HUNTINGTON DISEASE (CHOR-HTT)
A Peruvian Perspective

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DISCLOSURES

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OUTLINE

Clinical spectrum of CHOR- HTT

HD Phenotypes

Genetic aspects of HD and diagnosis

HD in Latin America

HD in Peru

HD isolate in Cañete Valley

Challenges on HD management

Final remarks
A polymorphic DNA marker genetically linked to Huntington’s disease

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George Huntington (1850-1916)
*On Chorea* published at age 22

Minimum Prevalence (per 100,000)
- > 5
- 1 - 5
- 0.5 - 1.0
- 0.1 - 0.5
- unknown
Clinical spectrum in HD

Abnormal movement disorders
Behavioral disturbances
Cognitive impairment

“han matado a mi hermano, han sido ellas, mi nuera y cuñada. Quieren hacer lo mismo con mi madre, tengo testigos mire..."
HD Phenotypes

JUVENILE HD PHENOTYPE

VIDEO

LATE ONSET PHENOTYPE

VIDEO
Genetics of HD

HTT gene (4p16.3)

Unaffected individuals

≤26 CAG
Normal

27-35 CAG
Intermediate

36-39 CAG
Reduced penetrance

≥40 CAG
Full penetrance

Affected individuals

Inverse correlation
(The more CAG repeat the earliest onset and more severe disease

Anticipation

Definitive diagnosis: HTT genotyping with CAG repeat counting

In Peru: PCR / TP-PCR genotyping available at CIBN-INCN since 2000
HD in Latin America

- First descriptions: Arostegui in Cuba (1890), Couto in Brazil (1891) and Costa (1894)
- First HD isolates: Maracaibo lake by Américo Negrette (1963) and by Ramon Ávila- Girón (1973)
- USA-Venezuela Collaborative Research Project, led by Nancy Wexler, discover the HTT gene (1993)
CHOR-HTT in Peru

First report
1950

1979-1998
HD Isolate in the Cañete Valley

HTT genotyping

HTT isolate in Cañete Prev
40/100 000
2003-2006

HD Phenotypes
Health costs
Haplotypes analysis
Quality of Life
2015-2108

Peruvian HD Cohort
over 400 HD families
from all over the Country (2000-2017)
HD isolate in Cañete Valley

Outreach clinic in the Cañete Valley
Origin of HD mutation in Latin America

Ancient origin hypothesis: HD came to Lat Am and Peru during the European colonization. However...

Recent evidence (haplotypes study) suggest an Amerindian origin of HD in Peru and Lat Am.
HD Management

Currently
No cure yet?

- Nutritional support
- Support groups
- Physical and occupational therapy
- Mental health
- Genetic counseling
- Pharmacological treatment
- Multidisciplinary approach
# Challenges when living with HD in Peru

The **economic impact of HD** for families and patients in Peru is catastrophic. USD 9,538.00/year.

Patient living with HD at home affects quality of life of whole family.

**Misconceptions of genetics** in HD families: “HD Chorea came from Korea the country” “it is genetic, but if you avoid certain behaviors you will be better” “if you get HD, do not have children”

HD is a rare disorder, for instance is **not a priority** for the healthcare system.
Final Remarks

▪ CHOR-HTT is an progressive neurodegenerative disorder affecting behavior, motor system and cognition.

▪ HD management requires multidisciplinary approach not only for patients, but also at-risk and non-affected family members and caregivers.

▪ Appropriate genetic counselling “family oriented” should be the rule when managing HD.

▪ HD isolates like Cañete requires innovative Health strategies for providing healthcare to those families

HD healthcare could incorporate global health strategies to improve it and became a model of Healthcare for other rare disorders in Peru and other developing countries.
AGRADECIMIENTOS