THE GENETIC BASIS OF NEUROLOGICAL DISORDERS



The Biomedical & Life Sciences Collection



A complete advanced undergraduate/graduate course with:

- 22 online lectures by leading authorities
- Resources for workshops, tutorials, journal clubs, projects and seminars
- Suggested exam questions and model answers
- Multiple choice questions and answers
- Recommended reading: original papers and review articles

View the content of the course on our website: hstalks.com/GeneticBasisOfNeurologicalDisorders



View our in-depth HSTalks: hstalks.com/CoursesBrochure

Course module with video lectures, material for tutorials (case studies, projects, workshops and recommended reading), multiple choice questions and suggested exam questions with model answers. A comprehensive course on a subject of major importance.

The material is especially designed to support research and teaching staff when presenting a comprehensive course at graduate or advanced undergraduate level with seminars, journal clubs, laboratory exercises, data workshops, online tests and end of course examinations.

The course is also suitable for continuing professional development/education programmes.

This brochure provides brief details of the complete module, including the lectures, lecturers and additional learning material.

Who is the course for?	The comprehensive material is especially suitable for teachers and researchers who wish to offer courses on specialist subjects to small groups of students (or even a single student) when it is not possible to justify the time and expense of preparing, internally, a course or there is not the range of expertise available locally to do so. All the lecturers are highly regarded experts in their fields and few institutions are likely to have a comprehensive group of faculty members with a similar range of experience and knowledge of the subject matter.
	The course material is designed to be used by local faculty and staff acting as course directors, tutors and mentors.
	The material is suitable for flipped classroom, blended, team and distance learning courses.
	New courses are time consuming and expensive to create. These modules cut both the cost and the time, enabling a wider range of options to be offered on specialist topics. Graduate students can take the courses, mentored by their supervisors, while pursuing their research.
Ideal for Virtual Learning Environments (VLE)	All course material, including the additional learning material, is arranged in a standard format that allows easy embedding into virtual learning environments such as Moodle, Blackboard or your institute's own system.
Supporting learning and teaching goals	In an age when faculty and staff face ever greater demands on budgets and time, these lectures and additional learning material will be of great help when preparing and delivering graduate and advanced undergraduate courses.



Course Summary

Neurological diseases represent one of the gravest challenges to humans in the 21st century, with the prevalence of disorders such as Alzheimer's and Parkinson's increasing as populations age across the globe. Over the past thirty years, huge progress has been made in understanding how genetics can influence the onset of neurological diseases - from dominant Mendelian inheritance through to common genetic variability enhancing lifetime disease risk. This course summarizes our current understanding of the genetics of neurological disorders, providing a detailed grounding in how clinical symptoms and pathology match up to genetic background. For each neurological disease loci to the cellular etiology of the disorder. This will be complemented with lectures on how the latest genomic and proteomic approaches are helping us develop novel therapeutic avenues of inquiry, with the aim of producing drugs to slow down the degenerative process in these disorders, while highlighting the huge challenges that face the biomedical research community in this area of drug development.



Prof. John Hardy Institute of Neurology, University College London, UK



Dr. Patrick A. Lewis University of Reading, UK

The course module is designed for:

- Researchers and graduate students in the fields of neuroscience, genetics and genetic counselling
- Researchers and decision makers in the pharmaceutical industry
- Clinicians and medical students
- Advanced undergraduate students
- Continuing Professional Education/Development

Course Lectures

Neurodegenerative disease: the medical > imperative for the developed world

Click the lecture title to access

Genetic basis of neurological disease

the genetic basis of neurological disorders

An overview of the neuropathology of

neurodegenerative diseases

Institute of Neurology, University College London, UK

Prof. Nicholas Wood University College London, UK Genetic methods in the study of neurological diseases:

Prof. John Hardy

Dr. Jose Miguel Bras University College London, UK

Dr. Tammaryn Lashley

Dr. Patrick A. Lewis University of Reading, UK

University College London, UK

Deciphering neurodegeneration: models and methods

Early clinical features of Parkinson's disease and related disorders

Dr. Alastair Noyce Institute of Neurology, University College London, UK

Autosomal recessive Parkinson's disease and mitophagy

Cellular pathways linked to dominant Parkinson's disease

> Dr. Patrick A. Lewis University of Reading, UK

Dr. Helene Plun-Favreau University College London, UK

Alzheimer's disease: current status of genetics research

Dr. Rita J. Guerreiro University College London, UK

The genetics of frontotemporal dementia

Prof. John Hardy Institute of Neurology, University College London, UK

Inherited neuropathies























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THE GENETIC BASIS OF NEUROLOGICAL DISORDERS

22 specially recorded, animated lectures by world leading authorities

	Huntington's disease and HD-like disorders				
Click ecture title o access	Genetics of epilepsy	Prof. Sarah Tabrizi University College London, UK			
		Prof. Sanjay Sisodiya University College London, UK			
	Functional insights from genetic channelopathies				
	The genetics of progressive supranuclear palsy and corticobasal degeneration (CBD)	Dr. Stephanie Schorge University College London, UK (PSP)			
	Pathogenesis and cell biology of amyotrophic lateral sclerosis (ALS)	Dr. Rohan de Silva University College London, UK			
		Dr. Pietro Fratta University College London, UK	0.1		
	The inherited ataxias				
		Dr. Paola Giunti University College London, UK	25		
	Genetics of hereditary spastic paraplegia				
		Dr. Arianna Tucci University College London, UK	F		
	The genetic basis of dystonia				
		Prof. Tom Warner University College London, UK			
	Pathogenic mechanisms in prion disease				
		Prof. Giovanna Mallucci University of Cambridge, UK	101		
	Next generation sequencing in genetic diagnostics				
		Dr. Alan Pittman University College London, UK			
	Biomarkers of neurodegeneration				
		Prof. Henrik Zetterberg Gothenberg University, Sweden	6		

































Examples of Course Materials

For each lecture, the course offers tutorials, workshops, recommended reading, multiple-choice questions, and suggested exam questions with model answers.



HST Moodle My Courses > The Genetic Basis of Neurological Disorders

Tutorial: Huntington's disease and HD-like disorders

A 40 year old male reports some loss of mental acuity at work over the last 4 months, leading to some performance issues. His wife describes some change in behaviour with increased irritability and social withdrawal, over the last year but puts it down to stress at work. Although he

- was estranged from his father, he was told he passed away aged 60 and spent his last years in a nursing home due to difficulties with self-care, communication and immobility. No formal diagnosis is available.
- Clinical examination reveals jerky smooth pursuit movements and mild distal chorea of all 4 limbs with mild slowing and loss of rhythm on finger taps.
- (A) What important differentials should one consider, and how the examination findings and family history affect them?
- (B) Does the absence of family history exclude the possibility of HD? (C) Explain the important points that should be covered when undertaking genetic counselling.
- (D) In the event that a subsequent HD test was negative, what would you do next? What clues in the history may help you? As it turns out the HD test is positive. Many years later, the gentleman attends with his 16 year-old daughter who has developed some symptoms quite
- different to his own. Examination reveals significant axial and limb rigidity, slowness of movement but no chorea. (E Compare and contrast the movement disorder within the same family? (use videos side by side)
- (F Is this consistent with Huntington's disease? (G What genetic features are being illustrated here?

?	Exam Questions and Model Answers	Question 1 Not yet answered Marked out of 1.00 V Flag question C Edit question	Compare and contrast HD with the HD-like disorders. A = B I := B C C C
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l≔D	Multiple-choice	Question 4	Which disorder is characterised by brain iron accumulation?

الللب questions and Not vet Select one: answered answers a) Spinocerebellar ataxia Type 17 Marked out of 1.00 O b) Neurodegeneration caused by PANK2 mutations* ○ c) Progressive supranuclear palsy C Edit question d) Multiple system atrophy e) Huntington's disease



Recommended reading supporting each lecture: Original research papers and review articles

Recommended Review Articles

- 1. Novak & Tabrizi, Huntington's Disease. BMJ. 2010 Jun 30;340:c3109
- 2. Ross & Tabrizi, Huntington's disease: from molecular pathogenesis to clinical treatment. Lancet Neurology 2011, 10:83-98
- 3. Wild & Tabrizi, The differential diagnosis of chorea. Pract Neurol. 2007 Nov;7(6):360-73
- 4. Gillian Bates, Peter Harper and Lesley Jones, Huntington's Disease. Oxford University Press



How to access the course

Extracts of lectures can be viewed at hstalks.com/biosci/. The full length lectures can be viewed by all members of universities, colleges and medical schools currently subscribing to The Biomedical & Life Sciences Collection. Institutions that do not subscribe to The Biomedical & Life Sciences Collection may take annual licenses at US \$2,000 covering an unlimited number of students.

Full supporting material: video lectures, material for tutorials (case studies, projects, workshops and recommended reading), multiple choice questions and suggested exam questions with model answers are provided to faculty members of subscribers.

To subscribe, obtain additional information and/or the additional learning material contact Dr. Eyal Kalie at eyalk@hstalks.com.

Upload to your VLE

The complete course (lectures and additional learning material) can be loaded into Moodle, Blackboard and other virtual learning environments.



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