

**The Central Database of Czech Duchenne/Becker Muscular Dystrophy Patients
The National DMD/BMD Registry**

Prezentace technologických řešení registru
Petr Brabec



The National DMD/BMD Registry





The National DMD/BMD Registry

ABOUT IBA MU

About IBA



Academic institute

- > Established in 2001
- > 45 employees
- > 65 scientific projects



Structure

- > Data analysis
- > Clinical trials
- > Environmental informatics
- > SW development & ITC

Research and teaching

- > Computational biology
- > Ecological risk assessment
- > Human risk assessment
- > Evidence-based medicine

Certification



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- **ISO 20000** (IT Service Management Standard)
 - Completed – August 2008
 - Implemented ITIL (IT Infrastructure Library)
- **ISO 9001** (Quality management systems)
 - In progress – March 2009
- **ISO 27000** (information security management system (ISMS))
 - In progress – June 2009



PDCA – basic idea



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Plan:

- establish the objectives and processes necessary to deliver results in accordance with customer requirements and the organization's policies;

Do:

- implement the processes;

Check:

- monitor and measure processes and services against policies, objectives and requirements and report the results;

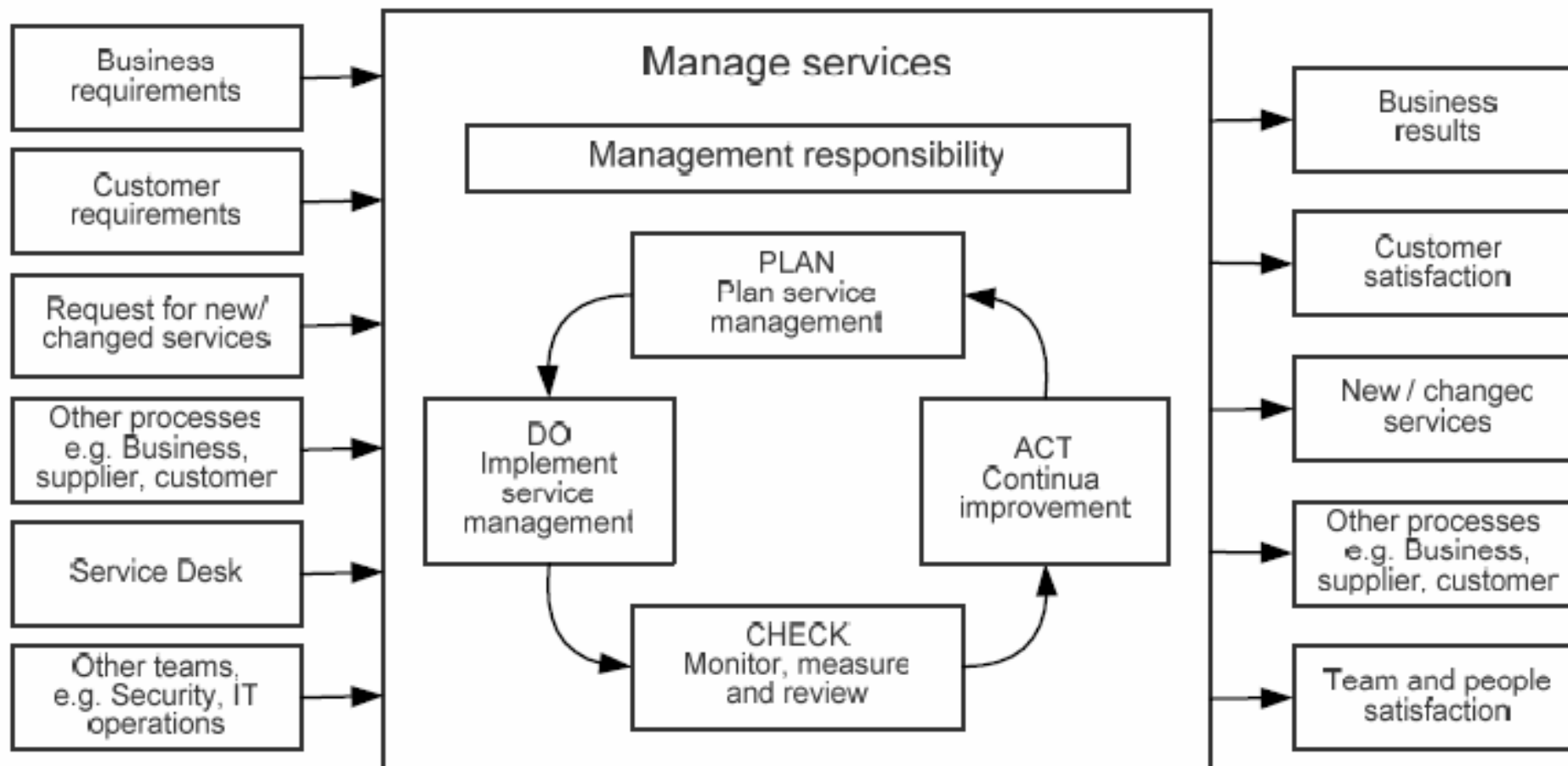
Act:

- Take actions to continually improve process performance.

Plan-Do-Check-Act methodology for service management processes



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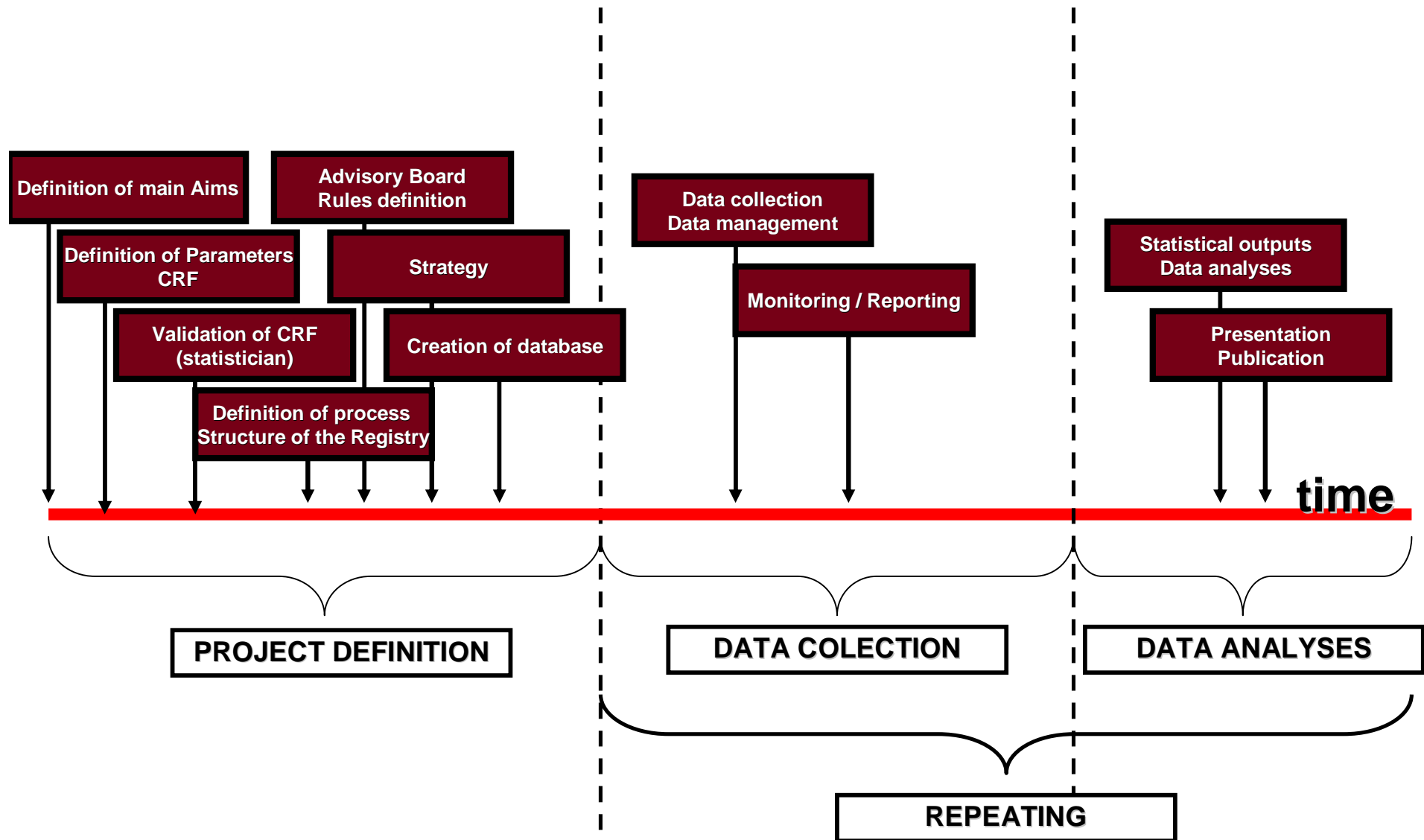
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ABOUT REGISTRY

Life cycle of Registry Project



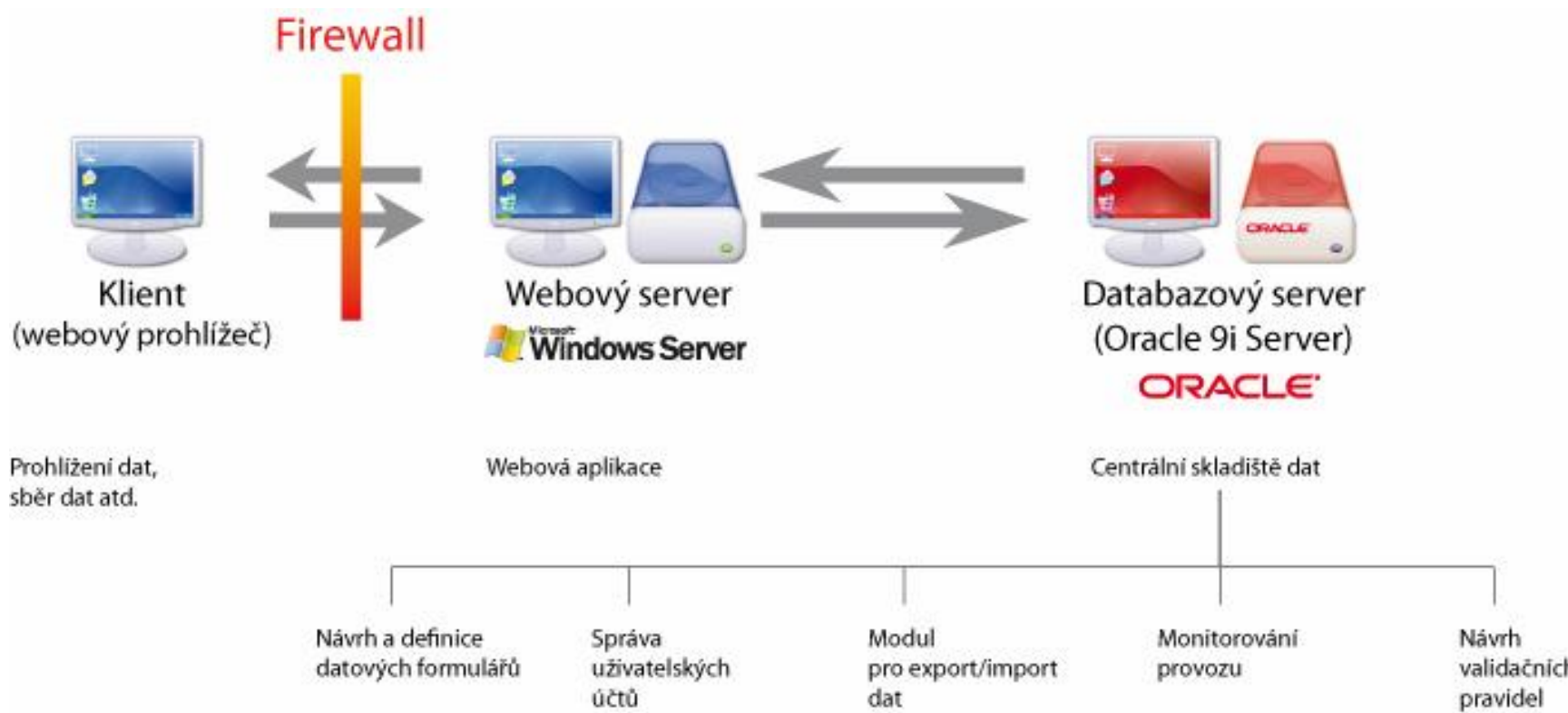
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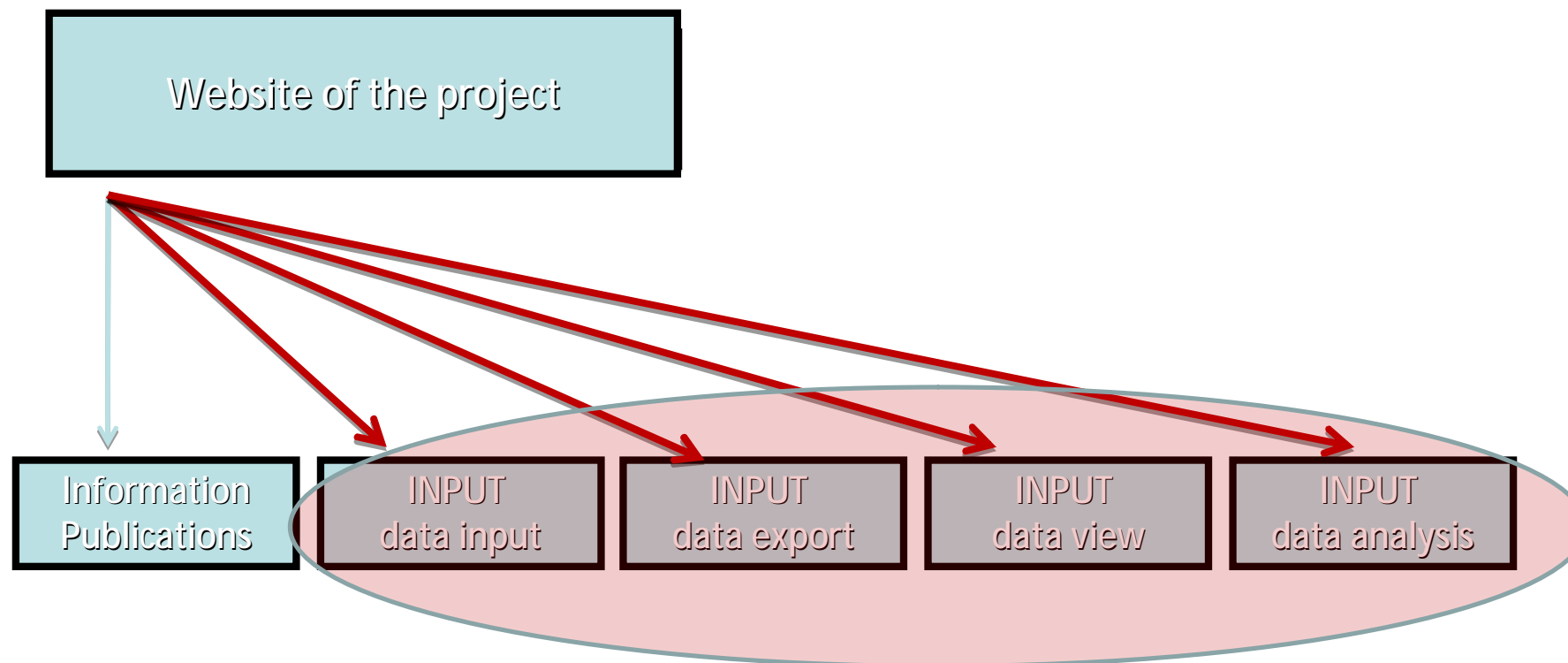


System



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General information, basic outputs for public

Applications that are accessible only for users with password. Accesses and rights for individual applications and data are assigned and controlled by register administrator.



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ABOUT REGISTER DMD/BMD

1. Webový informační systém Svalová dystrofie

Pro vstup do databáze použijte Internet Explorer

Vstupní adresa:

<http://dystrofie.registry.cz>



The Central Database of Czech and Slovak Duchenne/Becker Muscular Dystrophy Patients The International DMD/BMD Registry

Project | Data Summary | Muscular Dystrophy | Technology | Centers | Download | About us | Helpdesk | Entry to Registry

Project

In 2004 work started on the DMD/BMD patients database/registry. The original aim of the database was to effectively integrate Czech patients to international clinical studies. International cooperation and active searching for patients on an international level is necessary because suitable patients matching to the particular clinical and genetic criteria are insufficient in single countries. In the case of finding effective therapeutic methods the central database will speed up selection and indication of appropriate patients.

This registry provides a national database of DMD/BMD patients for medical experts and researchers to help accelerate the development and delivery of new treatments for Duchenne and Becker muscular dystrophy.

The registry of patients with DMB/BMD in the Czech Republic is now available online. This database is available to registered specialist and patients, who have access to their personal data. The registry provides the possibility to collect, view or search for data regarding patients' phenotype and genotype profiles and other medical information for the purpose of future international and intervention studies and to establish a therapeutic strategy. The substance of this project is to collect all the necessary information possible about Czech patients who have this serious hereditary disease.



The project is sponsored by Parent Project Czech Republic civil association.

We are looking for sponsors. Please contact us by email:
brabec@iba.muni.cz

The registry of Czech and Slovak Duchenne/Becker muscular dystrophy patients is a result of cooperation between the Institute of Biostatistics and Analyses, Masaryk University (technical realization, data analyses), Parent Project Czech Republic civil association (project initiation, financial support) and University Hospital Brno (scientific advisor).



Project goals:

- Describe Czech and Slovak DMD/BMD patients
- Recruit patients for international clinical studies
- Describe prevalence of specific types of mutations in the dystrophin gene
- Speed up selection of appropriate patients in the case of effective treatment
- Encourage research into DMD/BMD
- Improve understanding of DMD/BMD
- Inform participants about new research/latest developments in the area of DMD/BMD treatment
- Publish new results of the research

Základní vlastnosti

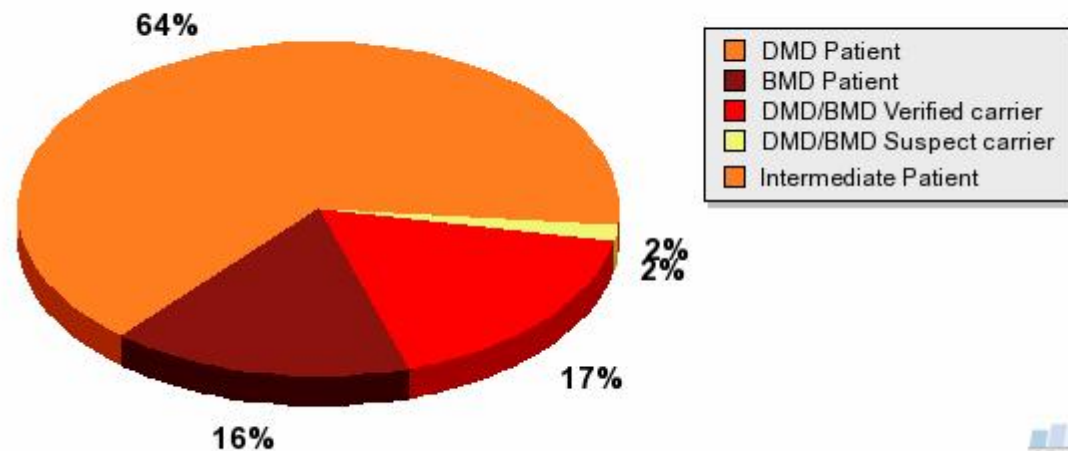
- Prohlížení statistik
- Detailní procházení záznamů pro registrované uživatele
- Možnost prohlížení záznamu konkrétního pacienta
- Možnost požádat elektronickou cestou o dokumentaci pacienta
- Mapa center
- Popis technologií
- Download materiálů
- Zadávání dat pro lékaře
- Zadávání dat pro pacienty

Data summary



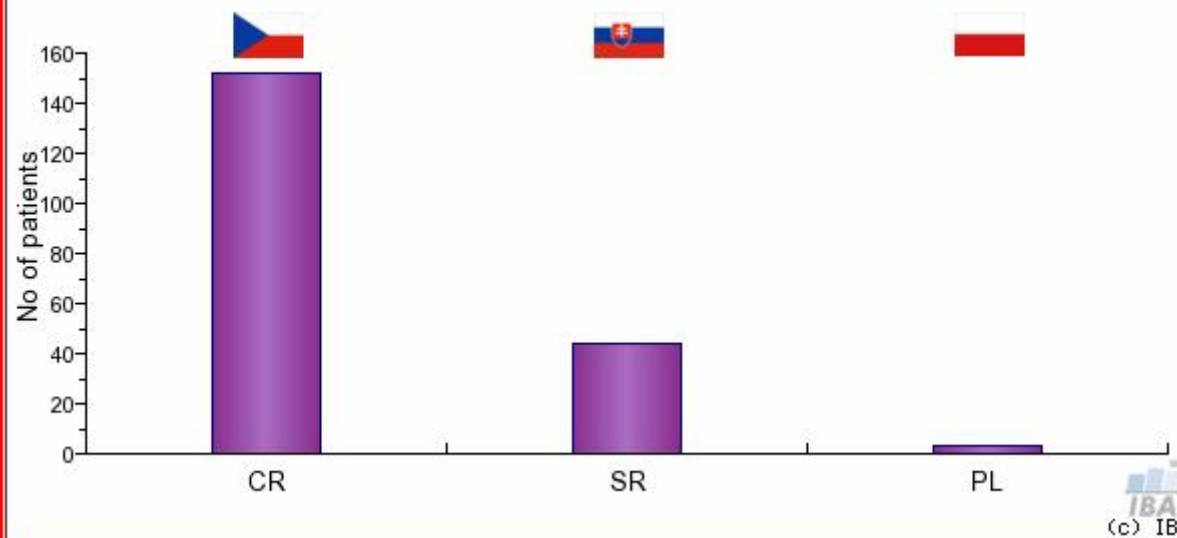
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Patients (n=198)



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Patients (n=199)

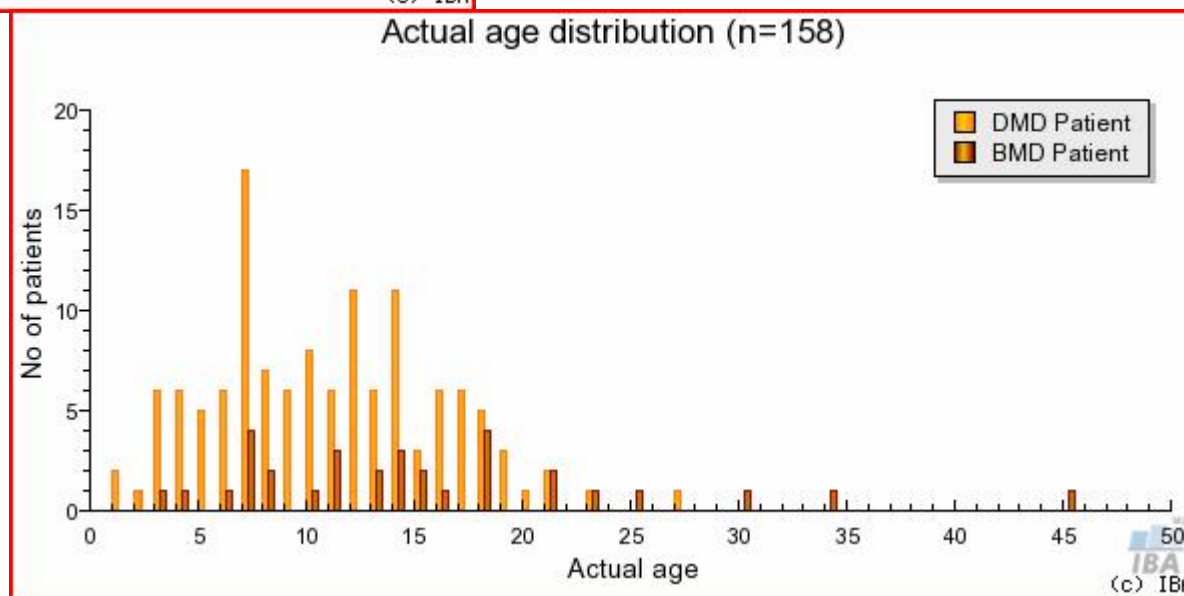
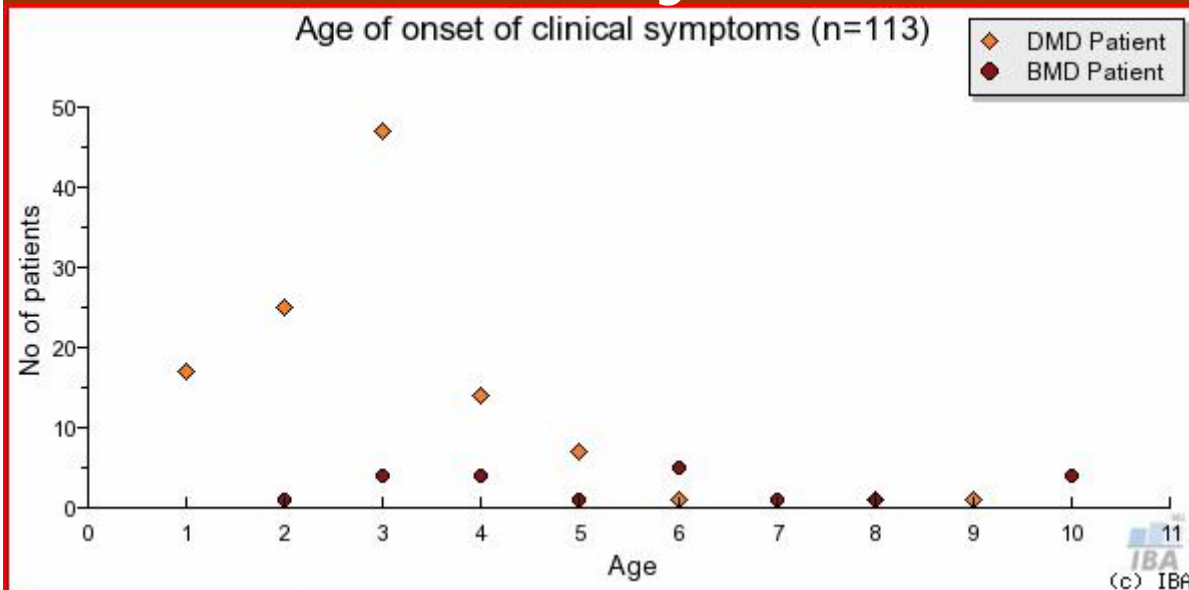


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Data summary



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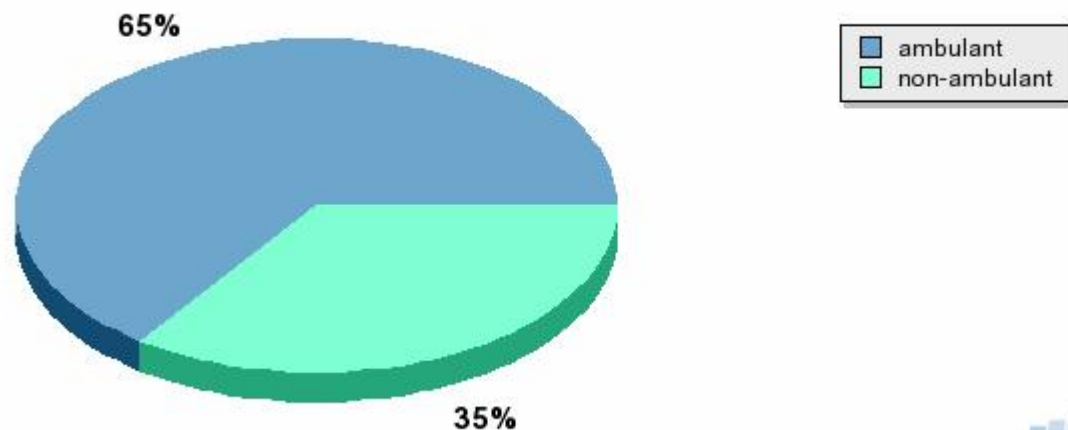


Data summary



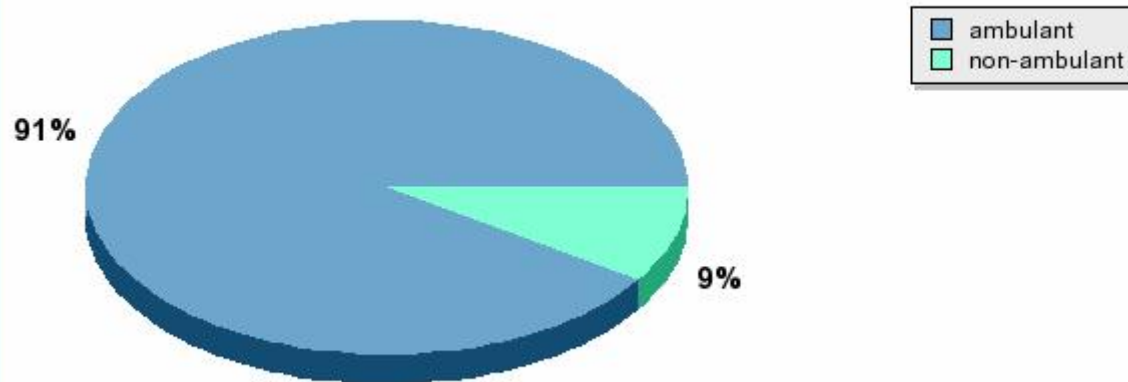
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DMD Patients - functional state (n=126)



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BMD Patients - functional state (n=32)



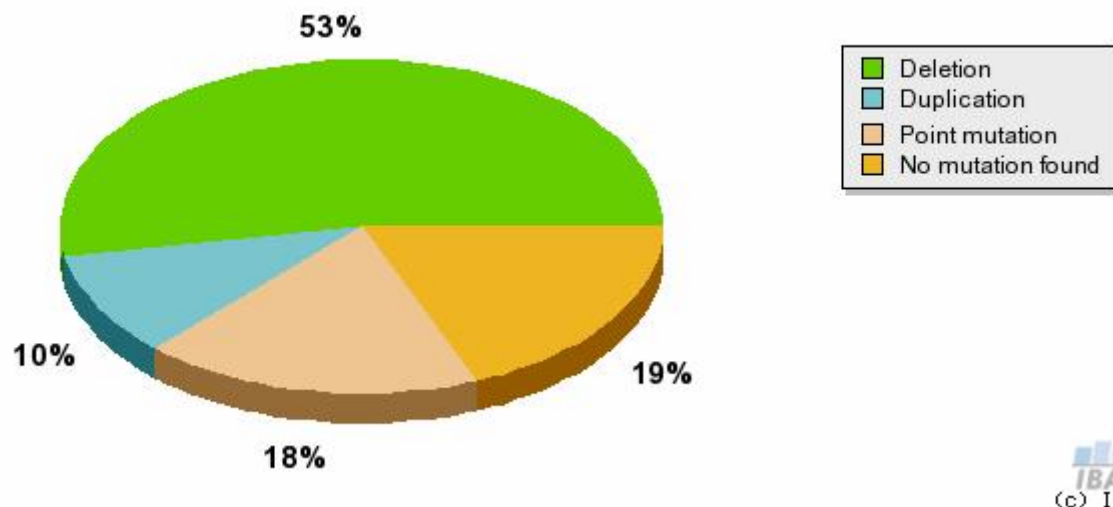
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Data summary

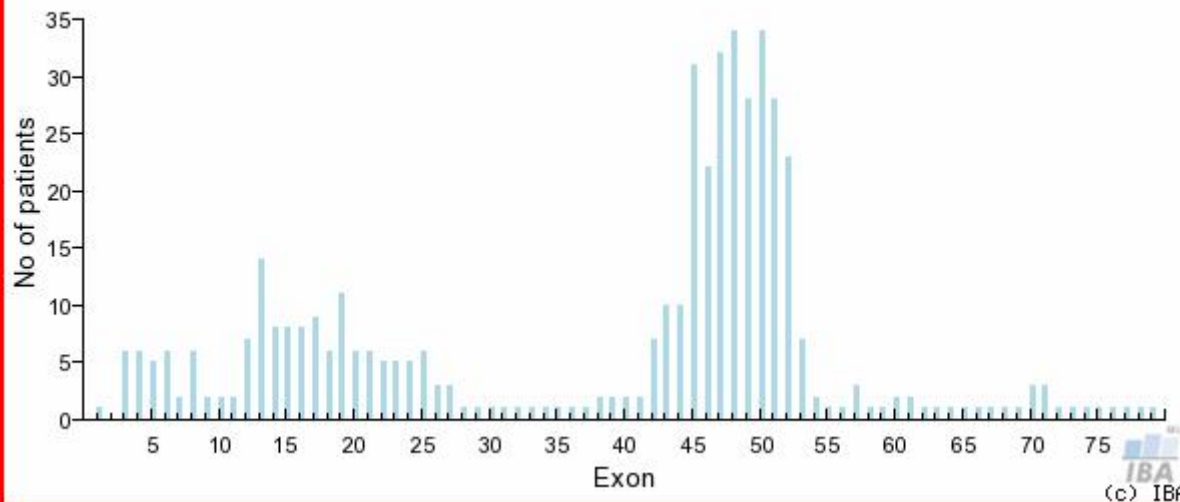


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Distribution of Mutations (n=197)



Deletion of Exons (n=467)



Data summary



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Data summary - more information

User: brabec [Logout]

Choose parameters:

Status code:

Still ambulatory:

Type of mutation:

RT-PCR Results:

Deletion of exons:

01 02 03 04 05 06 07 08 09 10 11 12 13 14 15 16 17 18 19 20 21 22 23 24 25
 26 27 28 29 30 31 32 33 34 35 36 37 38 39 40 41 42 43 44 45 46 47 48 49 50
 51 52 53 54 55 56 57 58 59 60 61 62 63 64 65 66 67 68 69 70 71 72 73 74 75
 76 77 78 79

- all of the selected deletions
 at least one of the selected deletions

Final diagnosis:

Submit

Reset

Selected patients (N = 199)

Patient ID <small>(click for details, check if interested)</small>	Gower's sing	Calf pseudo-hypertrophy	Contractures	Scolio-sis	Cardio-myophathy	Respiratory insuff.	Motor functions	Hist. of malignant hyper-thermia	Specification	Score	Status code	Still ambula-tory
0112296834 <input type="checkbox"/>	POSITIVE	YES	MILD	NO	NO	NO	ABLE TO WALK > 100M WITHOUT SUPPORT	No		60	Clinically symptomatic patient	ambula
92 <input type="checkbox"/>	POSITIVE	YES	SEVERE	YES	NO	NO	WHEEL CHAIR BOUND	No		30	Clinically symptomatic patient	non-am
MD-B-001 <input type="checkbox"/>	NEGATIVE	NO	NO	NO	NO	NO	NORMAL WALKING / RUNNING	Yes		100	Clinically still asymptomatic patient	ambula
MD-B-002 <input type="checkbox"/>	POSITIVE	YES	MILD	NO	NO	NO	WHEEL CHAIR BOUND	No		40	Clinically symptomatic patient	non-am

Data summary - more information

Patient MD-B-004

If you have serious interest in this patient: [Send message to Principal Investigator](#) [Back](#)

Parameter	Value
Basic Data	
Site	FN Brno - Klinika dětské neurologie
Patient ID	MD-B-004
Sex	M
Date of birth	2001-05-24
Country	CR
Phase	Diagnosis
Status Code	
Status Code	Clinically symptomatic patient
Clinical Data	
Date	2005-12-23
Age of onset of clinical data	1
Still ambulatory	ambulant
Non ambulatory since age	
Gower's sing	POSITIVE
Calf pseudohypertrophy	YES
Contractures	NO
Scoliosis	YES
Cardiomyopathy	NO
Respiratory insufficiency	NO
Motor functions	ABLE TO WALK < 100M WITHOUT SUPPORT
History of malignant hyperthermia	No
Specification	
Score	50
Biochemistry	
Date	2005-12-23
CK [uKAT/L]	363
LD [uKAT/L]	76
ALT [uKAT/L]	9
AST [uKAT/L]	8
CK [U/l]	
LD [U/l]	
ALT [U/l]	



Pro vstup do samotného systému Svalová dystrofie nejprve musíme zadat do políček „Login“ a „Heslo“ naše uživatelské jméno a heslo. Po stisknutí tlačítka „Login“ vstoupíme do další části systému.

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Please enter **login and password**:

Login:

Heslo:

Login

Login

Heslo

Login - stiskněte

IBA MU
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Masaryk University Brno
Kamenice 126/3, 625 00, Brno, Czech Republic

PARENT PROJECT
SVAZKA RODIČŮ
VYBRANĚ LÉČENÝCH
Parent Project
Supported by Parent Project, Czech Republic
www.parentproject.cz

Principal Investigator: Petr VONDRÁČEK, MD., PhD., pvondracek@fnbrno.cz
Data Manager: Petr BRABEC, MSc., brabec@iba.muni.cz
Developed by Institute of Biostatistics and Analyses, Masaryk University Brno, Czech Republic



Upozornění:

Po vstupu do systému, pokud v něm nebudeme 30 minut pracovat, nás systém sám odhlásí. Pro další práci je nutné provést opětovně přihlášení. Účelem této funkce je zabránit přístupu neoprávněné osoby do systému.



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Praktická ukázka řešení

<http://dystrophy.registry.cz>