DuchenneConnect

www.DuchenneConnect.org

Parent Project Muscular Dystrophy

1 What is DuchenneConnect?

Web-based patient self-report registry to link the resources and needs of the Duchenne/Becker muscular dystrophy community, including:

- **Care considerations**
- **Access to genetic testing & counseling**
- **Clinical trials & research studies**
- **Profile report**
- **Educational materials**

Prepares the community for Clinical Trials

Who should join?

- **Males with DBMD**
- **Female carriers**

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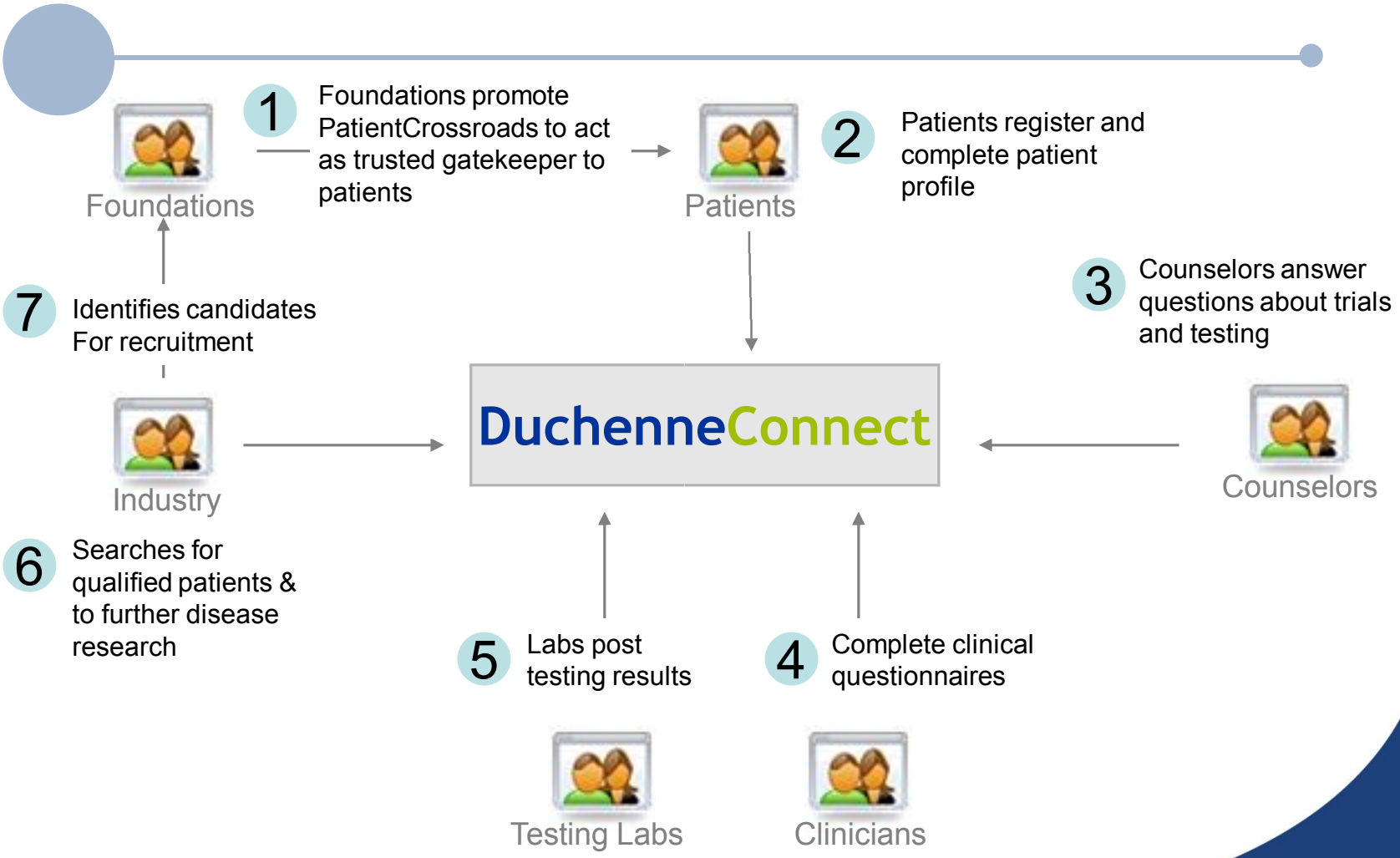
How It Works



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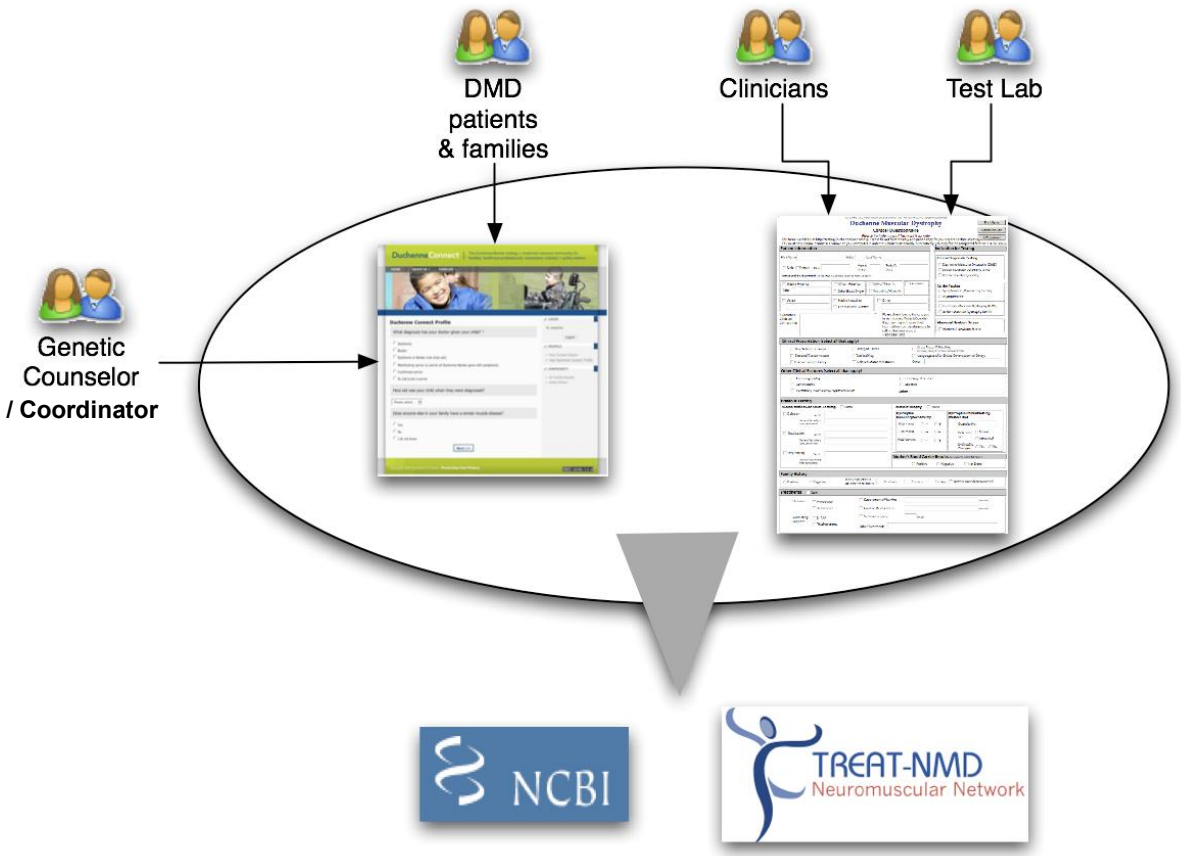
DuchenneConnect Process



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2 Mirrored site for Providers

- **Clinical**
  - **Ability to search for participant characteristics**
  - **Care and management considerations summarized by experts in neuromuscular disorders**
  - **Access to genetic test information & test coordinators**
  - **Educational materials for patients & families**

- **Research**
  - **Ability to search for participant characteristics**
  - **Research and clinical trial updates & developments**
  - **Study feasibility planning and recruitment**

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2 Participants: Patients (Parents)

Participants include:

- Individuals or guardians of those with Duchenne/Becker muscular dystrophy
- Women who are carriers (or at-risk) of Duchenne/Becker

Benefits:

- Resources to assist with early, appropriate and least invasive diagnosis
- Information genetic testing & counseling
- Educational resources
  - Latest information on care and treatment!
- Promote access to new treatment trials
- Co-registration with international TREAT-NMD registry

What will be asked of me?

- Participants submit health information and genetic test results
- Stored in a de-identified database?

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Major Participant Services:

- **DuchenneConnect profile**
  - Provides valuable information about you/your child
- **Profile Report**
  - Review your profile answers and communicate questions and care needs with your providers
- **Care Considerations**
  - Learn about general care strategies from experts in neuromuscular disorders
- **Genetic counseling and testing**
  - Access Connect Counselors for questions related to genetic testing/results, participation, or counseling
- **Education and resources**
  - Find answers to common questions
- **Clinical Trials**
  - Explore current clinical trials

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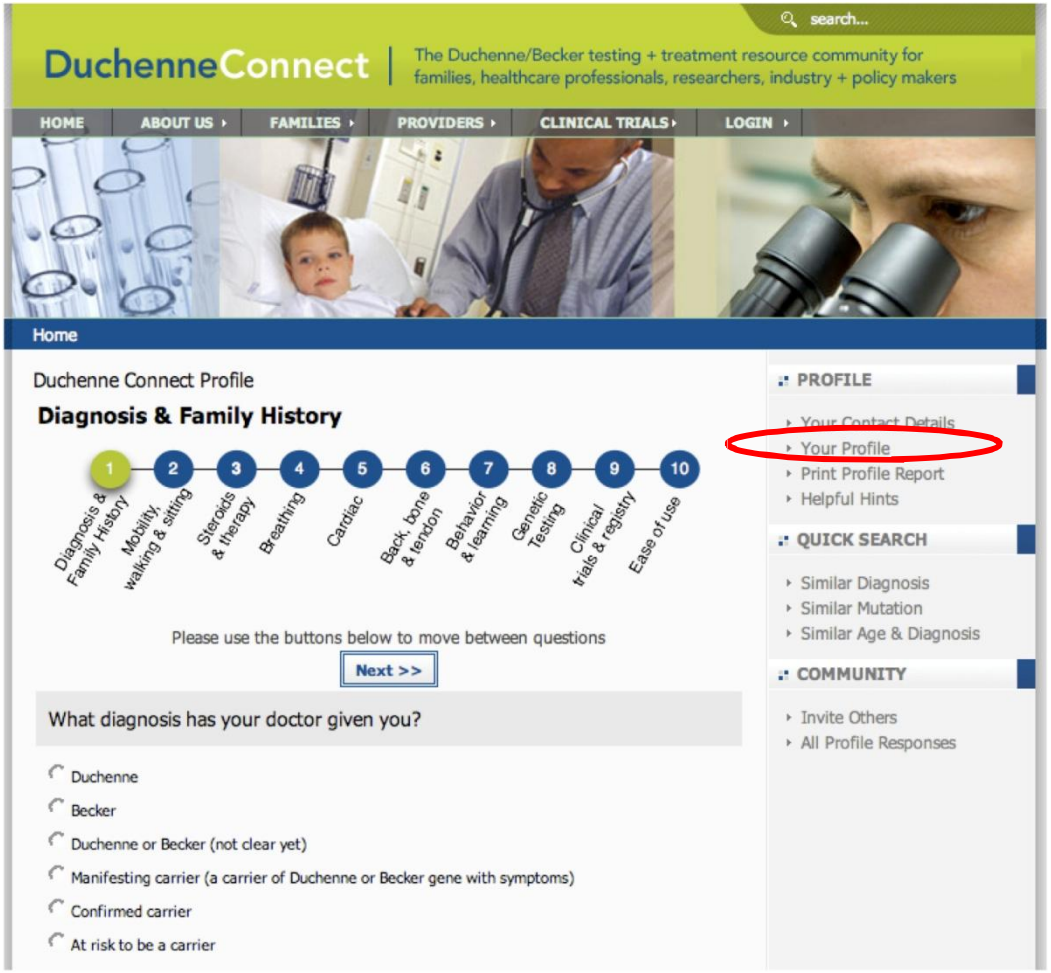
4 Patient (Parent) Participation



Complete the Profile

- Diagnosis & History
- Family history

Clinical Status

- Ambulation
- Medications
- Pulmonary function
- Cardiac function
- Bone, Back
- Behavior

Genetic Testing

- Submit report

Other Registries

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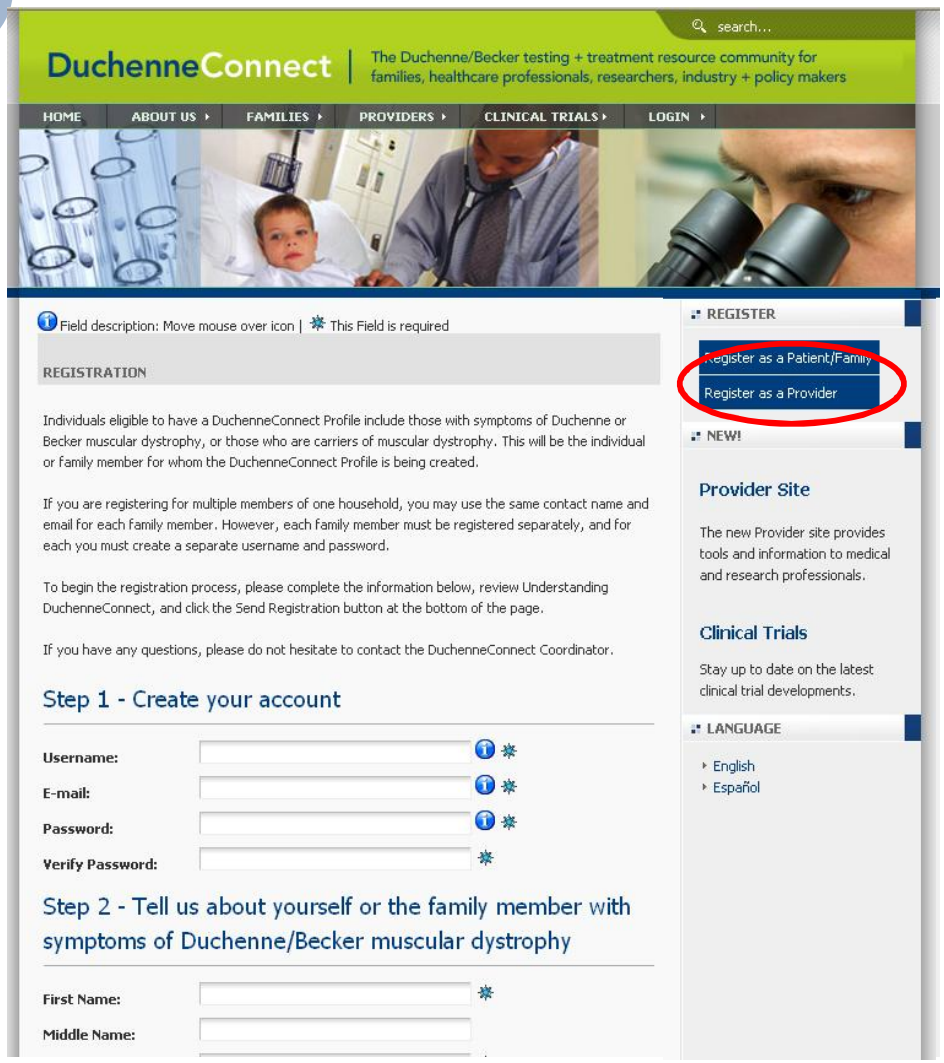
3 Registration



Provider registration

- Provide contact information
- Community affiliation
- Pending staff approval

Patient registration

- By individual, parent, family, caregiver, carrier
- Provide contact and participant information

What do patients provide?

- Complete a profile
- Submit test results
  - Genetic test result
  - FVC
  - LVEF/LVSF

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5 Profile Questions: Genetic Testing & Counseling

Genetic results

- Enter results & submit test report
- Reviewed by genetic counselors

Genetic counseling

- Answer questions
- Arrange testing
- Help obtain results
- Explain results & options

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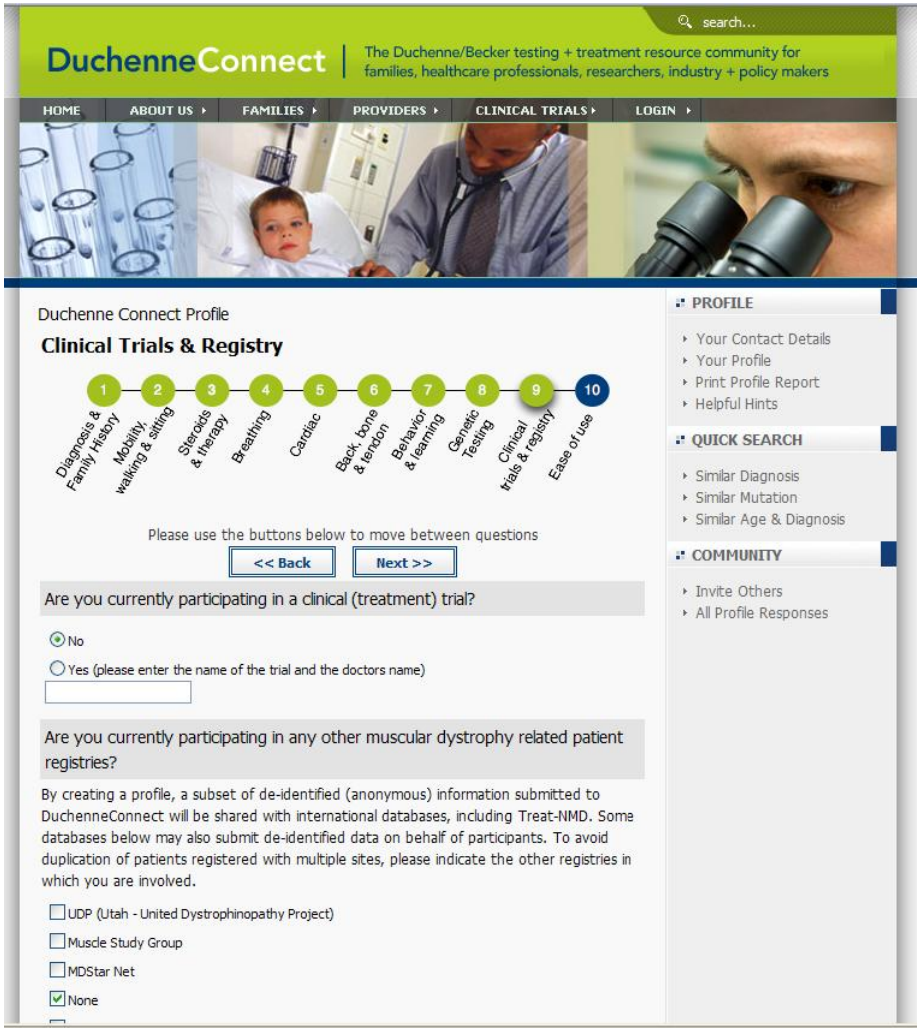
6 Profile Questions:Participation in Global Registries



TREAT-NMD & NCBI

- De-identified data send to TREAT-NMD
- Genetic data shared with NCBI

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Family Benefits

Access to Genetic Counselors

Connect to latest research information

Registry Comparison

- Review responses of entire registry
- Compare similarities
- Feeling of community

Educational Materials

- Duchenne/Becker muscular dystrophy
- Genetics & testing
- Carrier information
- Glossary

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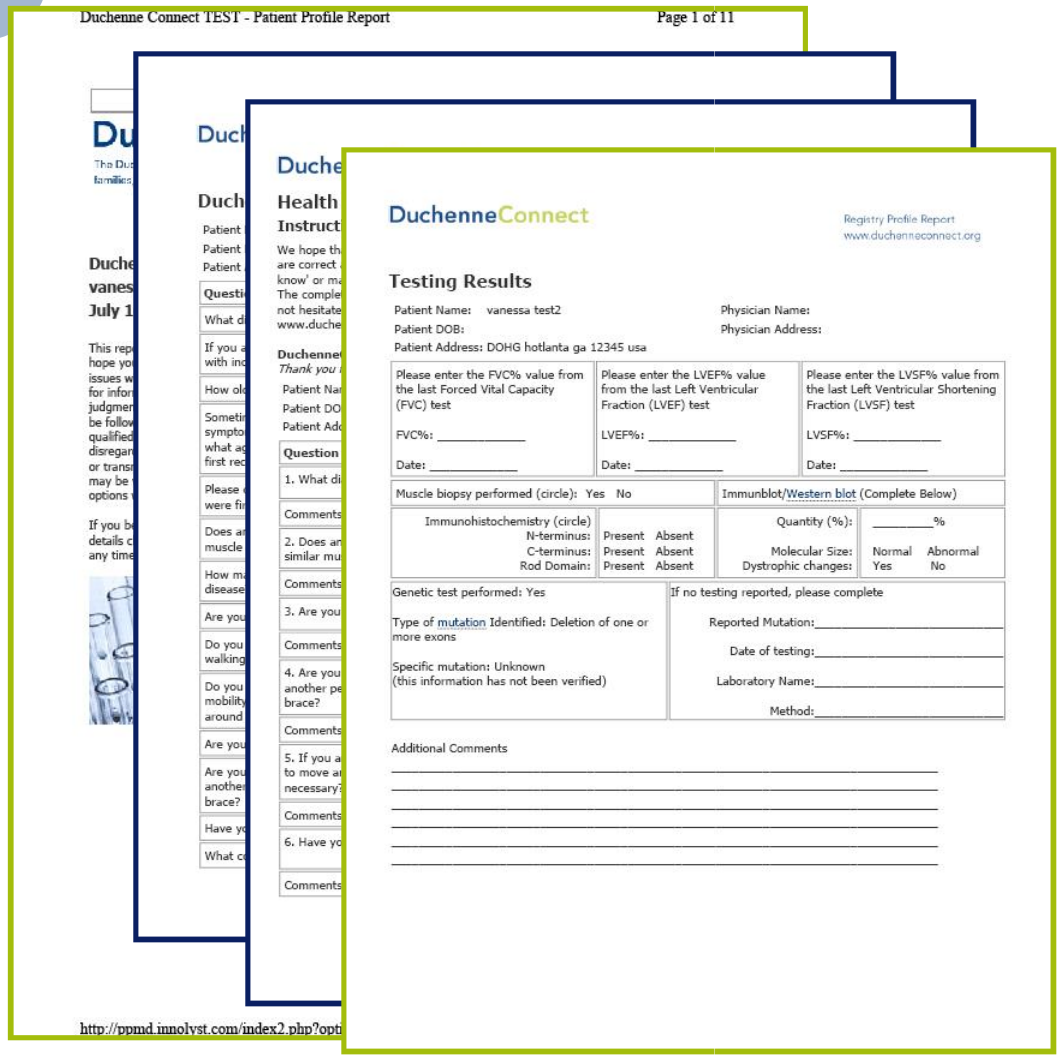
Profile Report Tool



- Communicate health information with care provider
- Identify further needs
- Explore care & management considerations
- Update profile survey responses

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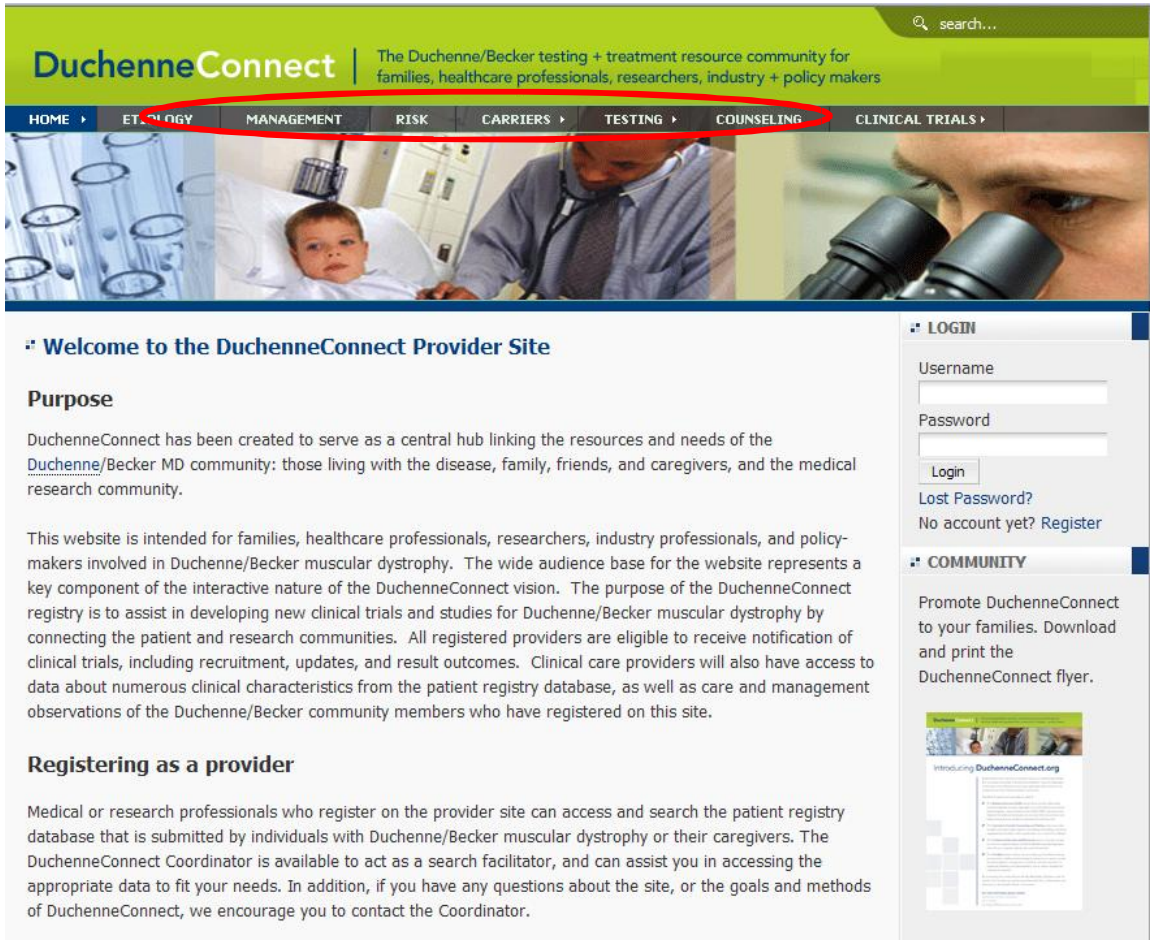
9 Provider Benefits



Access to Genetic Counselors

- Help ordering genetic testing

Educational Materials

- Patient ready materials
- Test pathway
- Understanding of report

Connect to latest research information

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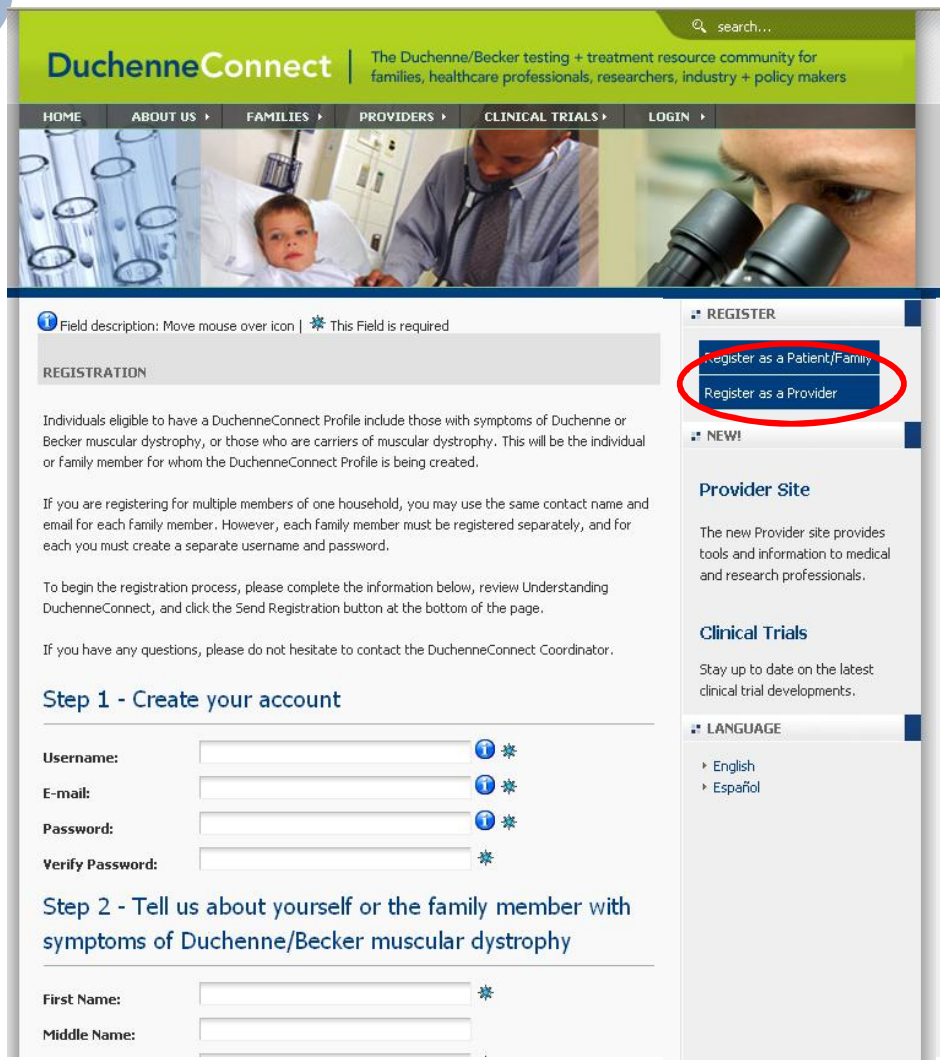
3 Registration



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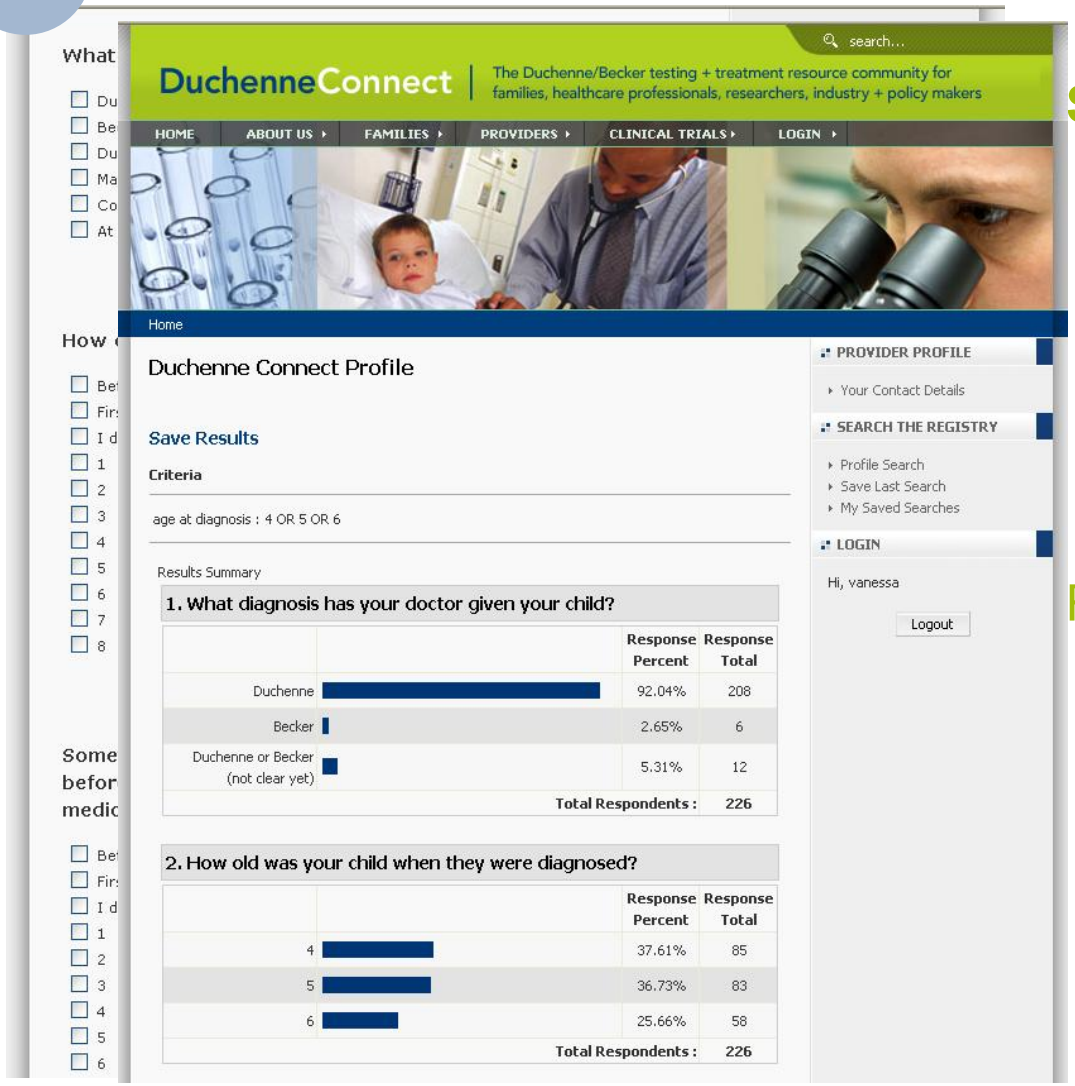
Search & Results



Search the Registry

- Treatment and outcome data
- Recruitment and feasibility planning
- Detailed searches **MUST** be approved by DuchenneConnect Program

Results display

- View aggregate profiles that meet the search criteria
- De-identified information

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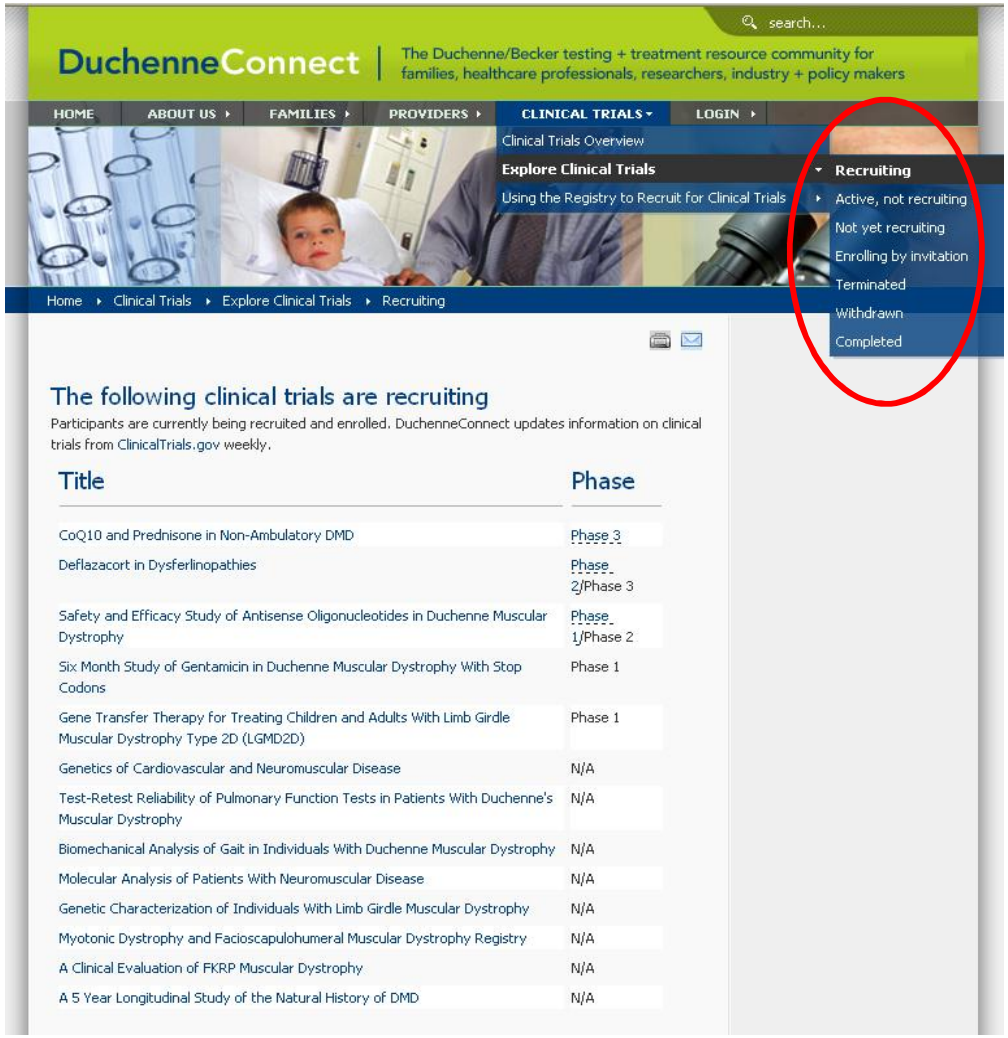
Clinical Trials & Research Recruitment



Find clinical trials

- Updated weekly
- Sorted by trial status

General overview about clinical trials

Use the registry to recruit for trials/studies

- Search for study planning & recruitment
- Review of recruitment material
- Notify patients and providers
- Assistance from coordinator
-

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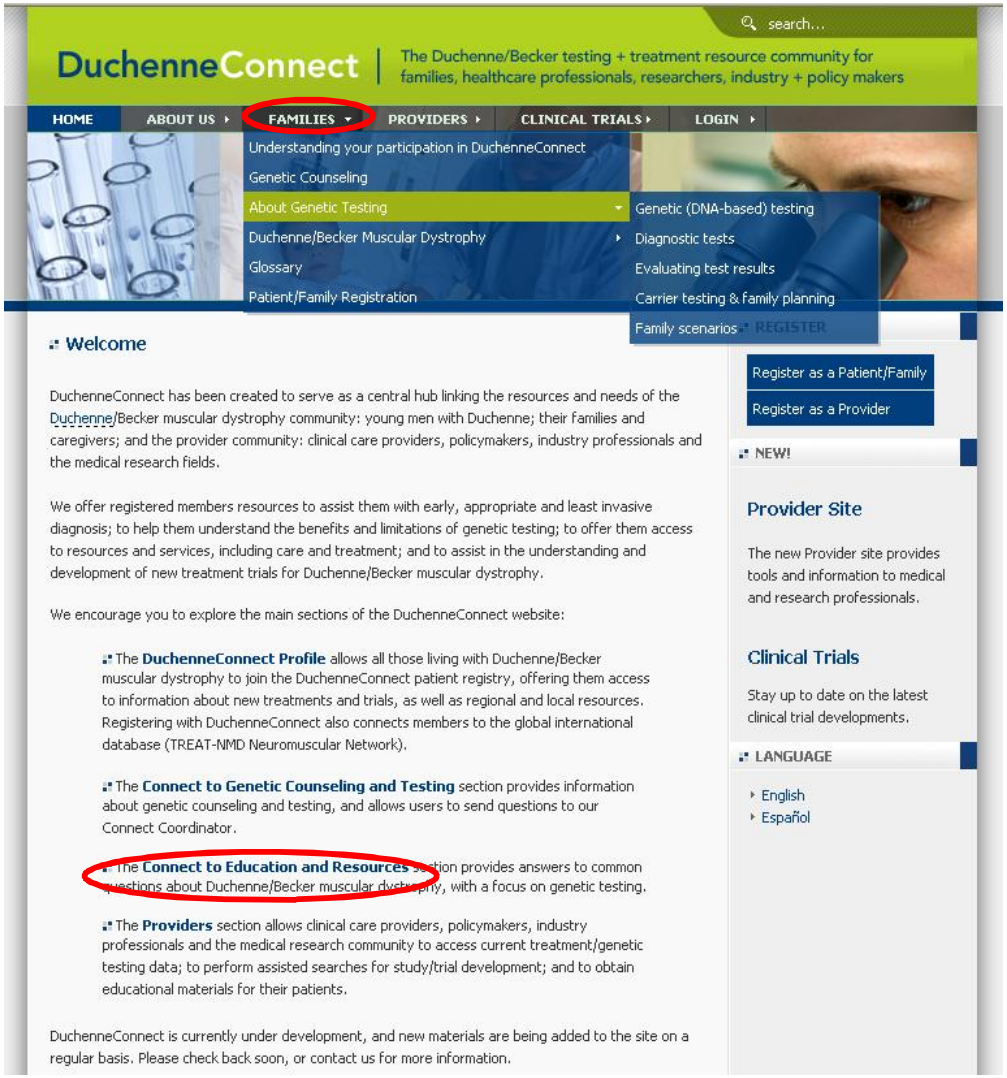
Educational Materials



Topics include

- Duchenne/Becker muscular dystrophy
- Genetics & testing
- Carrier information
- Glossary

Languages

- Spanish
- Additional languages in development

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13 Reporting - Over 60 Countries

Argentina	14
Australia	18
Austria	3
Belgium	4
Brazil	4
Bulgaria	1
Canada	62
Cayman Islands	1
Chile	2
China	1
Colombia	5
Cook Is	1
Costa Rica	1
Croatia	1
Cyprus	1
Czech Republic	5
Ecuador	2
Egypt	6
El Salvador	1
Finland	1
Germany	8
Greece	1
Honduras	2
Hungary	4
India	58
Iran	3
Israel	6
Italy	1
Jordan	3
Kyrgyzstan	2
Kuwait	1
Lebanon	3
Libya	1
Malaysia	1
Mexico	16
Nepal	1
Netherlands	6
New Zealand	2
Nigeria	1
Norway	5
Pakistan	3
Panama	2
Peru	9
Philippines	2
Poland	2
Portugal	1
Puerto Rico	3
Romania	3
Russian Federation	2
Rwanda	1
Saudi Arabia	7
Singapore	6
Slovakia	1
South Africa	9
South Korea	1
Spain	20
Sri Lanka	3
Sweden	2
Switzerland	4
Syria	2
Thailand	1
Turkey	10
Uganda	1
UK	20
Ukraine	1
UAE	1
USA	751
Venezuela	5
Zimbabwe	1

(October 2008)

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Experience to date

Patient/Provider Portal:

- 1600 Registered Participants from 53 Countries – *in 13 months*
  - 75% completed profiles, of which 84% report genetic testing
- 912 Questions from Participants
  - Explain test results, arrange testing, clinical trial & registry participation
  - More than 95% of participants – easy to use
- 210 registered providers

Clinical Trials/Research Studies:

- Screening for one clinical trial identified 39 participants
- Feasibility planning inquiry for potential participants in geographical regions

Future Goals

- Rapid response alerts
- Targeted surveys (ex: supplement use, carriers)
- Toolkit for non-neuromuscular care providers

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