

ConnectMPS registry project: Connecting mucopolysaccharidosis and mucolipidosis patients

Vanessa Rangel Miller¹, Terri Klein², Jill Wood³

¹Invitae, San Francisco, CA; ²The National MPS Society, Durham, NC; ³Jonah's Just Begun, Brooklyn, NY

Background

The disease advocacy groups sought to build an engaged patient community willing to share their health data and unmet needs in order to accelerate the development of treatments for mucopolysaccharidosis (MPS) and mucoliposis (ML) and to compel pharmaceutical and biotechnology companies to action. Individually, these rare diseases have very small patient populations, and it would be difficult to fund individual registries for each. Online patient-entered registries are an efficient solution to unite patients and collect insights directly reported by patients and families. In July of 2016, Jonah's Just Begun, Ben's Dream, the National MPS Society, and Invitae came together to re-brand the Sanfilippo Patient Registry to ConnectMPS.org, to extend the program to all forms of MPS, ML and glycoprotein deficiencies, with the support of 24 global patient advocacy organizations.

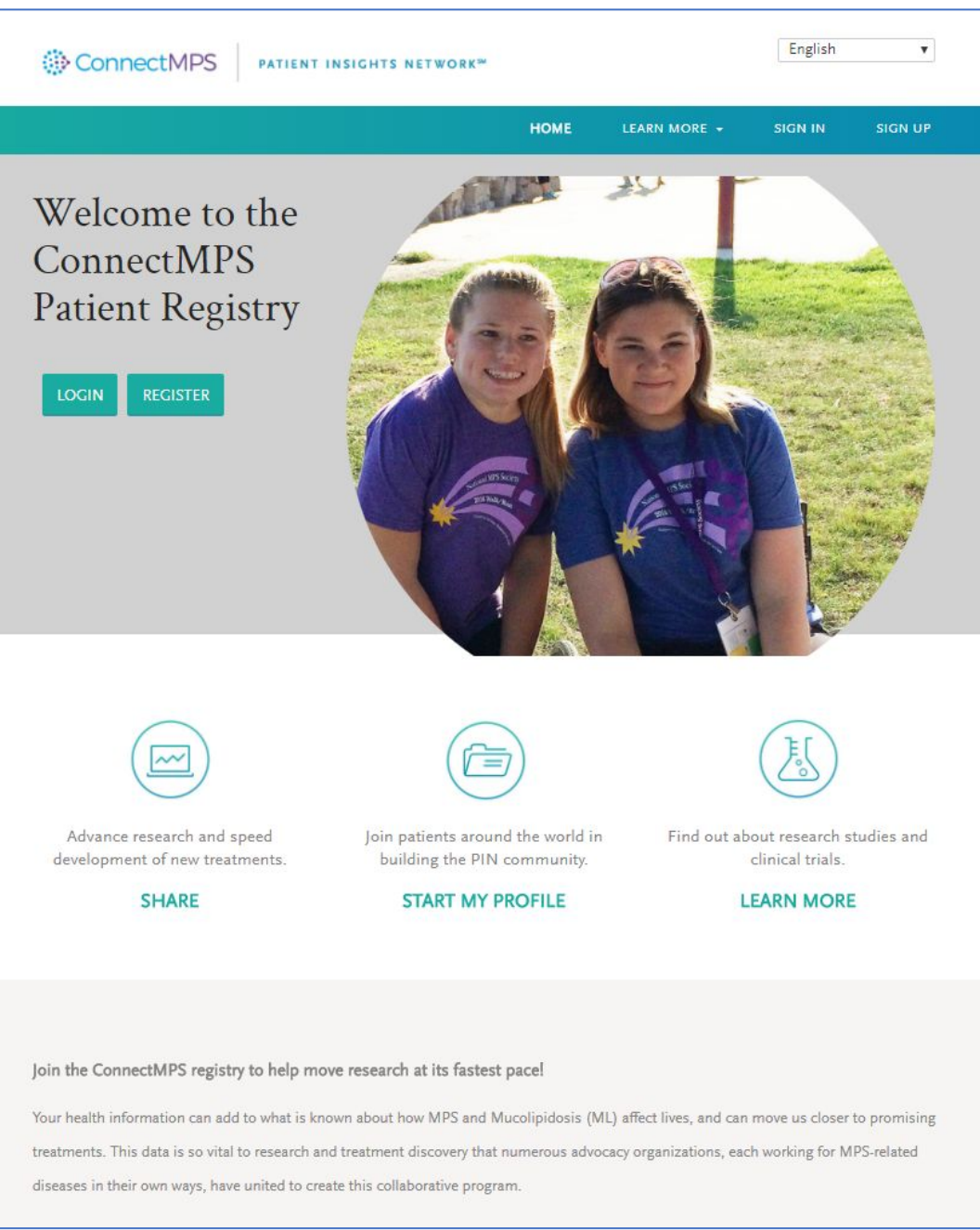


Fig 1. Homepage for ConnectMPS www.connectmps.org.

Design

Represented by 27 disease advocacy organizations, the ConnectMPS Patient Insights Network (PIN) enables patients to contribute medical data through a secure online patient opt-in platform. Participants complete one or more surveys to capture the patient-reported outcomes. Patients may opt in to share their data, which is made available to the greater community through a dedicated online data access portal.

- HIPAA-compliant platform, managed according to FISMA guidelines
- Users create a password-protected account and set data-sharing and contact preferences
- Users can add dependents; extended family members are linked using a unique code
- Data is collected longitudinally and de-identified prior to sharing with participants and professionals
- Languages: English, Portuguese, Spanish, Mandarin, Japanese, German, plus Google Translate
- Mobile responsive

Surveys

Core survey set administered across all 45 PIN programs in network to support pan-disease comparison, include:

- Diagnosis
- Referring organization
- Medical care
- General health
- Family history

Surveys include common data elements and review of systems survey used in rare disease registries (NIH GRDR CDEs, ClinicalGenome Resource).

Participants

1,019 participants enrolled worldwide, of which 944 (93%) have completed the account set-up. Of 920 who provided gender, 43% of participants are female and 57% are male.



Fig 3. Of those who provided their country (920), 680 (74%) are from the US, while 240 are from 53 other countries.

Participants report the diagnosis and subtype:

Alpha mannosidosis	1	MPS IIIB Sanfilippo B syndrome	103
Aspartylglucosaminuria (AGU)	1	MPS IIIC Sanfilippo C syndrome	53
Beta mannosidosis	1	MPS IIID Sanfilippo D syndrome	6
Galactosalisidosis	2	MPS IIIE Sanfilippo E Syndrome	1
MPS I Hurler syndrome	145	MPS IVA Morquio A syndrome	56
MPS I Hurler-Scheie syndrome	31	MPS IVB Morquio B syndrome	6
MPS I Scheie syndrome	10	MPS VI Maroteaux-Lamy syndrome	23
MPS II Hunter syndrome	223	MPS VII Sly syndrome	4
MPS III Sanfilippo (type unknown)	11	Mucopolipidosis II, II/III, III alpha/beta	22
MPS IIIA Sanfilippo A syndrome	243	Mucopolipidosis III gamma	2

Table 1. Participants report diagnosis at account registration.

Patient engagement

Through the expanded partnership of supporting disease advocacy organizations, enrollment has increased steadily, from 246 participants in July 2016 to 1,019 participants in January 2018. This was accomplished through engagement at disease specific family conferences to promote the program, enroll participants, and educate families regarding the importance of their data to accelerate research, treatments and trials. In addition, Jonah's Just Begun and the National MPS Society with funding from the Ryan Foundation donated and raffled prizes to participants. The collaborations between numerous patient organizations have been key to creating a powerful registry.

Data sharing and communication

In ConnectMPS participants own their data and control their data-sharing and contact preferences, which they may change at any time. From July 2016 through January 2018, 27 communications have been sent to participants regarding participation opportunities and research updates.

- Of 922 participants, 805 (87%) want to be contacted about research studies and clinical trials
- Of 872 participants, 872 (70%) agree to be contacted by the ConnectMPS advocacy organization partners

De-identified participant data, accessible to participants and professionals, are presented in chart form. Participants may compare their responses with others with the same diagnosis. Data is exportable into statistical programs using MS Excel or CSV.

- Of participants who finished the core surveys, 74-98 % agreed to share their de-identified survey data

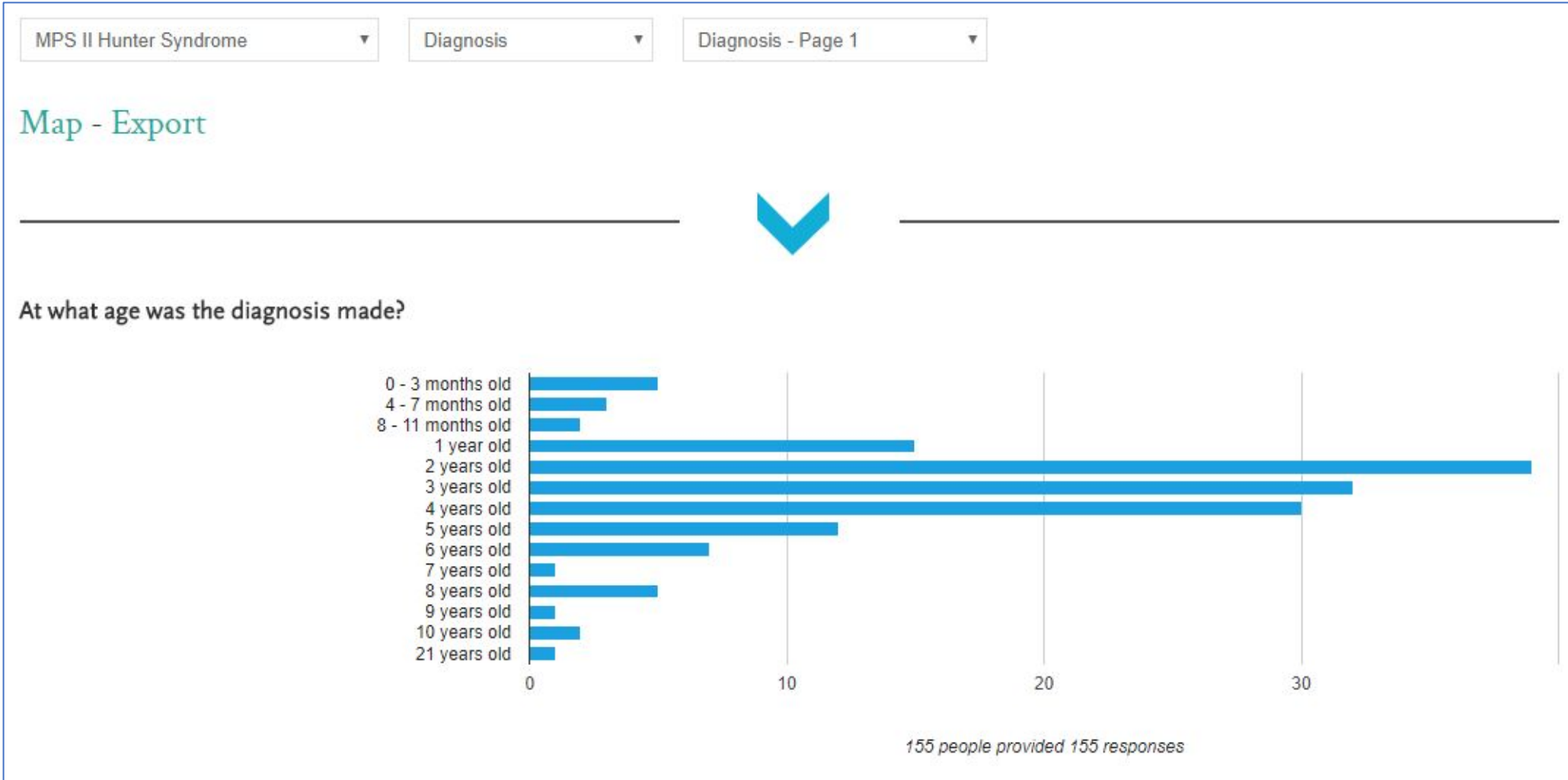


Fig 5. Example chart from data sharing portal.

Of the 944 who completed account set-up:

- 656 (69%) have completed two or more surveys
 - 285 (30%) have completed four or more surveys
- From a targeted request, 62 participants have uploaded copies of genetic, biochemical, developmental test reports.

Since July 2016, ConnectMPS has extended to support two data collections for specific cohorts in support of biopharmaceutical partners.

- 253 participants have completed one or more research surveys administered through ConnectMPS

Summary

ConnectMPS has provided a valuable, secure, and patient-centric format for collecting patient-reported outcome data and for connecting patients globally. The close collaborations between parents/patients let disease organizations and researchers to ensure that the limited funding and time of organizations is well spent as new treatments are pursued. Looking ahead, ConnectMPS aims to extend its model for patient engagement to further support the needs of patients, advocacy organizations, and researchers globally.

We gratefully acknowledge the contributions of the patients and organizations involved with ConnectMPS, and of Jo Anne Vidal (Invitae), the ConnectMPS PIN Coordinator.

