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Huntington's Disease in Germany – an update on prevalence and incidence, current treatment and diagnostic genetic testing

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1. Background

Huntington's disease (HD) is a rare, neurodegenerative, ultimately fatal hereditary disease for which there is currently no curative or progression-delaying medication [1,2]. In addition to specific neurological examinations, the diagnosis can be made via HD-specific genetic testing. However, evidence on the implementation of genetic testing is currently unavailable in Germany.

2. Aims

- To present recent estimates of the **prevalence and incidence of HD**
- To describe **routine care** of prevalent HD patients
- To assess the use of **HD-specific genetic testing** to diagnose HD based on German real-world data

[1] McColgan P, Tabrizi SJ. Huntington's disease: a clinical review. European journal of neurology. 2018;25(1):24-34.
[2] Walker FO. Huntington's disease. The Lancet. 2007;369(9557):218-28.

3. Methods

- This study was conducted in a retrospective, cross-sectional design.
- To estimate the one-year prevalence and incidence of HD, an observation period of 12 months (1 January 2018 to 31 December 2018) was chosen.
- Source of data was the Institute for Applied Health Research Berlin (InGef) Health Research Database, a four-million insurant sample, representative of the German population in terms of age and gender [3].

[3] Andersohn F, Walker J. Characteristics and external validity of the German health risk institute (HRI) database. Pharmacoepidemiology and drug safety. 2016;25(1):106-9.

3. Methods

Prevalent HD case identification: at least two documented ICD-10-GM codes G10 during the year 2018.

Incident HD case identification: Patients without any HD diagnoses in the four years preceding the first observable diagnosis were identified as incident HD cases.

Treatment of prevalent HD cases:

- proportion of patients receiving prescriptions for HD-relevant outpatient medications, medical aids, and remedies
- proportional involvement of medical specialties in the treatment of HD
- proportion of patients with hospitalizations and sick leaves due to HD

Frequency of **HD-specific genetic testing** and extrapolation to the German population

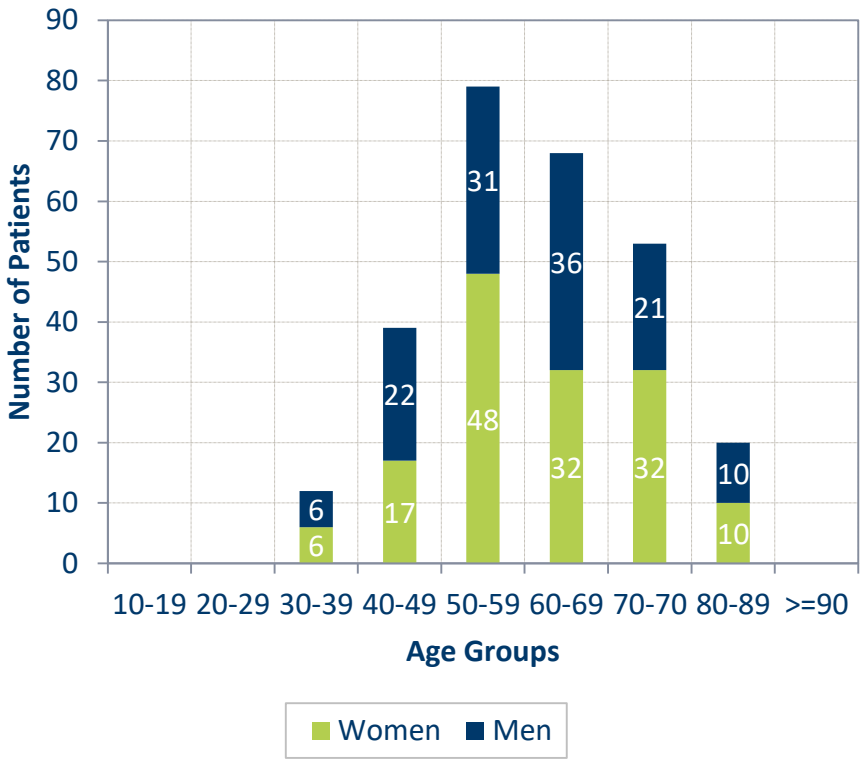
Occurrence of symptoms pre-, and the number of confirmed HD diagnoses post-genetic testing

4. Results

We identified 279 prevalent and 23 incident HD cases in 2018, corresponding to a one-year prevalence of 8.7 and an incidence of 0.7 per 100,000, respectively.

The mean age of prevalent cases was 60.1 years (SD: 13.8) while 53.0% of the patients were female.

Distribution of Prevalent Individuals with HD, stratified by age and sex, in 2018.



4. Results

Treatment of prevalent HD patients in 2018

- **Prescriptions** for antipsychotics were the most frequent (67.0%), followed by antidepressants (39.1%), anticonvulsants (16.5%), hypnotics (12.5%), and opioids (11.8%).
- The most frequently observed **medical aids** were incontinence pads (36.2%) and wheelchairs (14.3%).
- **Remedies**: Physical therapy and speech therapy were observed in 44.1% and 25.4% of HD patients, respectively.
- 50.0% of the involved general practitioners, neurologists, and psychiatrists treated 87.8%, 80.0%, and 81.7% of HD patients, respectively.
- Due to HD, 20.0% of HD patients had **sick leaves** (mean: 137.1 days), and 6.8% had **hospitalizations** (mean: 29.0 days).

4. Results

Occurrence of symptoms and confirmed diagnosis of HD prior to and after genetic testing in 2017 (based on N=33 tested individuals)

Diagnosis of HD prior to genetic testing:

- Number of confirmed HD prior to the test: n=<5 (/*)
- Number of patients with suspected HD in the four quarters before the test: n=5 (15.2 %)
- Number of patients with suspected HD in the quarter of the test: n=20 (60.6% %)

Number of individuals that received a diagnosis of HD within 60 days after testing: n= 11 (33.3 %)

Most frequent symptoms prior to testing:

- Extrapyramidal diseases and movement disorders: n=13 (39.4 %)
- Depressive episode: n=13 (39.4 %)

Number of individuals with no symptoms prior to testing: n= 12 (36.4 %)

Extrapolation to German population: expected testing number of 745

*Note: Numbers that are too low or might allow indirect calculability of too low case numbers cannot be displayed due to data protection reasons.

5. Conclusions

Based on a large representative sample, our study provides:

- Recent estimates of the prevalence and incidence of HD
- The current treatment situation of patients with HD in Germany
- Further insights into the use of HD-specific genetic testing. Most genetic tests appeared to be performed for diagnostic purposes rather than as predictive tests.

This study may provide a starting point for future research into the demand for HD-specific genetic testing as new treatments, some of which may target pre-symptomatic individuals, become available.

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Declaration of Conflict of Interest
Ich erkläre hiermit, dass ich seit dem 1. November 2020 keine geschäftlichen, persönlichen oder materiellen Beziehungen zu Industrieunternehmen, Consulting-Unternehmen oder Kostenträgern bzw. Trägern von medizinischen Einrichtungen unterhalten habe: **Basedow F., Galetzka W.**

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