• Impairment of the Inhibitory Functions of the BG
  • Dyskinesias – involuntary movements
    • Chorea (“dance-like”) – quicker irregular movements
    • Athetosis – slower writhing, twisting movements
  • Dystonias – abnormalities of excessive muscle tone

• Huntington’s Disease
  • Transmitted by a dominant gene on chromosome 4 (about 30,000 US cases with 150,000 at risk kids)
  • Deterioration of striatum produces involuntary chorea, athetosis & other motor difficulties
  • Cortical deterioration causes progressive & debilitating dementia, aggressiveness, mood swings, depression, psychosis
  • Death due to health complications in 15-20 yrs

• Huntington’s Disease
  • Bad gene has excess “CAG repeats” (more than 36-250 instead of usual 28 or fewer) resulting in an abnormal form of protein known as huntingtin.
  • The more repeats, the earlier symptoms appear.
  • # of repeats can increase across generations, especially in kids inheriting gene from father
  • Brain damage may be due to decrease in normal protective huntingtin + adverse effects of abnormal protein on critical growth factors keeping cells alive.
  • HD may cause increased susceptibility to excitotoxic glutamate and/or abnormal programmed cell death.

• CAG Repeats & Age of Symptom Onset

• Treatments for HD
  • Genetic testing to identify presence of the gene
  • Involuntary movements may be decreased by DA blockers (antipsychotics)
  • New drugs being tried to delay progression:
    • Rilutek (riluzole), Neurontin (gabapentin) decrease glutamate transmission
    • Rapamycin (transplant drug) speeds elimination of abnormal protein
  • Growth factor supplementation being studied
  • Experimentation with brain cell transplants/surgeries is underway
**Tourette Syndrome**

- Another hereditary BG disorder characterized by involuntary movements
- Multiple motor tics - simple tics of face or limbs and/or more organized complex tics (touching, grimacing, pinching, poking, adjusting, hitting, jumping, kissing, throwing, gestures) **plus**:
  - Phonic or vocal tics - both simple (throat-clearing, coughing, hiccuping, grunting, yelping) and/or complex tics (actual words, coprolalia, echolalia, palilalia, assuming different voices, talking to oneself in different voices)
- Seems to affect frontal lobe-BG connection that is important for our ability to inhibit actions

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**Tourette Syndrome**

- 40% report “sensory tics” — uncomfortable sensations that may be a reason for some of the involuntary movements
- Some degree of suppressibility, but individual experiences increased tension until tic is released
- Pattern of tics changes & waxes & wanes with changes in stress, anxiety, fatigue.
- Treated with DA blockers (antipsychotics). Milder tics may respond to NE agonist clonidine.
- Majority experience decreased tics as adults.

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**Link with Other Disorders**

- ~50-60% also suffer OCD (others estimate that up to 90% experience some involuntary touching compulsions, ritualistic behaviors, intrusive thoughts)
- ~50-90% show evidence of ADHD as well; first signs of GTS are usually impulsive, hyperactive behaviors (before tics appear)
- About 30% have learning disabilities, emotional lability, rage, aggressiveness; 40-50% depressed
- Evidence suggests a single gene with sex-linked, varied forms of expression of disinhibition
- 50-73% concordance in identical twins vs 8-22% in fraternal twins

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**Tourettes videos**

- Twitch and Shout
- I Have Tourette's
- John Stossel
- McGraw Hill abnormal sampler

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**Medications**

http://www.tourettesyndrome.net/tourette_treatment.htm#Introduction

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**Other Resources**

- [http://video.google.com/videoplay?docid=-5419552820725946842&q=tourette&hl=en&client=firefox-a#docid=8729287416499666301](http://video.google.com/videoplay?docid=-5419552820725946842&ei=FrKWS-KuMoygqQOnweCHAg&q=tourette&hl=en&client=firefox-a#docid=8729287416499666301)