

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:

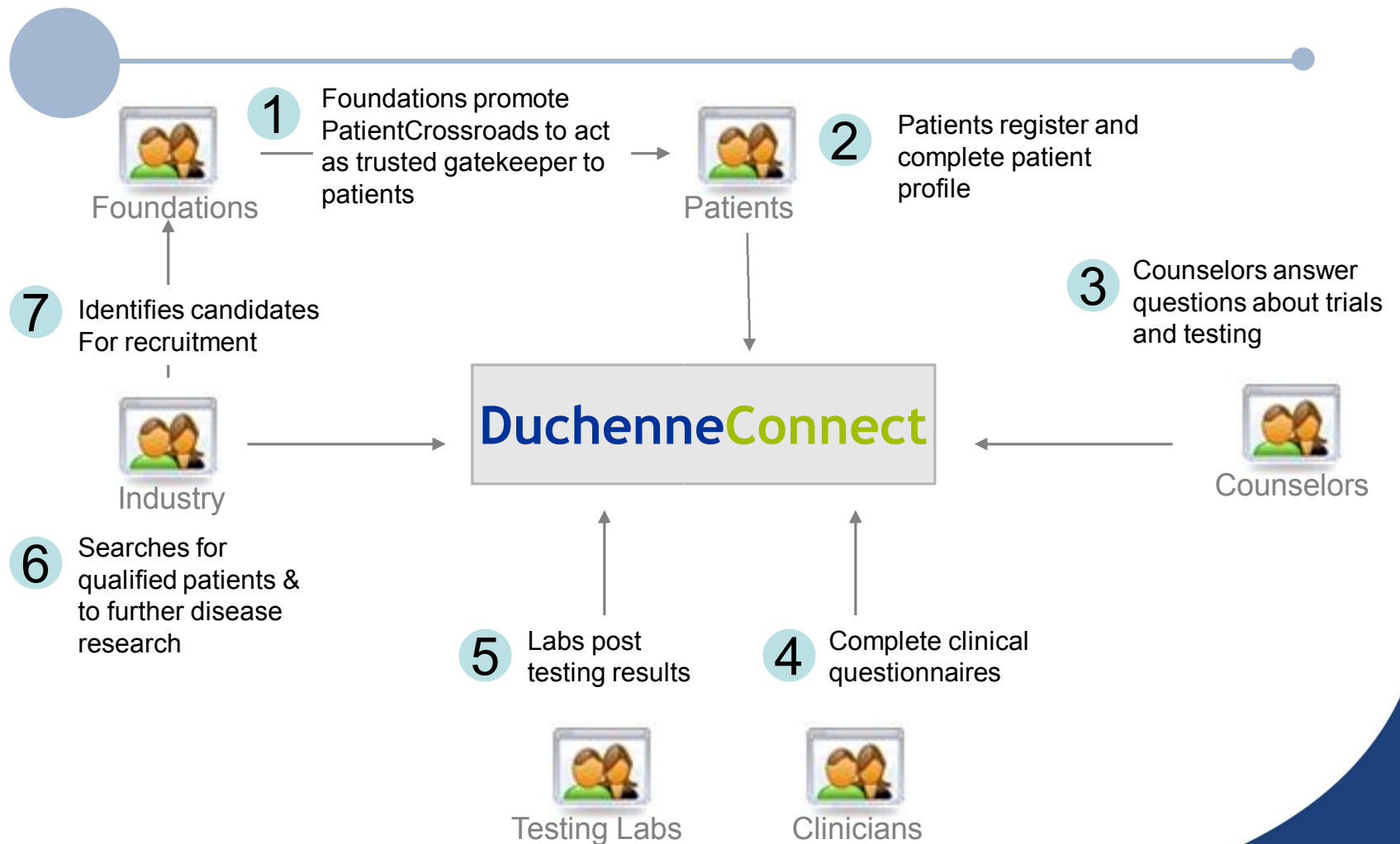
- **Care considerations**
- **Access to genetic testing & counseling**
- **Clinical trials & research studies**
- **Profile report**
- **Educational materials**

Prepares the community for Clinical Trials

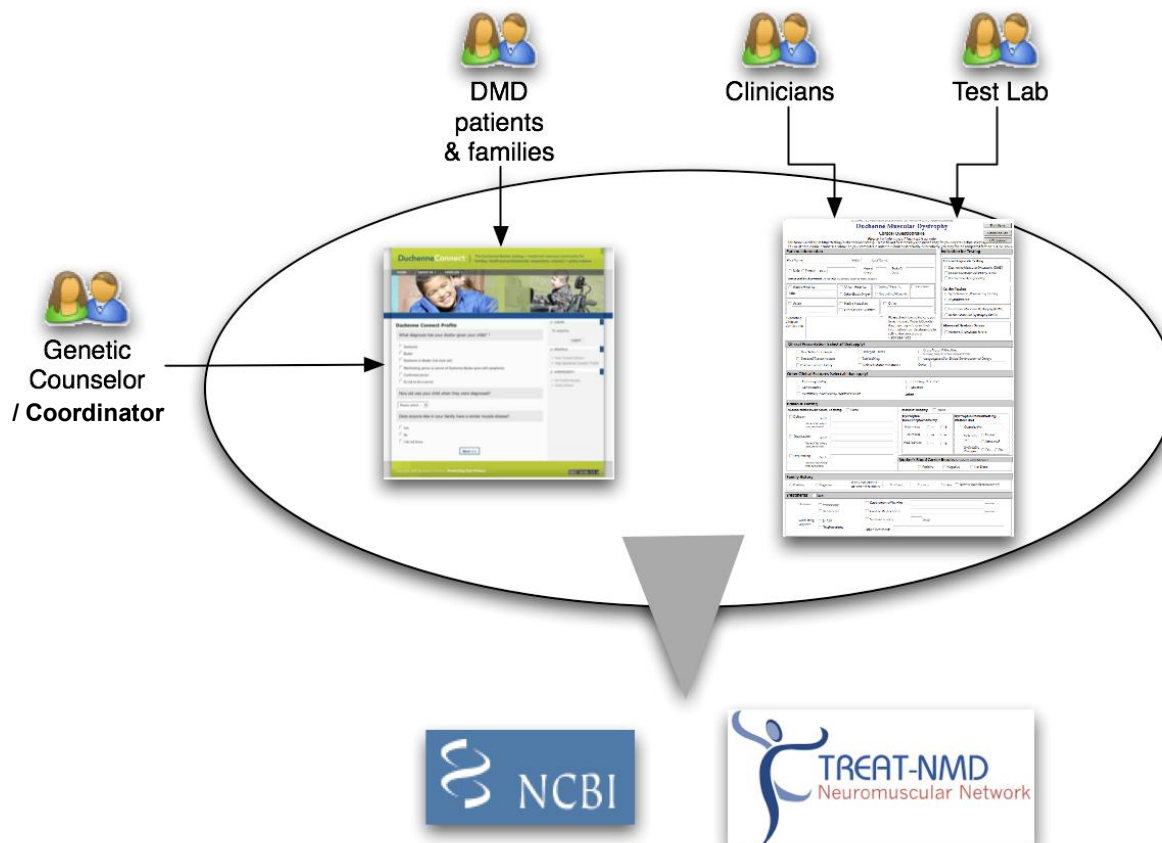
Who should join?

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

How It Works



DuchenneConnect Process



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**

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?

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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Patient (Parent) Participation

DuchenneConnect | The Duchenne/Becker testing + treatment resource community for families, healthcare professionals, researchers, industry + policy makers

HOME ABOUT US FAMILIES PROVIDERS CLINICAL TRIALS LOGIN

Home

Duchenne Connect Profile

Diagnosis & Family History

1 2 3 4 5 6 7 8 9 10

Diagnosis & Family History Mobility, walking & sitting Steroids & therapy Breathing Cardiac Back, bone & tendon Behavior & learning Genetic Testing Clinical trials & registry Ease of use

Please use the buttons below to move between questions

[Next >>](#)

What diagnosis has your doctor given you?

☐ Duchenne

☐ Becker

☐ Duchenne or Becker (not clear yet)

☐ Manifesting carrier (a carrier of Duchenne or Becker gene with symptoms)

☐ Confirmed carrier

☐ At risk to be a carrier

PROFILE

- ▶ Your Contact Details
- ▶ **Your Profile**
- ▶ Print Profile Report
- ▶ Helpful Hints

QUICK SEARCH

- ▶ Similar Diagnosis
- ▶ Similar Mutation
- ▶ Similar Age & Diagnosis

COMMUNITY

- ▶ Invite Others
- ▶ All Profile Responses

Complete the Profile

- Diagnosis & History
- Family history

Clinical Status

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

Genetic Testing

- Submit report

Other Registries

3

Registration

DuchenneConnect | The Duchenne/Becker testing + treatment resource community for families, healthcare professionals, researchers, industry + policy makers

HOME ABOUT US FAMILIES PROVIDERS CLINICAL TRIALS LOGIN

Field description: Move mouse over icon | * This Field is required

REGISTRATION

Individuals eligible to have a DuchenneConnect Profile include those with symptoms of Duchenne or Becker muscular dystrophy, or those who are carriers of muscular dystrophy. This will be the individual or family member for whom the DuchenneConnect Profile is being created.

If you are registering for multiple members of one household, you may use the same contact name and email for each family member. However, each family member must be registered separately, and for each you must create a separate username and password.

To begin the registration process, please complete the information below, review Understanding DuchenneConnect, and click the Send Registration button at the bottom of the page.

If you have any questions, please do not hesitate to contact the DuchenneConnect Coordinator.

Step 1 - Create your account

Username: ⓘ *

E-mail: ⓘ *

Password: ⓘ *

Verify Password: *

Step 2 - Tell us about yourself or the family member with symptoms of Duchenne/Becker muscular dystrophy

First Name: *

Middle Name:

REGISTER

Register as a Patient/Family

Register as a Provider

NEW!

Provider Site

The new Provider site provides tools and information to medical and research professionals.

Clinical Trials

Stay up to date on the latest clinical trial developments.

LANGUAGE

English

Español

Provider registration

- Provide contact information
- Community affiliation
- Pending staff approval

Patient registration

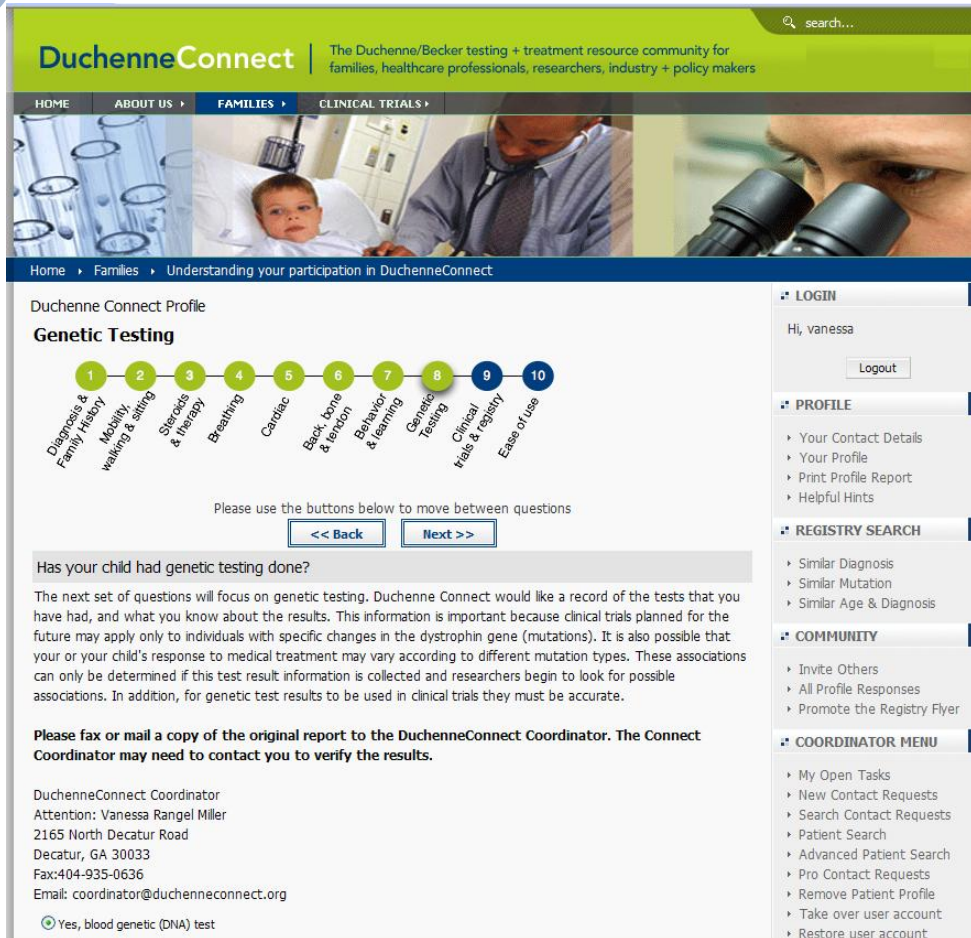
- By individual, parent, family, caregiver, carrier
- Provide contact and participant information

What do patients provide?

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

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Profile Questions: Genetic Testing & Counseling



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HOME ABOUT US FAMILIES CLINICAL TRIALS

Home » Families » Understanding your participation in DuchenneConnect

Duchenne Connect Profile

Genetic Testing

1 2 3 4 5 6 7 8 9 10

Diagnosis & Family History Mobility, walking & sitting Strengths & therapy Breathing Cardiac Back, bone & tendon Behavior & learning Genetic Testing Clinical trials & registry Ease of Use

Please use the buttons below to move between questions

<< Back Next >>

Has your child had genetic testing done?

The next set of questions will focus on genetic testing. Duchenne Connect would like a record of the tests that you have had, and what you know about the results. This information is important because clinical trials planned for the future may apply only to individuals with specific changes in the dystrophin gene (mutations). It is also possible that your or your child's response to medical treatment may vary according to different mutation types. These associations can only be determined if this test result information is collected and researchers begin to look for possible associations. In addition, for genetic test results to be used in clinical trials they must be accurate.

Please fax or mail a copy of the original report to the DuchenneConnect Coordinator. The Connect Coordinator may need to contact you to verify the results.

DuchenneConnect Coordinator
Attention: Vanessa Rangel Miller
2165 North Decatur Road
Decatur, GA 30033
Fax: 404-935-0636
Email: coordinator@duchenneconnect.org

☒ Yes, blood genetic (DNA) test

LOGIN
Hi, vanessa
Logout

PROFILE
Your Contact Details
Your Profile
Print Profile Report
Helpful Hints

REGISTRY SEARCH
Similar Diagnosis
Similar Mutation
Similar Age & Diagnosis

COMMUNITY
Invite Others
All Profile Responses
Promote the Registry Flyer

COORDINATOR MENU
My Open Tasks
New Contact Requests
Search Contact Requests
Patient Search
Advanced Patient Search
Pro Contact Requests
Remove Patient Profile
Take over user account
Restore user account

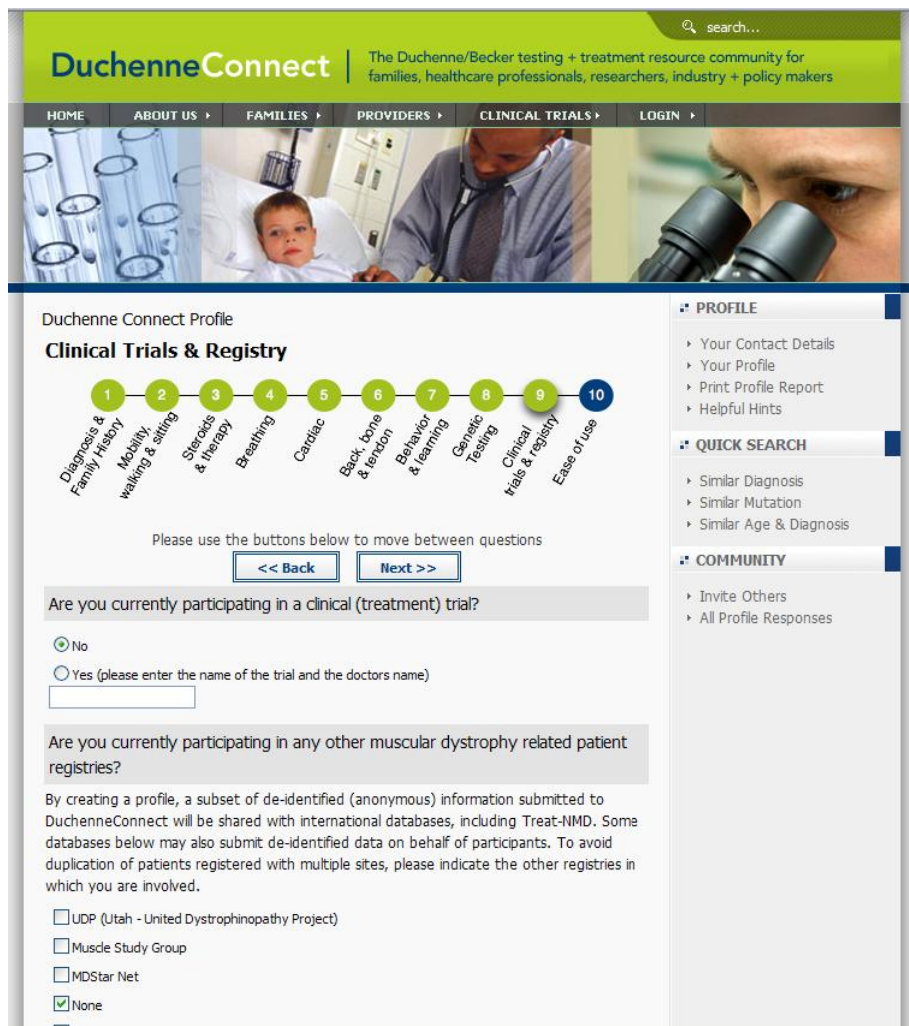
Genetic results

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

Genetic counseling

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

Profile Questions: Participation in Global Registries



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HOME ABOUT US FAMILIES PROVIDERS CLINICAL TRIALS LOGIN

Duchenne Connect Profile

Clinical Trials & Registry

1 2 3 4 5 6 7 8 9 10
 Diagnosis & Family History Mobility, walking & sitting Steroids & therapy Breathing Cardiac Back, bone & tendon Behavior & learning Genetic Testing Clinical trials & registry Ease of Use

Please use the buttons below to move between questions

<< Back Next >>

Are you currently participating in a clinical (treatment) trial?

☒ No

☐ Yes (please enter the name of the trial and the doctors name)

Are you currently participating in any other muscular dystrophy related patient registries?

By creating a profile, a subset of de-identified (anonymous) information submitted to DuchenneConnect will be shared with international databases, including Treat-NMD. Some databases below may also submit de-identified data on behalf of participants. To avoid duplication of patients registered with multiple sites, please indicate the other registries in which you are involved.

☐ UDP (Utah - United Dystrophinopathy Project)

☐ Muscle Study Group

☐ MDStar Net

☒ None

PROFILE

- Your Contact Details
- Your Profile
- Print Profile Report
- Helpful Hints

QUICK SEARCH

- Similar Diagnosis
- Similar Mutation
- Similar Age & Diagnosis

COMMUNITY

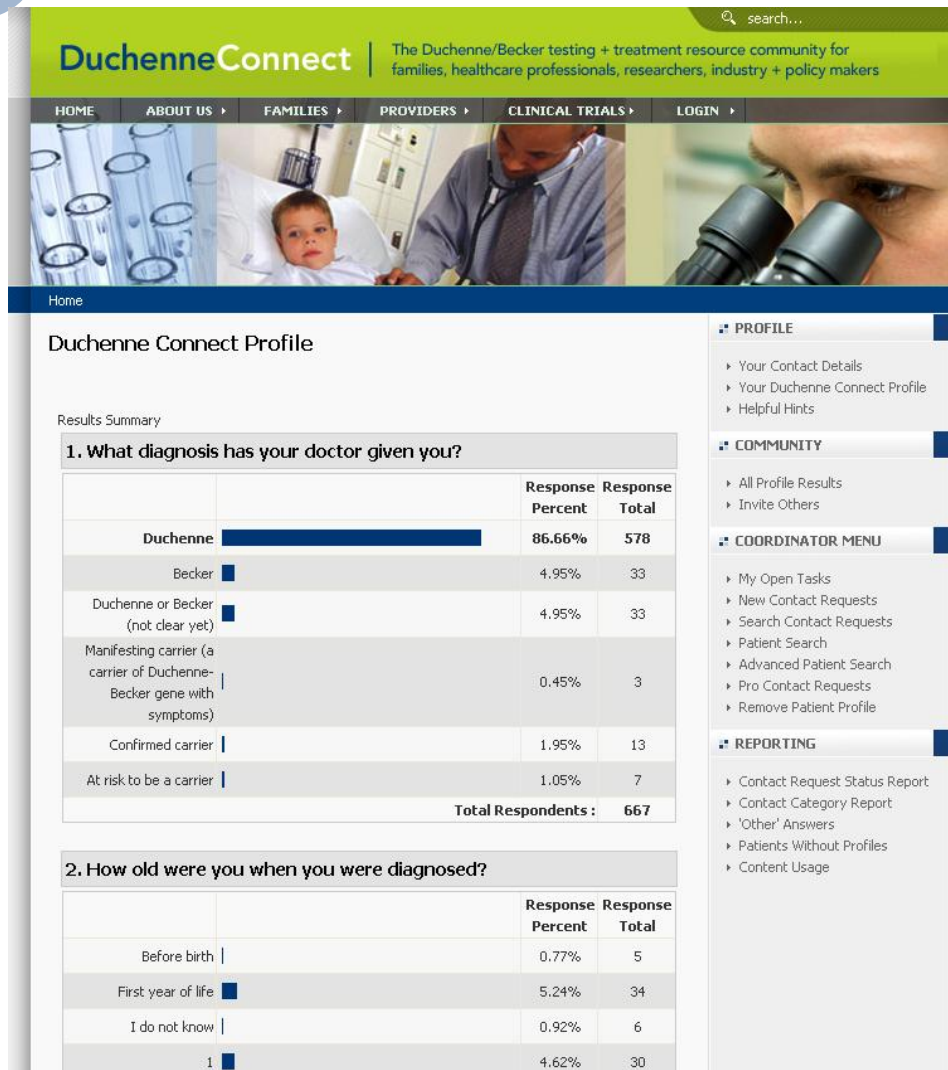
- Invite Others
- All Profile Responses

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

Profile Report Tool

Duchenne Connect TEST - Patient Profile Report

Page 1 of 11

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Testing Results

Patient Name: vanessa test2

Patient DOB:

Patient Address: DOHG hotlanta ga 12345 usa

Physician Name:

Physician Address:

Please enter the FVC% value from the last Forced Vital Capacity (FVC) test

FVC%: _____

Date: _____

Please enter the LVEF% value from the last Left Ventricular Fraction (LVEF) test

LVEF%: _____

Date: _____

Please enter the LVSP% value from the last Left Ventricular Shortening Fraction (LVSP) test

LVSP%: _____

Date: _____

Muscle biopsy performed (circle): Yes No

Immunoblot/Western blot (Complete Below)

Immunohistochemistry (circle)
N-terminus: Present Absent
C-terminus: Present Absent
Rod Domain: Present Absent

Quantity (%): _____ %
Molecular Size: _____
Dystrophic changes: _____

Normal Abnormal
Yes No

Genetic test performed: Yes

If no testing reported, please complete

Type of mutation Identified: Deletion of one or more exons

Reported Mutation: _____

Specific mutation: Unknown (this information has not been verified)

Date of testing: _____

Laboratory Name: _____

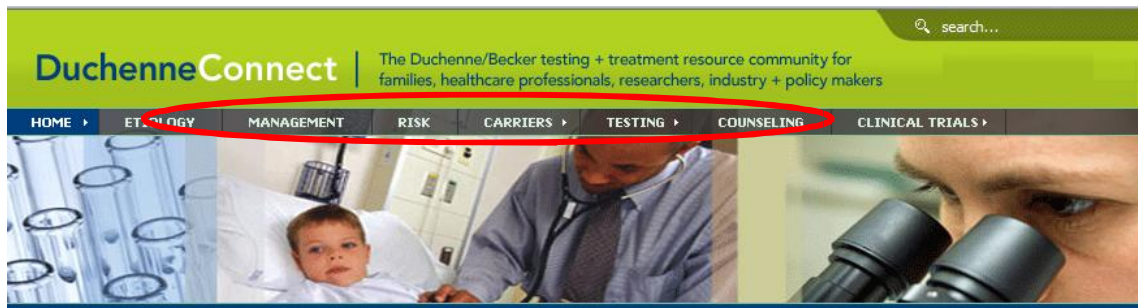
Method: _____

Additional Comments

<http://ppmd.innovyst.com/index2.php?opt>

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

Provider Benefits



DuchenneConnect | The Duchenne/Becker testing + treatment resource community for families, healthcare professionals, researchers, industry + policy makers

HOME > ETIOLOGY > MANAGEMENT > RISK > CARRIERS > **TESTING > COUNSELING** > CLINICAL TRIALS >

Welcome to the DuchenneConnect Provider Site

Purpose

DuchenneConnect has been created to serve as a central hub linking the resources and needs of the Duchenne/Becker MD community: those living with the disease, family, friends, and caregivers, and the medical research community.

This website is intended for families, healthcare professionals, researchers, industry professionals, and policy-makers involved in Duchenne/Becker muscular dystrophy. The wide audience base for the website represents a key component of the interactive nature of the DuchenneConnect vision. The purpose of the DuchenneConnect registry is to assist in developing new clinical trials and studies for Duchenne/Becker muscular dystrophy by connecting the patient and research communities. All registered providers are eligible to receive notification of clinical trials, including recruitment, updates, and result outcomes. Clinical care providers will also have access to data about numerous clinical characteristics from the patient registry database, as well as care and management observations of the Duchenne/Becker community members who have registered on this site.

Registering as a provider

Medical or research professionals who register on the provider site can access and search the patient registry database that is submitted by individuals with Duchenne/Becker muscular dystrophy or their caregivers. The DuchenneConnect Coordinator is available to act as a search facilitator, and can assist you in accessing the appropriate data to fit your needs. In addition, if you have any questions about the site, or the goals and methods of DuchenneConnect, we encourage you to contact the Coordinator.

LOGIN

Username


Password

Login

Lost Password?
 No account yet? Register

COMMUNITY

Promote DuchenneConnect to your families. Download and print the DuchenneConnect flyer.



Access to Genetic Counselors

- Help ordering genetic testing

Educational Materials

- Patient ready materials
- Test pathway
- Understanding of report

Connect to latest research information

3

Registration

DuchenneConnect | The Duchenne/Becker testing + treatment resource community for families, healthcare professionals, researchers, industry + policy makers

HOME ABOUT US FAMILIES PROVIDERS CLINICAL TRIALS LOGIN

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Username: ⓘ *

E-mail: ⓘ *

Password: ⓘ *

Verify Password: *

Step 2 - Tell us about yourself or the family member with symptoms of Duchenne/Becker muscular dystrophy

First Name: *

Middle Name:

REGISTER

[Register as a Patient/Family](#)

[Register as a Provider](#)

NEW!

Provider Site

The new Provider site provides tools and information to medical and research professionals.

Clinical Trials

Stay up to date on the latest clinical trial developments.

LANGUAGE

English

Español

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

Search & Results

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The screenshot shows the DuchenneConnect website interface. The header includes the logo and navigation links: HOME, ABOUT US, FAMILIES, PROVIDERS, CLINICAL TRIALS, and LOGIN. Below the header is a banner image showing a child in a hospital bed and a doctor using a microscope. The main content area displays search results for the question '1. What diagnosis has your doctor given your child?'. The results are summarized in a table with columns for diagnosis, Response Percent, and Response Total. The data shows that 92.04% (208 responses) are for Duchenne, 2.65% (6 responses) are for Becker, and 5.31% (12 responses) are for 'Duchenne or Becker (not clear yet)'. The total number of respondents is 226. Below this, there is another question '2. How old was your child when they were diagnosed?' with a similar table showing response percentages and totals for ages 4, 5, and 6.

Diagnosis	Response Percent	Response Total
Duchenne	92.04%	208
Becker	2.65%	6
Duchenne or Becker (not clear yet)	5.31%	12
Total Respondents :		226

Age	Response Percent	Response Total
4	37.61%	85
5	36.73%	83
6	25.66%	58
Total Respondents :		226

PROVIDER PROFILE

▶ Your Contact Details

SEARCH THE REGISTRY

▶ Profile Search

▶ Save Last Search

▶ My Saved Searches

LOGIN

Hi, vanessa

Logout

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

Clinical Trials & Research Recruitment

The screenshot shows the DuchenneConnect website. The header includes the site name and a search bar. The navigation menu has links for Home, About Us, Families, Providers, Clinical Trials, and Login. The Clinical Trials dropdown menu is open, showing options like Clinical Trials Overview, Explore Clinical Trials, and a Recruiting submenu. The Recruiting submenu is circled in red and lists: Active, not recruiting; Not yet recruiting; Enrolling by invitation; Terminated; Withdrawn; and Completed. The main content area displays a list of clinical trials currently recruiting, with columns for Title and Phase.

Title	Phase
CoQ10 and Prednisone in Non-Ambulatory DMD	Phase 3
Deflazacort in Dysferlinopathies	Phase 2/Phase 3
Safety and Efficacy Study of Antisense Oligonucleotides in Duchenne Muscular Dystrophy	Phase 1/Phase 2
Six Month Study of Gentamicin in Duchenne Muscular Dystrophy With Stop Codons	Phase 1
Gene Transfer Therapy for Treating Children and Adults With Limb Girdle Muscular Dystrophy Type 2D (LGMD2D)	Phase 1
Genetics of Cardiovascular and Neuromuscular Disease	N/A
Test-Retest Reliability of Pulmonary Function Tests in Patients With Duchenne's Muscular Dystrophy	N/A
Biomechanical Analysis of Gait in Individuals With Duchenne Muscular Dystrophy	N/A
Molecular Analysis of Patients With Neuromuscular Disease	N/A
Genetic Characterization of Individuals With Limb Girdle Muscular Dystrophy	N/A
Myotonic Dystrophy and Facioscapulohumeral Muscular Dystrophy Registry	N/A
A Clinical Evaluation of FKRP Muscular Dystrophy	N/A
A 5 Year Longitudinal Study of the Natural History of DMD	N/A

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

Educational Materials

DuchenneConnect | The Duchenne/Becker testing + treatment resource community for families, healthcare professionals, researchers, industry + policy makers

HOME ABOUT US **FAMILIES** PROVIDERS CLINICAL TRIALS LOGIN

Understanding your participation in DuchenneConnect
Genetic Counseling
About Genetic Testing
Duchenne/Becker Muscular Dystrophy
Glossary
Patient/Family Registration

Genetic (DNA-based) testing
Diagnostic tests
Evaluating test results
Carrier testing & family planning
Family scenarios **REGISTER**

Welcome

DuchenneConnect has been created to serve as a central hub linking the resources and needs of the Duchenne/Becker muscular dystrophy community: young men with Duchenne; their families and caregivers; and the provider community: clinical care providers, policymakers, industry professionals and the medical research fields.

We offer registered members resources to assist them with early, appropriate and least invasive diagnosis; to help them understand the benefits and limitations of genetic testing; to offer them access to resources and services, including care and treatment; and to assist in the understanding and development of new treatment trials for Duchenne/Becker muscular dystrophy.

We encourage you to explore the main sections of the DuchenneConnect website:

- The **DuchenneConnect Profile** allows all those living with Duchenne/Becker muscular dystrophy to join the DuchenneConnect patient registry, offering them access to information about new treatments and trials, as well as regional and local resources. Registering with DuchenneConnect also connects members to the global international database (TREAT-NMD Neuromuscular Network).
- The **Connect to Genetic Counseling and Testing** section provides information about genetic counseling and testing, and allows users to send questions to our Connect Coordinator.
- The **Connect to Education and Resources** section provides answers to common questions about Duchenne/Becker muscular dystrophy, with a focus on genetic testing.
- The **Providers** section allows clinical care providers, policymakers, industry professionals and the medical research community to access current treatment/genetic testing data; to perform assisted searches for study/trial development; and to obtain educational materials for their patients.

DuchenneConnect is currently under development, and new materials are being added to the site on a regular basis. Please check back soon, or contact us for more information.

Register as a Patient/Family
Register as a Provider

NEW!

Provider Site

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Clinical Trials

Stay up to date on the latest clinical trial developments.

LANGUAGE

English
Español

Topics include

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

Languages

- Spanish
- Additional languages in development

13

Reporting - Over 60 Countries

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

(October 2008)

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

www.DuchenneConnect.org

