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

1. Foundations promote PatientCrossroads to act as trusted gatekeeper to patients
2. Patients register and complete patient profile
3. Counselors answer questions about trials and testing
4. Complete clinical questionnaires
5. Labs post testing results
6. Searches for qualified patients & to further disease research
7. Identifies candidates for recruitment

Foundations

Industry

DuchenneConnect

Clinicians

Counselors

Testing Labs
DuchenneConnect Process

- Genetic Counselor / Coordinator
- DMD patients & families
- Clinicians
- Test Lab

NCBI - TREAT-NMD Neuromuscular Network
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
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
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
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
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
Profile Questions: Participation in Global Registries

TREAT-NMD & NCBI

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

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

Provider registration
- Provide contact information
- Community affiliation
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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**

**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

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

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

Languages
- Spanish
- Additional languages in development
### Reporting - Over 60 Countries

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(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