HUNTINGTON'S DISEASE

Andrea Ros Peña
Genetics Degree
Universitat Autònoma de Barcelona

All you need to know

Home | Epidemiology | Incidence & Prevalence | Etiology | Diagnosis | Therapies | Genetic Counseling

The creation of this website was born from the needing to create a precise and verified information source about Huntington's disease (HD), which is an autosomal dominant progressive neurodegenerative disorder characterized by chorea, cognitive decline and behavioural manifestations.
It is addressed to students and non-specialised professionals and its purpose is spreading relevant knowledge about this rare syndrome in different languages.

CONTENTS

1 Home

1. Diet and Huntington's disease

A research of Columbia University with 1110 individuals from Canada and United States showed no relation between Mediterranean diet and Huntington's disease age of onset.

News — Progress and discoveries in Huntington's disease

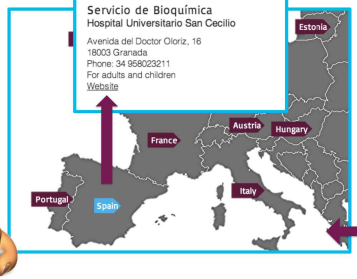
Events — Search for Huntington's disease meetings, speeches and lectures

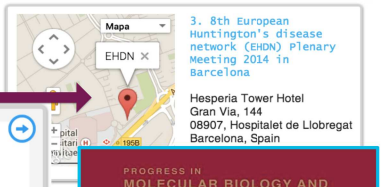
Centres — Find out pionner centres in the research and treatment of Huntington's disease

Articles — Bibliography related to Huntington's disease

SPANISH REFERENCE CENTRES

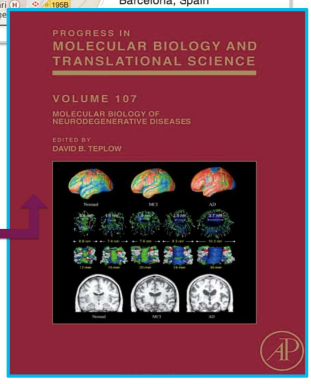
Servicio de Bioquímica
Hospital Universitario San Cecilio
Avenida del Doctor Oloriz, 16
18003 Granada
Phone: 34 958023211
For adults and children
Website





3. 8th European Huntington's disease network (EHDN) Plenary Meeting 2014 in Barcelona

Hesperia Tower Hotel
Gran Via, 144
08907, Hospitalet de Llobregat
Barcelona, Spain



2 Epidemiology

3 Incidence & Prevalence

4 Etiology

Neuropsychiatric symptoms

First to appear

- Lack of emotions
- Failure to recognition of others'needs
- Irritability
- Moodiness
- Euphoria and aggression
- Hipersexuality (5% cases)
- Restlessness
- Tendency towards suicide
- Depression
- Paranoia
- Hopelessness
- Apathy
- Anxiety
- Stress
- Delusions and compulsions (5-12% cases)

Motor symptoms

Progressive degeneration

- Abnormal facial movements
- Involuntary limbs'movements and other parts
- Difficulties in swallowing (improper movement of mouth muscles)
- Loss of co-ordination
- Steady gait

Cognitive symptoms

Progressive degeneration

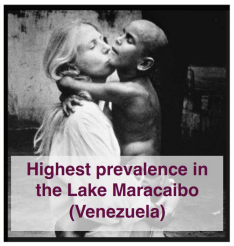
- Dementia
- Abnormal awareness and perception

WORLD'S INCIDENCE

0.38 per 100000

WORLD'S PREVALENCE

2.71 per 100000



Highest prevalence in the Lake Maracaibo (Venezuela)

- Prevalence in Asia is lower than in Europe and North America because of differences in CAG size and in HTT haplotypes distribution

Huntingtin (HTT) gene

- 4p16.3
- It contains CAG repeats which encode polyglutamine

CAG REPEATS	INDIVIDUAL
5 - 35	Normal
>36 - >100	Affected
35 – 38	Incomplete penetrance
<40	Later HD
>60	Juvenile HD

Huntingtin (HTT) protein

- Its proteolysis produces **N-terminal fragments** that:
  - bear polyglutamine tracts
  - display neuronal toxicity and aggregation in neuronal intranuclear inclusions and dystrophic neurites in cortex and striatum.
- Selective degeneration of neurons in caudate and putamen causes HD

5 Diagnosis

- **CLINIC** → presence of unequivocal main signs of HD
- **MOLECULAR** → Targeted mutation analysis of the huntingtin gene to detect CAG expansion (PCR + Southern Blot)
- **NEUROIMAGING**
  - Structural
  - Functional

  HD PATIENTS → **STRIATAL AND BASAL GANGLIA REGIONS VOLUME LOSS**
- **DIFFERENTIAL** → to distinguish HD from choreatic syndromes and HD phenocopies

6 Therapies

- There are therapeutic solutions for palliating some symptoms, especially depression and aggression
- Drugs
  - **Clozapine**: antipsychotic drug
  - **Tetrabenzine**: chorea suppressor
- Therapies under investigation:
  - Fetal striatal tissue transplantation
  - Reducing aggregation involving GLUT1
  - MiR-196a overexpression

7 Genetic Counseling

- Autosomal dominant pattern of inheritance
- Risk estimation:
  - Offspring of an affected individual → **50 %**
  - Progeny of an intermediate allele carrier → **5 %**
  - Expanded penetrant allele: **1/6241 – 1/951**

  Longer expansion → higher risk
- CCG repeats (repetitive sequence adjacent to CAG) → more stable CAG repeats
- Preimplantational genetic diagnosis
- Prenatal diagnosis

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