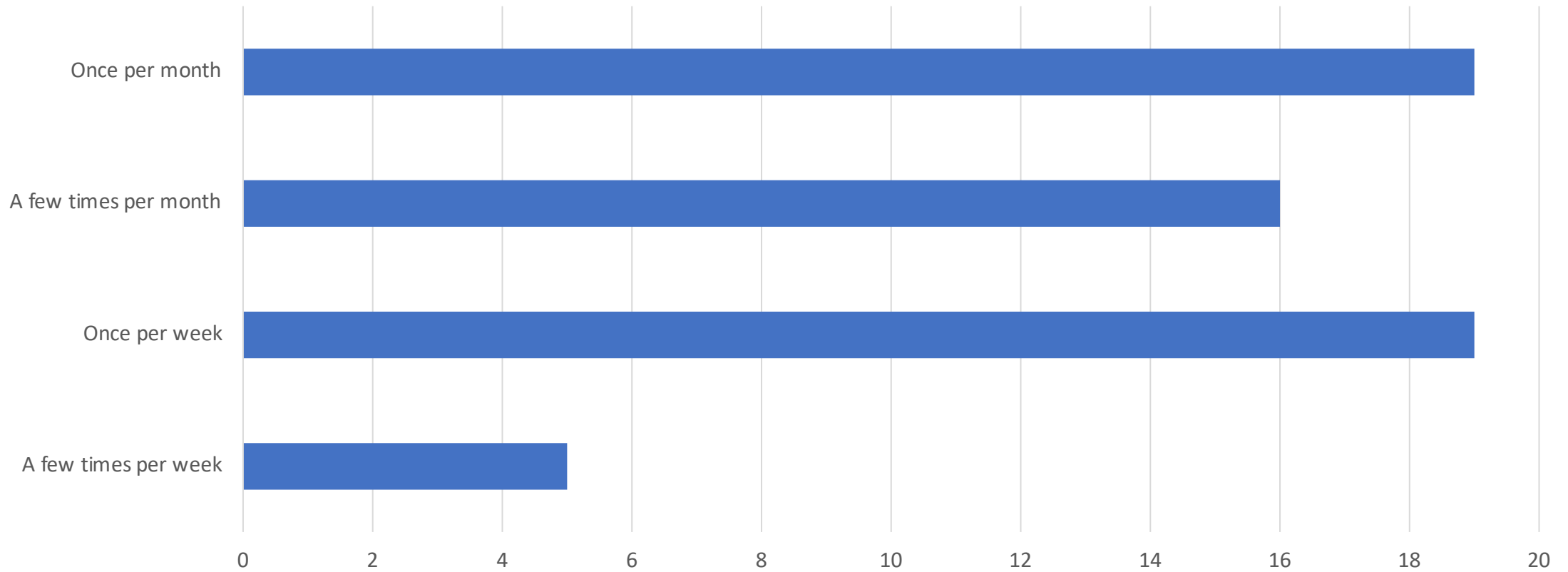
Series 1

Preference for sharing updates and resources



Series 1

Preferred communication frequency



Series 1

Takeaways

- Great need for resources related to care management
- Growing the network of doctors and researchers is important
 - Providing some education for non-specialists would be helpful
- Conferences/webinars are not a high priority when weighed against other priorities
- Email, Facebook and the website are the preferred methods of communication

