DATA BUSINESS CHALLENGE

Group 3

A TEAM OF 4 **DATA SCIENTISTS** TO HELP YOU CREATE VALUE FROM YOUR DATA

Aicha BOKBOT
Data-scientist
Aicha.bokbot@hec.edu
06 63 93 41 64

Leon LEITAO
Data-scientist
leon.leitao@hec.edu
07 54 59 29 51

Pragya SINGH
Data-scientist
Pragya.singh@hec.edu
07 50 91 21 71

Corentin SENE
Data-scientist
corentin.sene@hec.edu
06 51 26 77 76

PROJECT PRESENTATION & OBJECTIVES

DATA ANALYSIS

OUR METHODOLOGY

MODELS & RESULTS

SUGGESTIONS, LIMITATIONS & NEXT STEPS

DATA SOLUTIONS TO AID MEDICAL PROFESSIONALS

KEY OBJECTIVES AND EXPECTED BENEFITS

PROJECT OBJECTIVES

- Extract and **structure** the **valuable information** from treatment journals
- Create a model that **identifies patients** who may suffer from a disease

BUSINESS IMPACT

- Develop early, **accurate diagnoses**, which lead to quicker treatment and mitigate the long-term damage caused by the disease
- **Reduce risk** of misdiagnoses

OUR APPROACH

- Develop **Personalize solutions** for each disease

DATA AVAILABLE

PATIENT INFO

ANAMNESTIC DATA

DIAGNOSIS

LAB RESULTS

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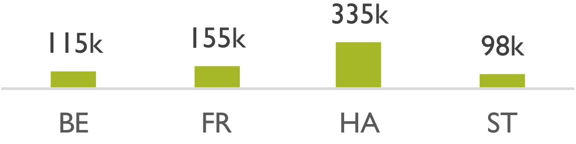
NUMEROUS DATA ON PATIENTS AND YET LIMITED DATA ON SOME DISEASES

Data analysis

OVERVIEW OF THE DATA

702 258 patients



on which we have a lot of data

on average

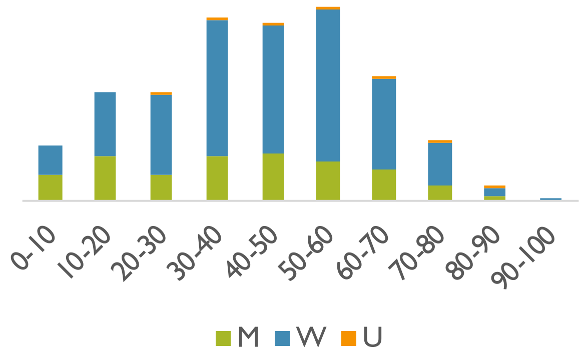
12 appointments
in 4 years
100 rows per patient

data from 1985 to 2020 on

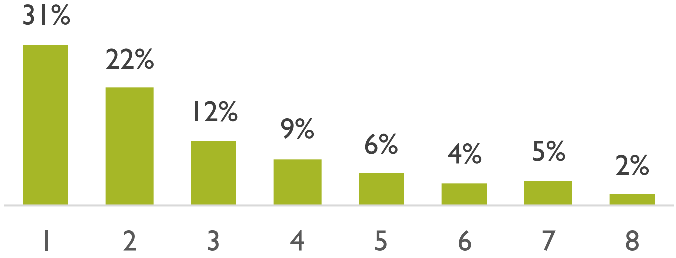
c. 6000 diseases

75% of them concern
less than 60 patients

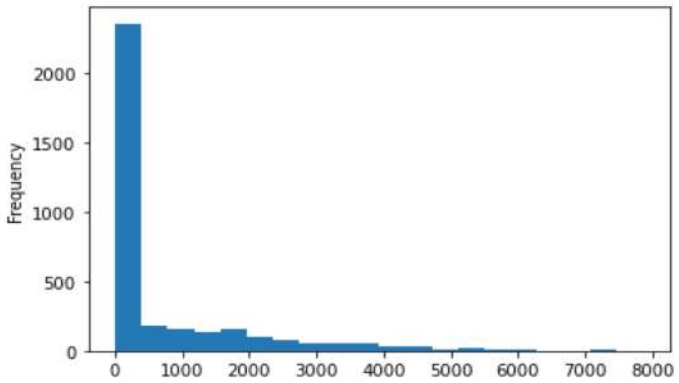
Age distribution by sex



Number of distinct diseases among diagnosed patients



Distribution of time between diagnosis and first visit (in days)



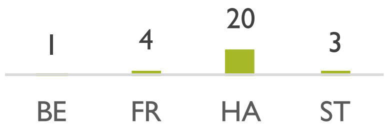
Data analysis

A **HETEROGENEOUS** NUMBER OF PATIENTS FOR EACH DISEASE

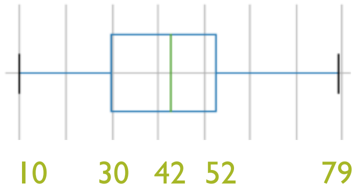
OVERVIEW OF THE 4 DISEASES

GAUCHER DISEASE – E75.22

28 patients

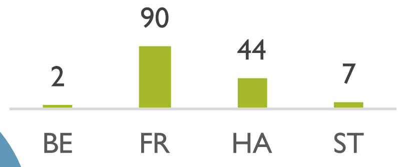


53% women
diagnosed at 43 y-o on avg

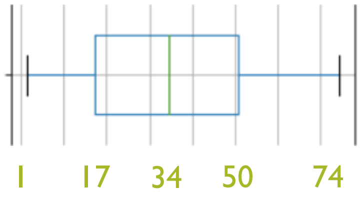


FAMILIAL HYPERCHOLESTEROLEMIA – E78.01

143 patients



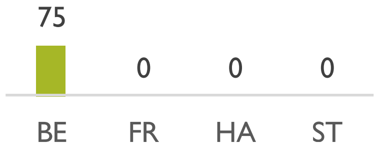
70% women
diagnosed at 34 y-o on avg



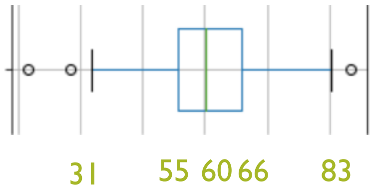
DISEASES

CHYLOMICRONEMIE – E78.3

75 patients

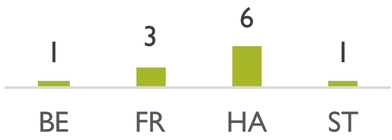


65% men
diagnosed at 60 y-o on avg

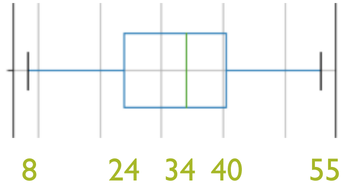


BETA OXYDATION DEFFECT – E71.3

11 patients



65% men
diagnosed at 31 y-o on avg



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THE TEXT COLUMN COMPLETES PROVIDES **VALUABLE INFORMATION** TO PROCESS

FEATURE EXTRACTION

PATIENT_HASH	ZENTRUM_ID	PATIENT_ID	PAT_GEBDATUM	PAT_GESCHLECHT	DATUM	TYP	TYP_EXT	TEXT	ICD10	SICHERHEIT
145858	FRA01	150256	07.07.07	W	27.11.17	Y		GLU=71; HS=2.9; GPT=15; GOT=32; GGT=9; AP=259;...	NaN	NaN
145858	FRA01	150256	07.07.07	W	27.11.17	A		Jessica wird uns zur Beurteilung der Körperhöh...	NaN	NaN
145858	FRA01	150256	07.07.07	W	15.11.18	Y		GLU=107 +; HS=3.6; GPT=14; GOT=33; GGT=10; AP=...	NaN	NaN
145858	FRA01	150256	07.07.07	W	10.01.19	*		Hypercholesterinämie	E78.0	G

Age

Average age of the patient (between the first and last visit)

Sex

Sex of the patient

Test results

Results of a selection of relevant tests

Symptoms

Does the patient show any relevant symptoms?

Co-morbidity

Does the patient have any other relevant diseases?

METHODOLOGY BASED ON THE SEARCH FOR **SYMPTOMS**, **TESTS** AND **CO-MORBIDITY**

OUR METHODOLOGY

Preprocessing

- **DATUM:** convert to Datetime (e.g."10.06.50", "10.06.90", "31.12.20")
- **PAT_GESCHLECHT**: convert to Datetime (e.g. "27.19.19")
- **ICD10**: treat missing values
- **SICHERHEIT**: treat missing values
- **TEXT**: interpret test results (from MCV=99.2 + to MCV = High)

For each of the four diseases:

1 – Research

Research on the disease to identify:

1. Symptoms: what are the common symptoms?
2. Tests: which tests enable to diagnose the disease?

2 – Pattern recognition in the data

Analyze the data of people diagnosed with the disease to identity:

1. Symptoms: what are the most recurring symptoms?
2. Tests: which are the most recurring tests with abnormal results?
3. Co-morbidity: which other other diseases do the patients also have?

3 – Feature creation

Build the features

1. Symptom
2. Test results
3. Co-morbidity

EXAMPLE : GAUCHER DISEASE

1 – Research

- **Reasons for referral:**
  - Splenomegaly
  - Hepatosplenomegaly
  - Bone Involvement
  - Chololithiasis
  - Thrombocytopenia
  - Pancytopenia
  - Leucopenia
  - Anemia
  - Member of patient family
- **Diagnosis**
  - Enzyme test called Beta-glucosidase leukocyte (BGL) test

2 – Pattern recognition in the data

- **Symptoms**
  - Fatigue
  - Bone pain
  - Splenomagaly
  - Thrombocytopenia
- **Co-Morbidity**
  - E55.9: Vitamine D defficiency
  - I10.90: Hypertension
  - D69.61: Thrombocytopenia
  - G93.3: Fatigue Syndrom
  - R16.1: Splenomegaly
- **Relevant tests**
  - High Osteocalcin (68%)
  - High Kappe Free Light chains (48%)
  - High Albumin (36%)
  - Low Thrombocytes (36%)
  - High Ferritin (32%)
  - Low Transferrin saturation (32%)
  - High DPD (32%)
  - High GGT (32%)
  - Low MCH (32%)
  - Low Hematocrit (32%)

Source: "Gaucher disease: Biochemical and molecular findings in 141 patients diagnosed in Greece", Molecular Genetics and Metabolism Reports, Volume 24, 2020

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ADABOOST ALGORITHM ALLOWS US TO OBTAIN A 96% ACCURACY

MODELING METHODOLOGY

Model intuition

Patient ID	Sex	Age	Test results	Symptoms	Co-morbidity	diagnosed
	…	…	…	…	…	1
	…	…	…	…	…	1
	…	…	…	…	…	0
	…	…	…	…	…	0
	…	…	…	…	…	0

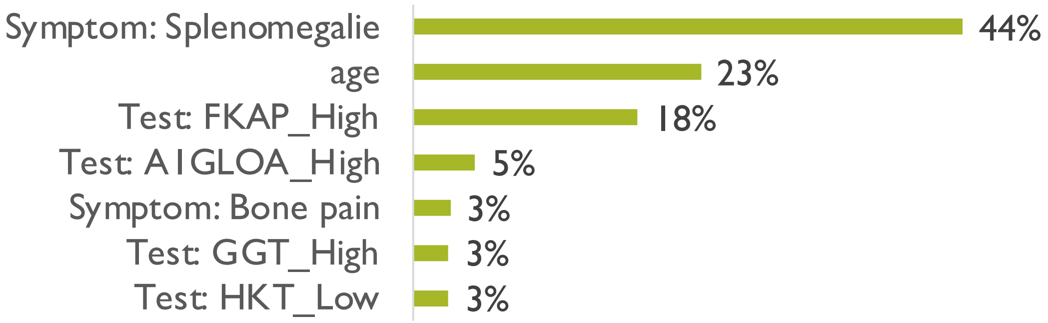
Best performing model

AdaBoost Classifier

Scores after hyper-parameter tuning and resampling :

- Classifies correctly 68% of positive patients
- Classifies correctly 99% of negative patients
- Accuracy: 96%

Feature Importance for Gaucher Disease

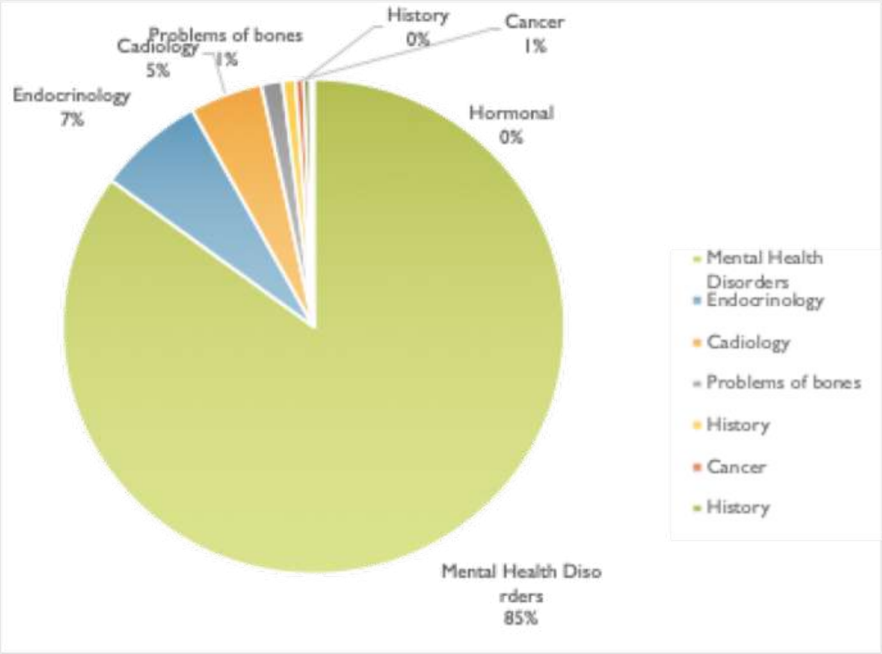


KEY FINDINGS FROM THE DOCTORS NOTES

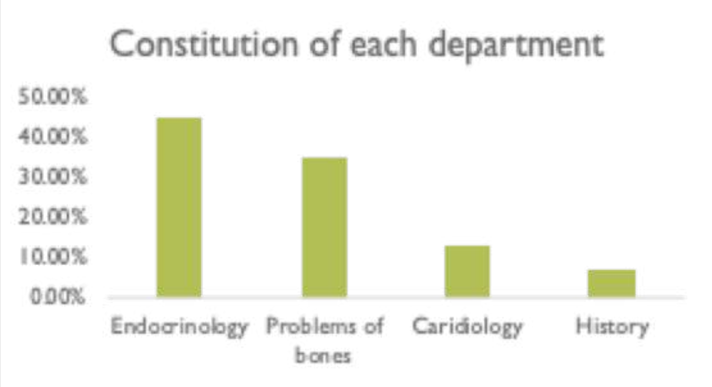
Methodology

EXAMPLE : ANAMENESTIC DATA

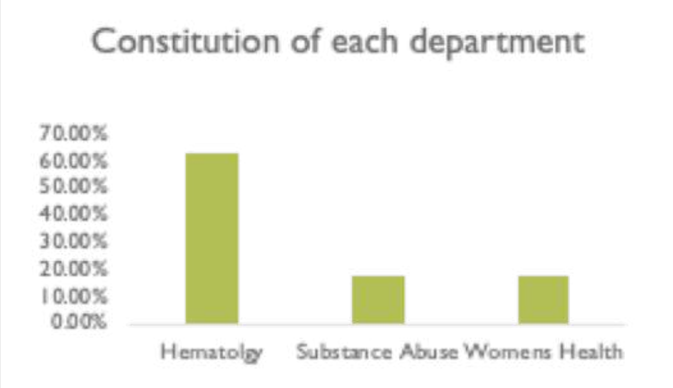
Most common categories amongst all patients



Familiare Hypercholesteramie



Gaucher



Most frequently occuring words	Departments assigned
Burned out,sleep,nocturna,therapi,weight loss	Mental Health Disorders
Wanting children, mammareduction, dysmenorrhoea, cycle monitoring	Women's reproductive health
Glucose tolerance, hypertoni, hormone replacement, sugar	Hormonal Disorders
Nicotine, alcohol, hypertonic nicotine, cancer, stage	Substance abuse
Hypothyroid, endocrino	Endocrinology
Personal history,social history,family anamnes	Personal History
Gonathros,hws syndrome,coxarthrosis,joint problem	Problem of bones

Key Ideas

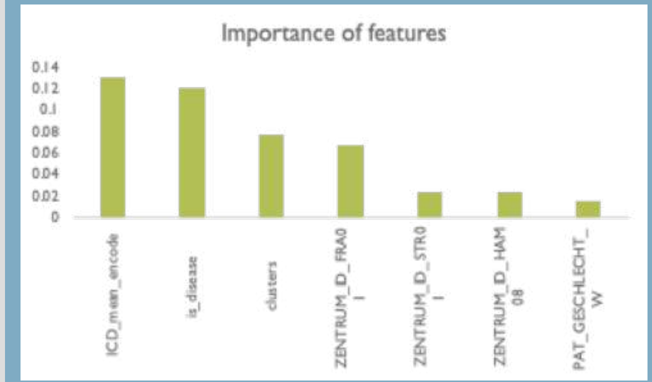
- Split the doctors notes, clean them and perform text analysis
- Group words that most frequently with each other together

MODEL COMPARISON

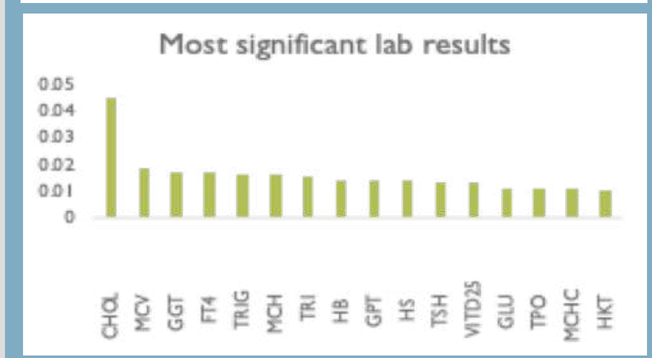
Features considered

- Location
- Sex
- Presence of other diseases
- Text clusters
- Non-normal lab results

Feature Importance

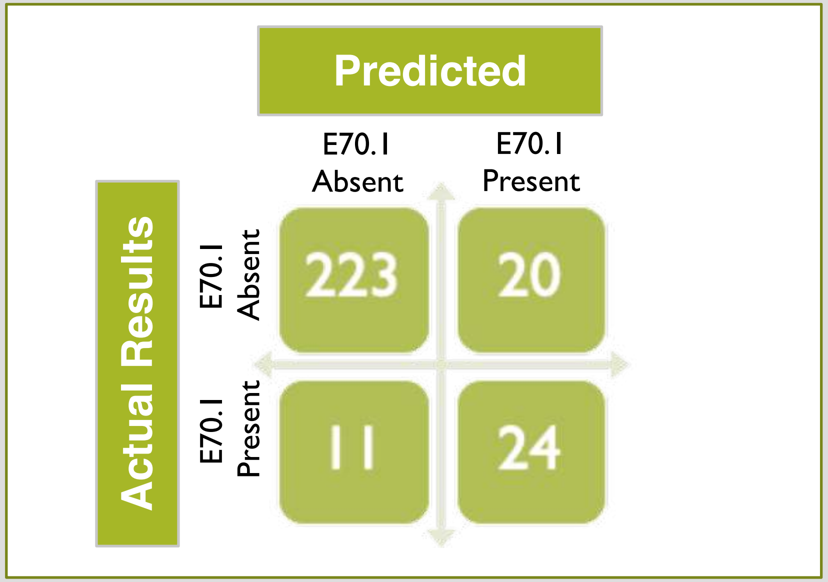


- Correlation with other diseases
- Clusters assigned



- Abnormalities in the lab results of LDL(cholestrol) MGV(blood) and GGT (liver and bone)

Confusion Matrix



Results

- Best performing model: Random Forest
- Accuracy of the model: 90%
- The model successfully classifies 24 of the 35 patients with the disease

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NEXT STEPS

Extract more information from the anamnestic data using natural language processing techniques

Improve the model performance by enriching data with external datasets

Build a more global model that can detect any disease

OUR SUGGESTIONS

Data quality	• Reducing missing values (columns ICD10, SICHERHEIT) • Date formatting (year with 4 digits)
Standardization	• Laboratory test codes • Diseases names
Additional information	• Subcategories in doctor notes (symptom, diagnosis, treatment) • Specialty of doctor (GP, specialist)

A MODEL HAS BEEN DEVELOPED FOR **EACH OF THE DISEASES**

EXECUTIVE SUMMARY

KEY OBJECTIVE	Generate **business value** from Amedes' treatment journals using **machine learning**
OUR APPROACH	Develop **personalized solutions** for each disease based on **research** and **pattern recognition** from the data
FEATURES	Create features based on **tests**, **symptoms** and **co-morbidity**
INTERPRETABILITY	Relevant **interpretabiliity** as we based our appoach on disease characteristics
RESULTS	**Algorithms** that allow us to **detect** diseases with a **relevant accuracy**

APPENDIX

EXAMPLE : β-oxidation defect

1 – Research

- **Reasons for referral:**
  - Adrenoleukodystrophy
  - Adrenomyeloneuropathy
  - Member of patient family

2 – Pattern recognition in the data

- **Symptoms**
  - Fatigue
  - Abdominal pain
  - Adrenal insufficiency
  - Irritability
- **Relevant tests**
  - High SHGB Protein
  - High Thyroxine
  - Low Red blood cell level
  - Low Uric acid
- **Co-Morbidity**
  - E27.1 : Primary adrenocortical insufficiency
  - E06.3 : Autoimmune thyroiditis
  - G40.9 : Epilepsy
  - M62.89 : Other specified disorders of muscle
  - G40.6 : Grand mal seizures

EXAMPLE : Hypercholesteramie

1 – Research

- **Reasons for referral:**
  - Chest pain
  - Family History
  - Member of patient family

2 – Pattern recognition in the data

Symptoms:

- Cholesterol deposits in the eyelids
- Chest pain
- Sudden stroke-like symptoms

- **Relevant tests**
  - LDL Tests
  - GGT Tests
  - MGV Tests

ADABOOST ALGORITHM ALLOWS US TO OBTAIN A **91% ACCURACY** ON THE DETECTION OF NEGATIVE PATIENTS

MODELING METHODOLOGY
Example on Gaucher Disease

Dataset definition

Prevalence of Gaucher Disease: 1/40000
We have 32 patients with GD : we need a sample of size 1.28 million to match the prevalence!

How to define the dataset on which to run and evaluate the model? How many non-GD patients to pick?

Proportion of patients with Gaucher Disease

- Berlin : 0.017 %
- Frankfurt : 0.066 %
- Hamburg : 0.319 %
- Stuttgart : 0.373 %

minimum = 0.017 % / # diagnosed = 32
=> 32 / 0.017 % = 188 000

Model Results with size 1880

Best performing models

```
Logistic Regression 0.989
        Positives:  56.00000000000001 % misclassifed      18 / 32
        Negatives:  0.0 % misclassifed       3 / 1798
Decision Tree 0.981
        Positives:  47.0 % misclassifed       15 / 32
        Negatives:  1.0 % misclassifed       18 / 1798
Neural Net 0.986
        Positives:  59.0 % misclassifed       19 / 32
        Negatives:  0.0 % misclassifed       6 / 1798
AdaBoost 0.986
        Positives:  50.0 % misclassifed       16 / 32
        Negatives:  1.0 % misclassifed       9 / 1798
```

After Hyperparameter Tuning and Resampling (SMOTE and Edited Nearest Neighbors Undersampling)

```
AdaBoost
        Positives:  19.0 % misclassifed       6 / 32
        Negatives:  9.0 % misclassifed       167 / 1798
```