An Introduction to Huntington’s Disease

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Regional Care Advisor

Essex
The Huntington's Disease Association exists to support people affected by the disease and to provide information and advice to professionals whose task it is to support Huntington's disease families.

The HDA is financed through the generosity of trusts, foundations, the statutory and corporate sectors, branches of the HDA, members and friends.

Head office is based in Liverpool
The office is open from 9:00am to 5:00pm Monday to Friday
Tel: 0151 298 3298
Regional Care Advisory Service

This consists of a team of Regional Care Advisers who:

- provide information and advice to families
- answer crises calls and liaise with other professional service providers
- promote and develop a full range of local services
- identify suitable respite and residential care facilities
- liaise with local branches and self-help groups
- give talks and organise seminars and training days
- provide speakers for training sessions
- provide workshops for service providers and users such as health, social services, nursing homes, and residential care staff teams
Local branches and groups throughout the country

These groups provide a local, informal setting for families and individuals to:

- mix socially
- offer support and advice
- share experiences and ideas
- form local links with professionals and other groups in their community
- raise funds
- organise speakers

and above all, provide a forum to share a common purpose and reduce the feeling of isolation.

Essex Branches in Chelmsford and Southend.
What is Huntington’s Disease?

- Huntington’s Disease is an inherited degenerative neuro-psychiatric disorder.
- Approx 10 per 100,000 people have HD
- Each child has a 50:50 chance of inheriting the disease
What is Huntington’s disease?

• Huntington’s Disease is a dominant genetic condition caused by a mutation on Chromosome 4.
• Symptoms of Huntington’s Disease usually start between the ages of 30-50 years, although late onset and juvenile forms do occur.
• The prognosis is usually 15-20 years from onset of symptoms.
Passing on the Gene

- Anyone who inherits the gene will, at some stage, develop the disease, unless they die before that happens. They can pass it on to any children they have.
- If a person doesn’t inherit the gene, they won’t get the disease and they can’t pass it on to their children, who won’t pass it on to their children.
- Sometimes a person who has the HD gene dies before any symptoms appear. When this happens they may have passed it on to their children without anyone realising it has happened.
Why is it called Huntington’s Disease?

- It was named after George Huntington, a New York doctor, who first described it in 1872.
- It used to be called Huntington’s Chorea.
- We now know that the disease causes many other changes, so the name was changed to Huntington’s Disease, or HD for short.
What does HD do?

- The faulty gene damages the nerve cells in certain areas of the brain, which leads to gradual physical, emotional and cognitive changes.
- The gene was identified in 1989 and predictive testing has been available since 1992 via genetic clinics.
- The first signs have been seen in people as young as 2 and as old as 80, although this is unusual.
- Symptoms begin gradually, often they are so mild and change so slowly that it can take a while before anyone realises there is anything wrong.
- Everybody is different, so everyone’s response to HD is different, even in the same family.
A Triad of problems with Huntington’s Disease

These problems interact with each other

- Physical problems
- Intellectual problems
- Mood and behavioural problems
The tricky bit.....

“The Genetics”
Genetics

• Genes are made up of DNA
• DNA is made up of 4 chemicals
  • A Adenine          C Cytosine
  • G Guanine         T Thymine
• One section of the HD gene contains 3 of these chemicals repeated many times in a chain
  CAGCAGCAGCAGCAGCAGCAGCAGCAGCAGCAGCAGCAGCAG
• The gene responsible for HD is on Chromosome 4
Genetics

• Normal CAG repeat under 27
• Between 27 and 35 described as ‘grey’ area
• 36-39 repeats means you have a positive result, but symptoms may occur later
• 40+ abnormal
• Count does not tell you when symptoms will start
The basic genetic problem

- The chemicals combine to form a protein
- Expanded HD gene produces too much
- Excess is stored in the brain
- The abnormal protein accumulates over time
- It interacts with many other genes and proteins
- It progressively damages the brain
- The brain can ignore this damage until...Symptoms appear, usually after many years
Healthy vs HD Brain
Typical Physical Changes

- Often starts with fidgety movements
- Might seem clumsy or stumble more than usual
- Speech might sound a bit slurred
- As the disease progresses, swallowing problems are common and choking on food
- Movements become more pronounced and problematical
Typical Emotional Changes

• Often starts with subtle changes to mood / behaviour

• Some people get frustrated leading to temper outbursts

• Depression is common, but very treatable

• Some people become aggressive, demanding, stubborn and self-centred.

• People can be impulsive or irrational, behaving in a disinhibited way or obsessive with things
Typical Cognitive (thinking) Changes

- Harder to plan and think ahead
- Harder to switch from one task to another or to multitask
- Lack of motivation – appear lazy
- Memory and concentration problems
- Inability to recognise they have a problem
What does it feel like to think with HD?

**Characteristics of thinking with Huntington’s**

- Slower thinking
- Recognition is easier than recall
- More difficult to change topics
- More difficult to organise, plan and sequence
- Can’t wait

We’ll do a few exercises to show you how it might feel to think with HD.
Slower thinking... how it feels

Take the pen and paper...
When I tell you to begin, write your name again and again...
Comfortably...
Until I tell you to STOP.
Slower thinking….how it feels

• Count the number of words you wrote
• That was 15 seconds
• Multiply your number by 4 to get a minute
• How many words per minute?

Fluent writing is a minimum of 16 words per minute
Slower thinking….how it feels

Now write your name very slowly
6 letters per minute
1 letter every 10 seconds
I will talk you through it........
STOP!

Here’s mine…

Alison Heaney.
Slower thinking....how it feels

Do you want to continue?
How did that feel?
It’s tiring isn’t it?
Concentrating is exhausting isn’t it?
Distractions come easily...
Slowing it down affects quality...
• It feels exhausting...annoying...lousy...more effort

• What was unconscious becomes conscious.

• ”It was automatic, now you have to pay attention to it.”

• “My automatic pilot’s broken!”

• Something can be so slow that you just don’t do it.
Recognition is easier than recall

Huntington’s disease is NOT Alzheimer's disease….memories are there!

Accessing them and expressing them is more difficult

There are different kinds of memory.
Recognition is easier than recall

Recognition
Recall
Relearning
Recognition is easier than recall…How it *feels*

- What is recall?
- What is recognition?
- They *feel* different…let me show you
- Take your paper again…..
- Draw a penny, both sides, from memory

- How did you do?
Recognition feels like this!
Recognition is easier than recall…How it feels

How did recognition feel?

Easier than recall?

When you see it and recognise it, it feels like relief

Imagine answering every simple question with the effort of recalling that penny!
Recognition is easier than recall...when we see it

We can see it in ourselves.

When we sit an examination we hope there are no ‘open ended questions’ or ‘essay type’ questions.....

We hope for multiple choice questions.

Open questions need recall

Multiple choice questions need recognition

The answer is there! We all prefer multiple choice...
Topic changes are difficult…what we know

• “Divided attention” is difficult.

• People get “locked on” to a topic, they persevere.
Topic changes are difficult...how it feels

Say the colours of the word as fast as you can

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Topic changes are difficult...how it feels

Say the colours of the word NOT what the word says as fast as you can.
Topic changes are difficult…how it feels

• Difficult?
• Confusing?
• An unconscious effort is made conscious.
• **Feels** like…
Topic changes are difficult…how we can help

• Minimize distractions
• Go slow
• One thing at a time
• Obvious
• Simple
• Absolutely helpful

Too easy to think that they are important?
Can’t wait! What we know

• Impulse control is a problem.
• Lack of inhibition is a problem.
• Sense of time seems altered.
• He gets locked onto a topic…over and over and over…
Can’t wait! When we see it

• If you have Huntington’s Disease, you often ask, “What’s taking so long?”
• “When someone with Huntington’s Disease wants something, they want it NOW!!!”
• “It seems that people with HD just can’t WAIT!!!”
Can’t wait! How we can help

• Avoid “No” and “Wait!”
• “A minute” is 60 seconds.
• Never make promises that you can’t keep.
• It is easier to do it now…if you can.
• Do not reason.
• You can always do it early; you can never do it late.
Can’t wait!

Could you endure…
...with grace…
..waiting for anything…
...or everything you want?

Might it feel like…….?
Thank you for listening.

Any Questions?

Further information available:

Understanding Behaviour in HD
Book available on order from HDA head office

Fact Sheets 9 & 10
Available by post from HDA or downloadable from

www.hda.org.uk