MYOTONIC DYSTROPHY FOUNDATION

Care and a Cure

2018
MDF ANNUAL CONFERENCE
September 14-15, 2018
Nashville, TN

ACCELERATING THE SEARCH FOR THERAPIES: WHAT'S HAPPENING, WHAT'S NEXT

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MYOTONIC DYSTROPHY FOUNDATION

Care and a Cure

To enhance the quality of life of people living with myotonic dystrophy (DM) and advance research focused on treatments and a cure

MYOTONIC DYSTROPHY FOUNDATION

CARE & A CURE

WEBSITE & NEWSLETTER

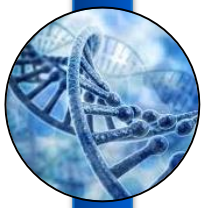
WARMLINE

TOOLKITS

ANNUAL CONFERENCE

CARE GUIDELINES

BASIC RESEARCH

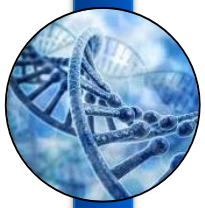


TRANSLATIONAL RESEARCH

CLINICAL RESEARCH

REGULATORY

PAYORS & ACCESS & ADVOCACY



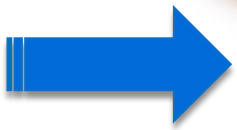
BASIC RESEARCH

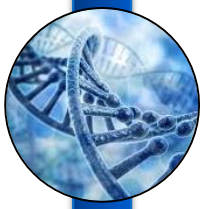
TRANSLATIONAL RESEARCH

CLINICAL RESEARCH and TRIALS

REGULATORY

PAYORS & ACCESS & ADVOCACY





BASIC RESEARCH

Laboratory experiments to understand basic biology

TRANSLATIONAL RESEARCH

Use information from basic research to create tools or techniques to support and inform drug development

CLINICAL RESEARCH and TRIALS

Research on normal healthy volunteers or patients

REGULATORY

Government bodies that regulate drug development

PAYORS & ACCESS & ADVOCACY

Government agencies funding DM research; insurance and reimbursement stake-holders

Our Goal is to Accelerate the Development of Treatments and a Cure for Myotonic Dystrophy

Continue to eliminate or lower barriers to therapy development

Support researchers and companies already committed to DM

Increase number of companies working on myotonic dystrophy

Scientific Advisory Committee

Other Patient Advocacy Organizations

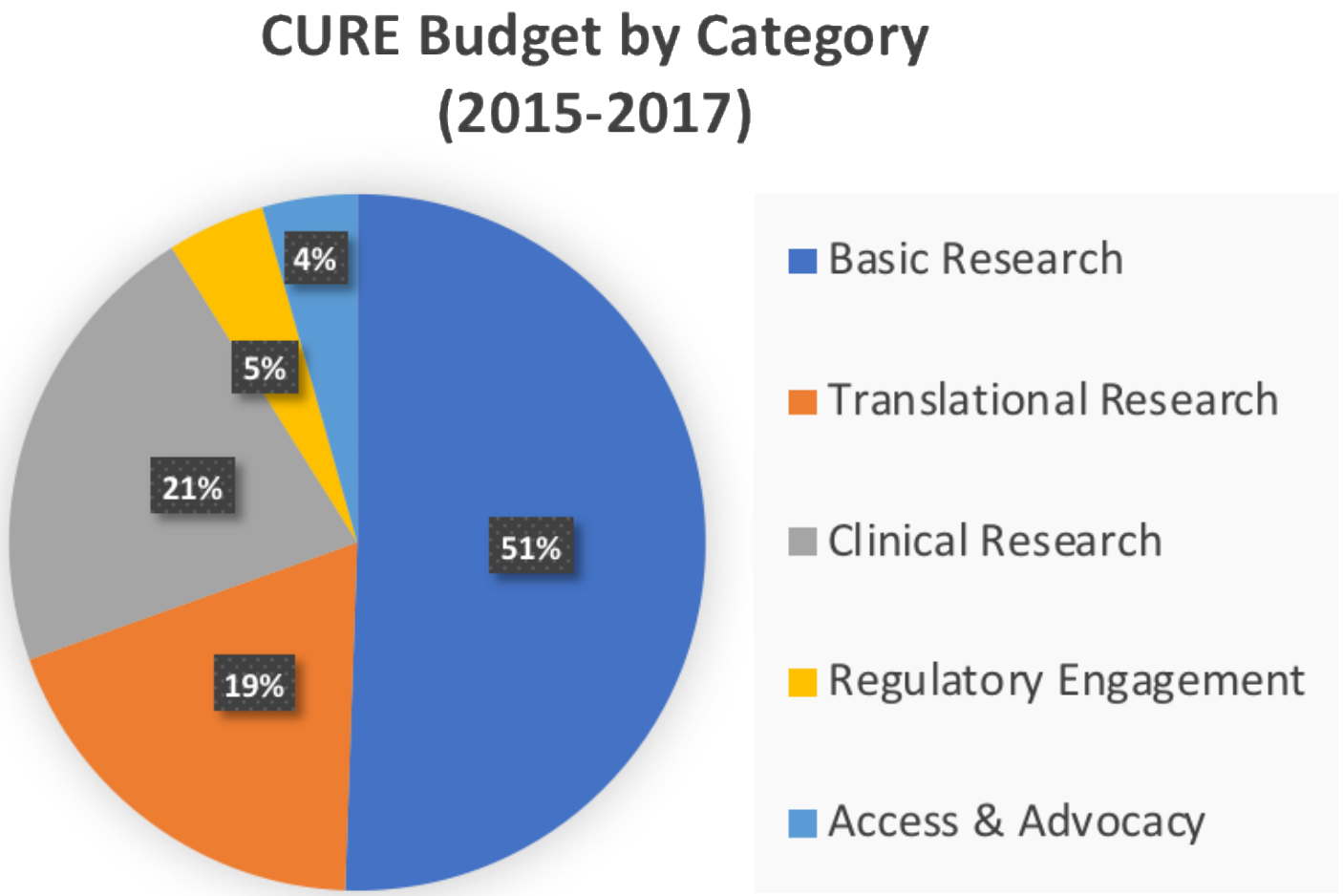
Industry Partners

Researchers

Patients

Regulatory Bodies (e.g. FDA)

Three Year Cure Research Plan Launched in 2015 ($5M)

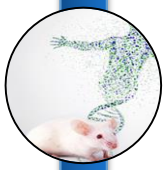


MDF Research Programs Target All Aspects of the Drug Development Pathway

BASIC RESEARCH

- 19 Post-doctoral and Pre-doctoral Fellows Funded
- Population-based prevalence study

TRANSLATIONAL RESEARCH



- Development of muscle MRI as a potential biomarker
- Development of RNA slicing in urine as potential biomarker
- iPS cell-lines for DM1 and DM2
- New Tg BAC Mouse model

CLINICAL RESEARCH

- DMCRN 500 Patient Natural History Study (US)
- PHENO-DM1 Natural History Study (UK)
- International Muscle Endpoint SOP consensus developed
- Myotonic Dystrophy Family Registry

REGULATORY

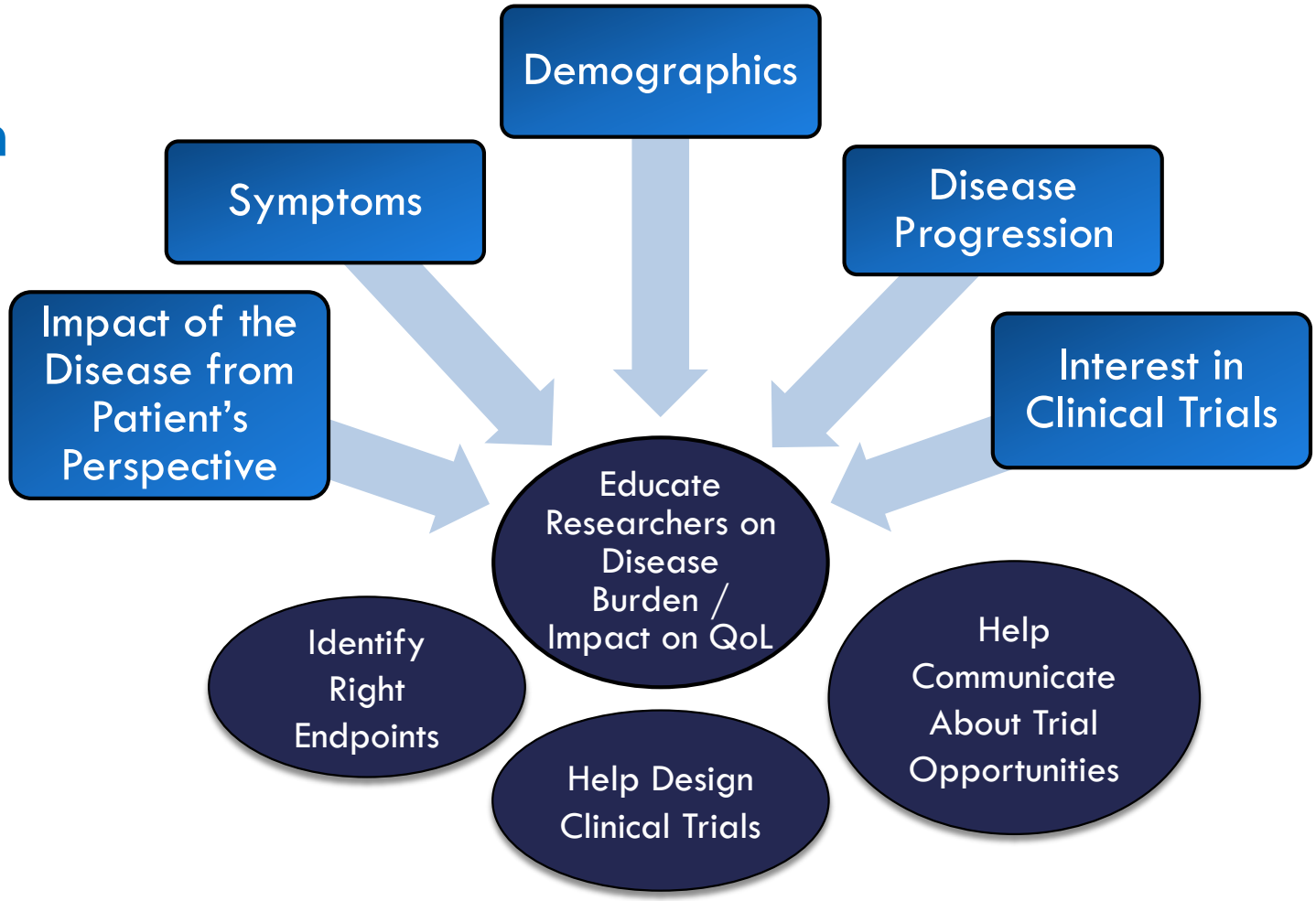
- CNS Patient voice workshop
- MDF FDA Workshop focused on DM
- PFDD Workshop; Voice of the Patient Report
- FDA Educational meetings

PAYORS & ACCESS & ADVOCACY

- Burden of disease analysis
- MDF active participant on Muscular Dystrophy Coordinating Committee
- DM added to DoD Funded Diseases (PRMRP)
- CDM added to SSA list for reimbursement

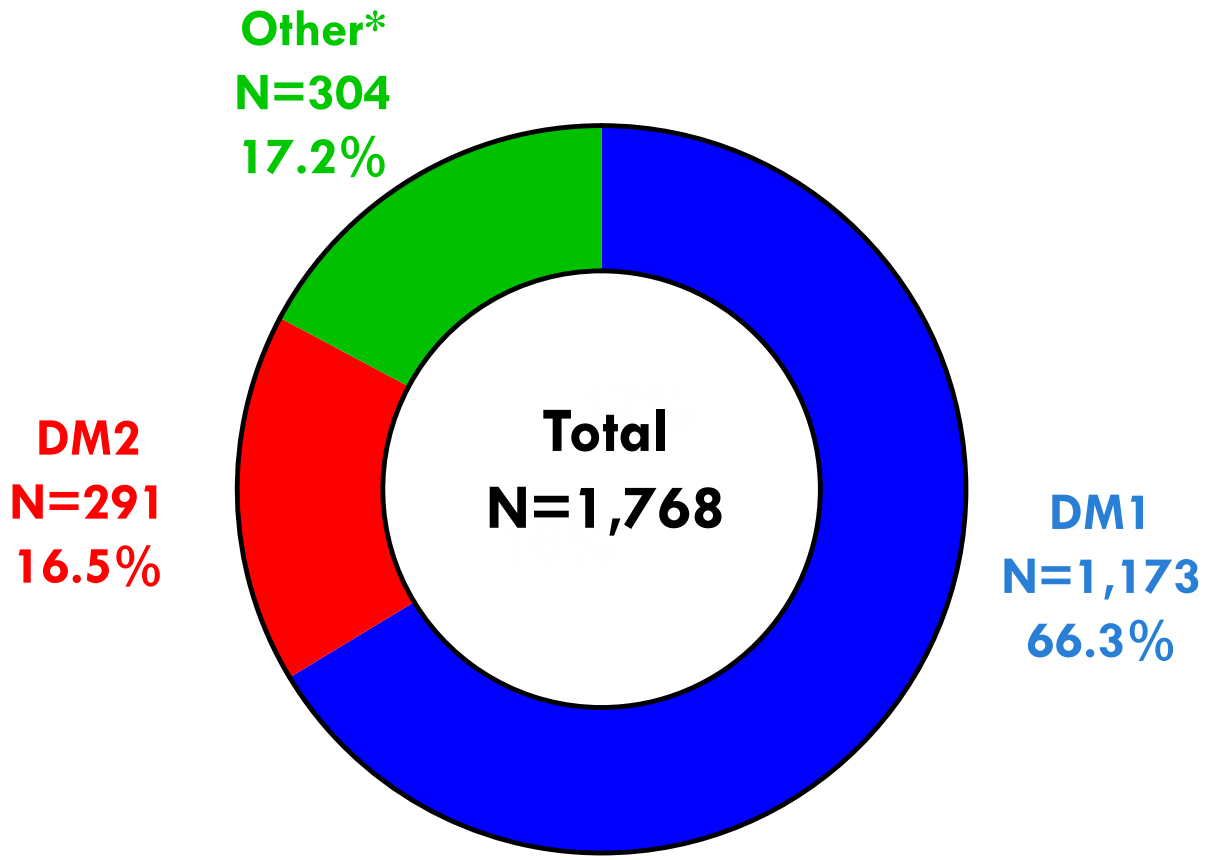
Patient Registries are Important!

Type of Information Collected



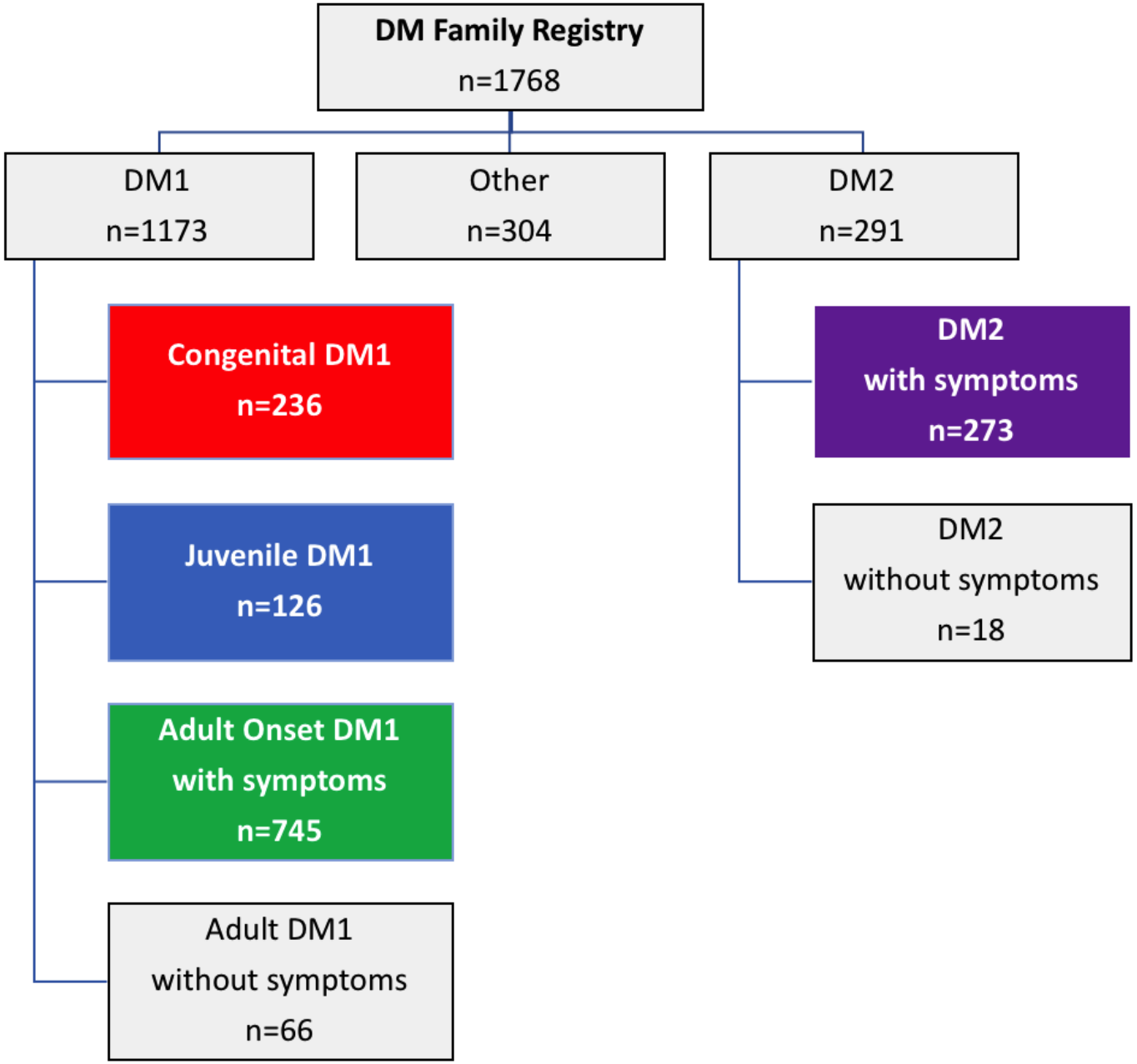
What It Is Used For

Myotonic Dystrophy Family Registry



(*) Includes subjects that selected "Other" or "I don't know" or left the question blank

Data cut July 22, 2018


DM Family Registry
n=1768
DM1
n=1173
Other
n=304
DM2
n=291
Congenital DM1
n=236
Juvenile DM1
n=126
Adult Onset DM1
with symptoms
n=745
Adult DM1
without symptoms
n=66
DM2
with symptoms
n=273
DM2
without symptoms
n=18

Demographics of Patients in MDF Registry






44 yr
Average Age

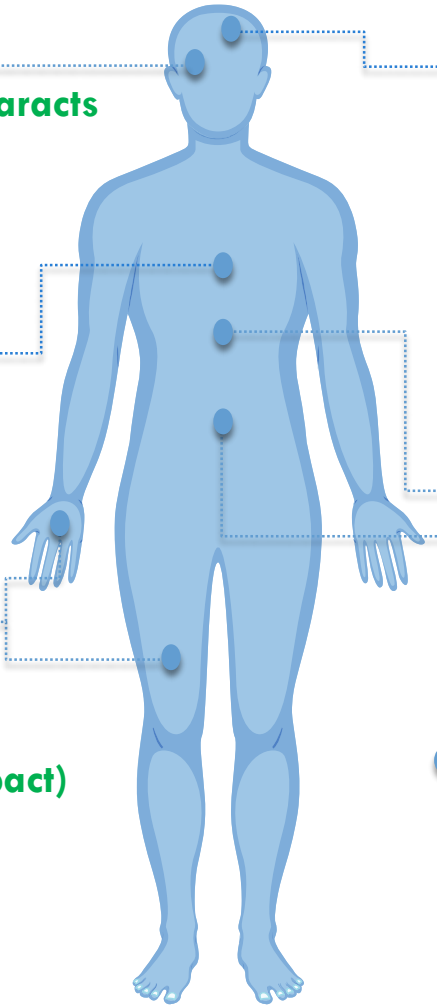
~90%
Identify as White

~1:1
Male to Female Ratio

~45%
First Person in Family with Diagnosis

~76%
Born in US; Over 60 other Countries Represented

MDF Registry Collects Prevalence of Multiple Disease Symptoms



Eye
- Vision trouble due to cataracts
- Cataract surgery

Central Nervous System
- Daytime sleepiness
- Other sleep issues
- Fatigue (negative impact)
- Pain (negative impact)
- Psychiatric / emotional disorders

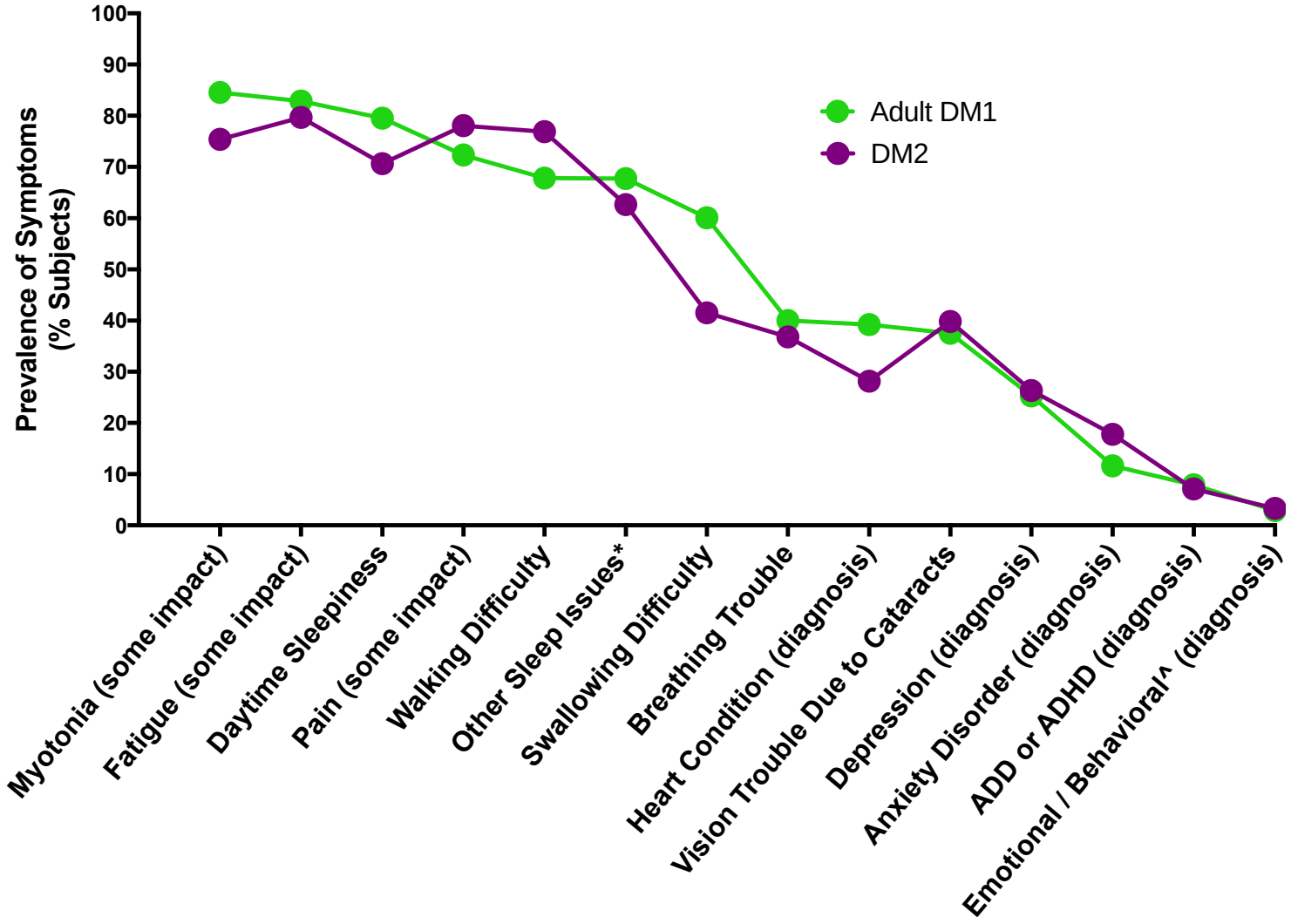
Cardiovascular System
- Heart condition diagnosis
- Pacemaker or ICD

Respiratory System
- Breathing difficulty
- Use of ventilation device

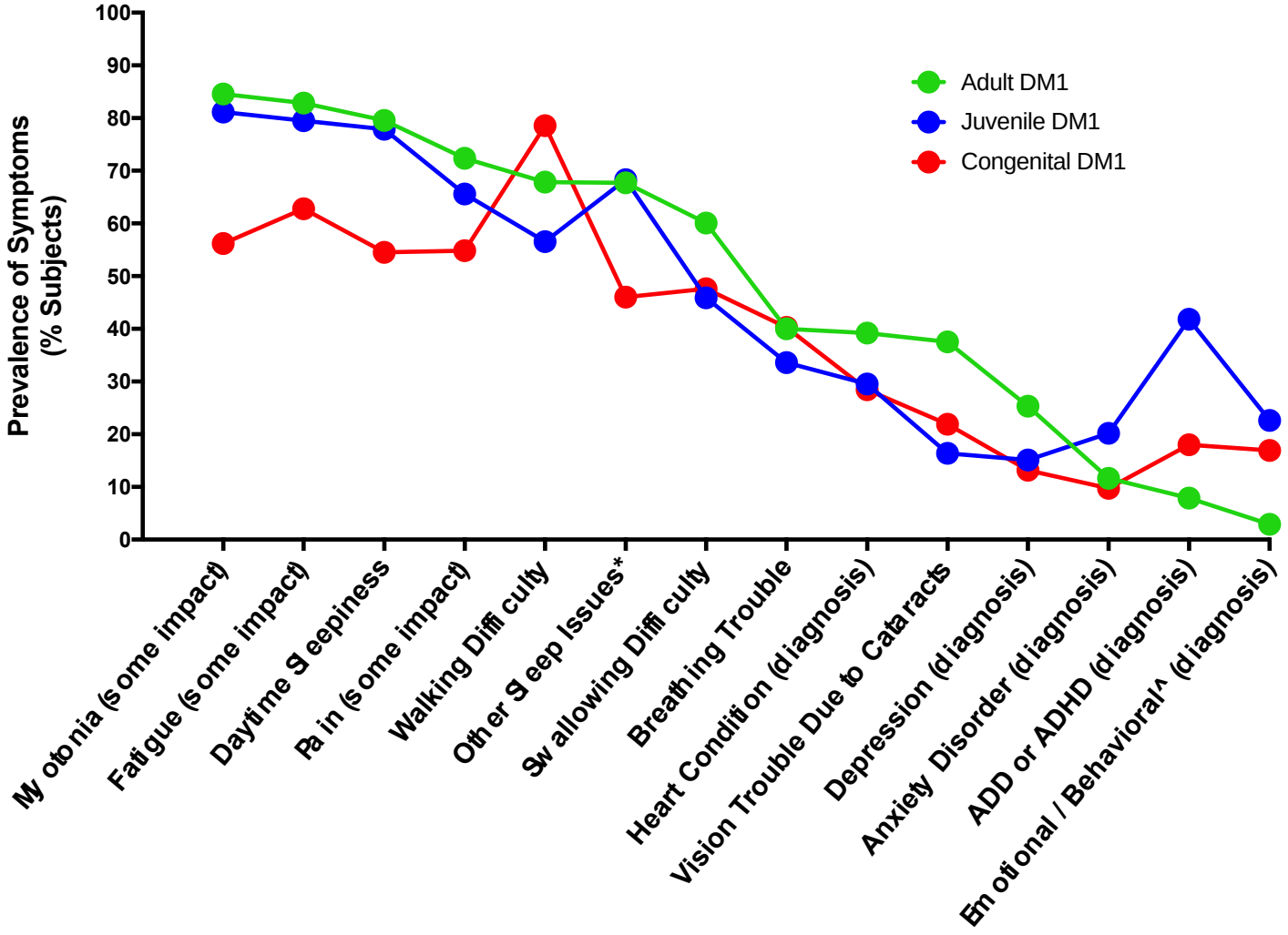
Skeletal Muscle
- Walking difficulty
- Use of orthopedic aids
- Myotonia (negative impact)

Gastrointestinal Tract
- Swallowing difficulty
- Use of gastric/nasal feeding tube

Prevalence of Symptoms (Adult DM1 vs DM2)



Prevalence of Symptoms (DM1 Subtypes)

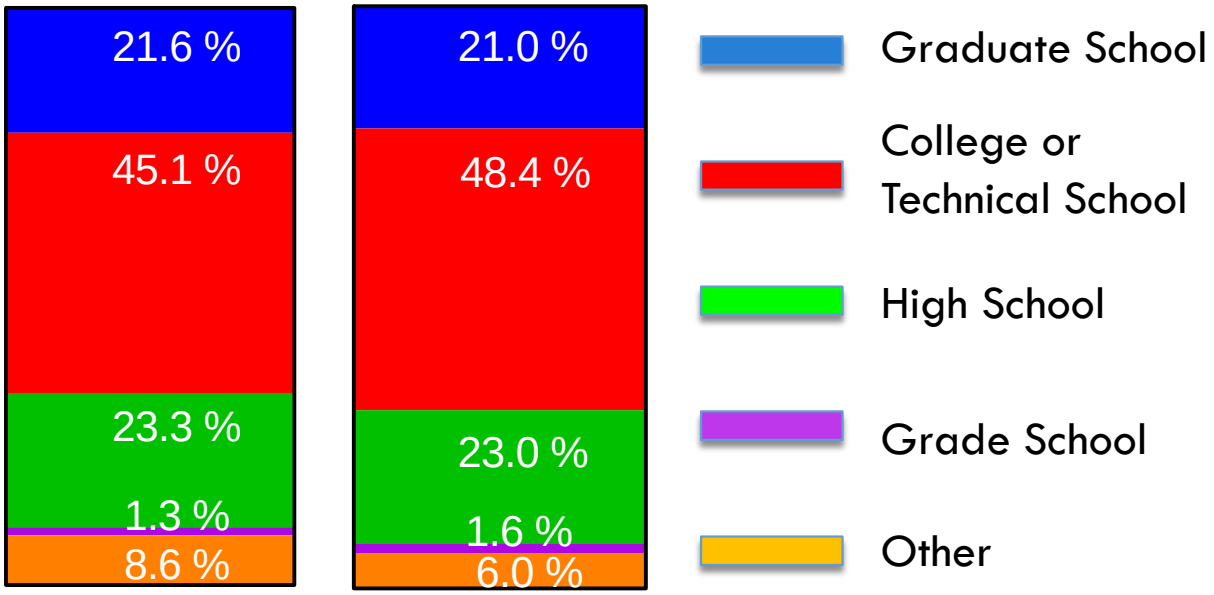


Significant Use of Orthopedic Devices or Physical Therapy Observed in All Groups

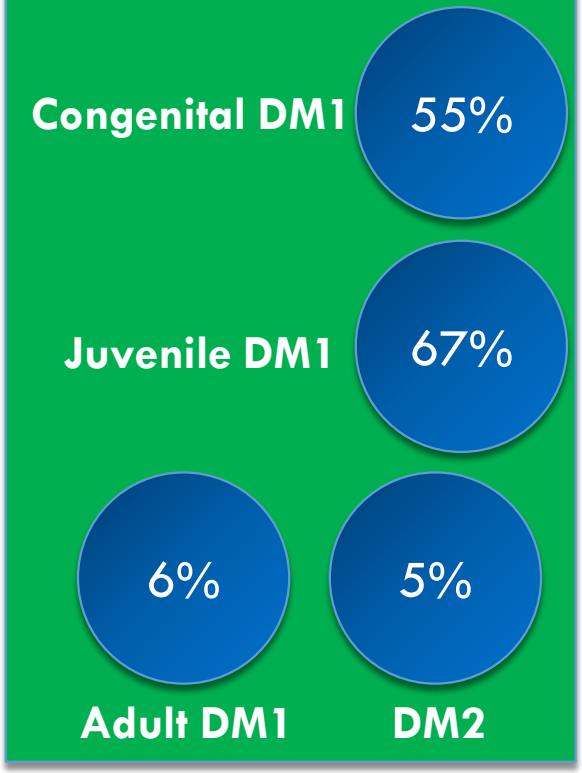
	Congenital DM1 (n=211)	Juvenile DM1 (n=119)	Adult Onset DM1 (n=689)	DM2 (n=250)
Use at Least One Orthopedic Device or Physical Therapy	68%	38%	48%	50%
Physical Therapy	36%	10%	12%	17%
Orthotics	22%	11%	7%	6%
Ankle/Leg Braces	38%	13%	12%	8%
Cane	7%	8%	21%	26%
Walker	9%	3%	8%	15%
Wheelchair	23%	11%	13%	17%

Education Status and Use of IEP Program

Highest Level of Education Completed



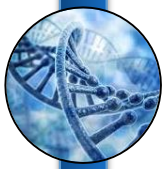
Used An Individualized Education Program (IEP) in School



Other Includes the following answers: I don't have any formal education, Not applicable-Participant is an infant or child, I don't know, Other-open text

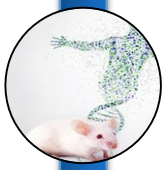
What's Next?
Plans for the Next 3-Year Initiative

BASIC RESEARCH



- Post- & Pre-doctoral Fellows
- Genome Editing Program

TRANSLATIONAL RESEARCH



- Mouse Drug Testing Facility
- Biomarker Development
- iPS Cell-Line Characterization
- Tg Mouse Model SOPs

CLINICAL RESEARCH

- Expansion of Patient Registry
- CNS Natural History Study

VENTURE PHILANTHROPY PROGRAM

REGULATORY

- Continue FDA and EMA Interactions and Educational Meetings

PAYORS & ACCESS & ADVOCACY

- Continue to lobby for PRMRP Funding each year
- MDF Active Participant on Muscular Dystrophy Coordinating Committee

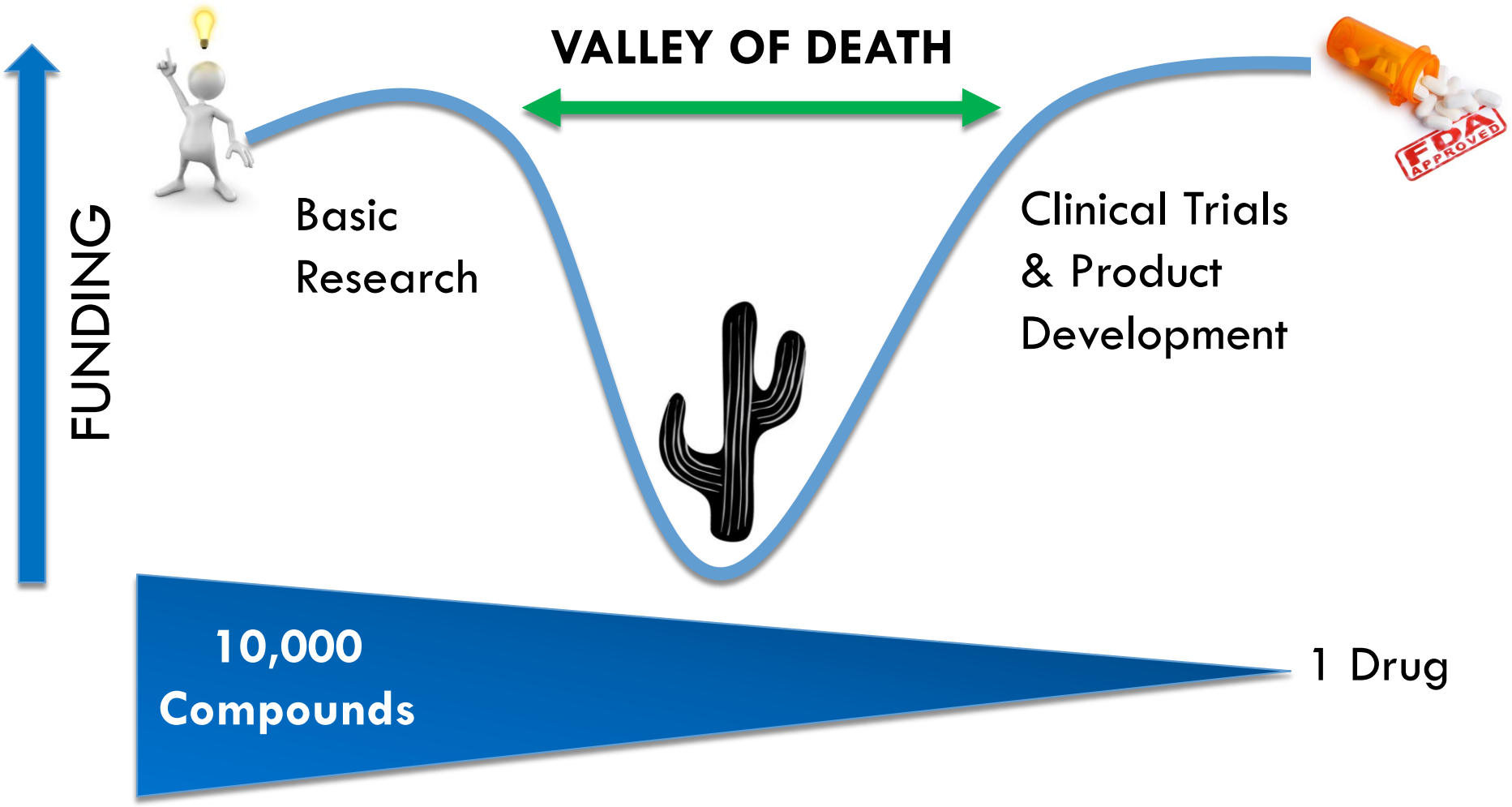
What is Genome Editing?

- Way to precisely make changes to the DNA of a cell or organism
  - Cut out pieces of DNA
  - Add pieces of DNA
  - Change sequence of DNA
- Genome editing can potentially be used to treat myotonic dystrophy
  - Cut out the repeat sequence
  - Delete the gene
- Much work is still needed to evolve the technology before it is transformed into an effective therapy for DM

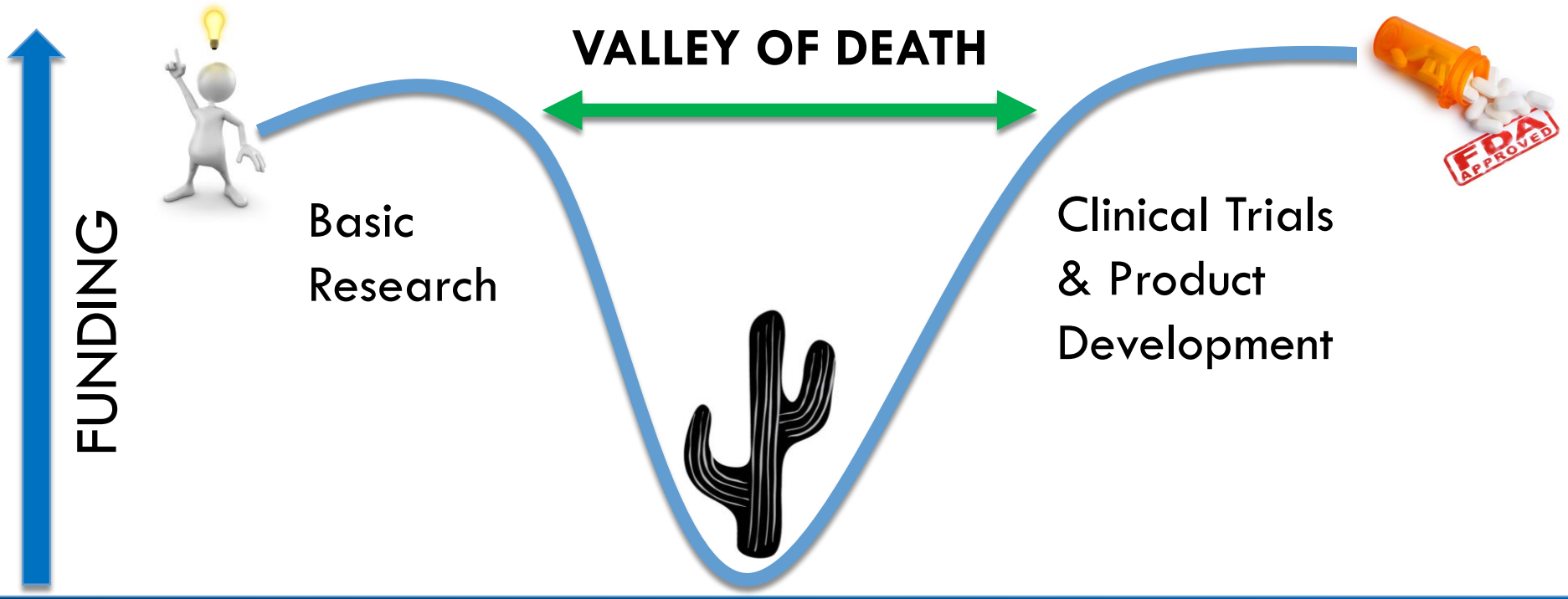
MDF's Genome Editing Initiative to Accelerate Development for DM

- Workshop held on April 17th, 2017 with 14 experts from universities, NIH, FDA, MDF and donors
- Request for Applications was released July 27th 2018
- Fund 2 awards of up to $250,000 to evaluate genome editing strategies for DM1 that target the DMPK gene

Venture Philanthropy Helps Promising New Therapies Cross the Valley of Death



Venture Philanthropy Helps Promising New Therapies Cross the Valley of Death



Make Strategic Investments in For-Profit Companies (Small Biotech, Early Stage Drug Development)

- Prioritize Myotonic Dystrophy
- Bring new companies into DM drug development
- Transition promising drugs across the 'valley of death'

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