

Prof Edward Tobias
Univ. of Glasgow,
Scotland, UK


The ESHG's dedicated website for educational resources

www.eurogems.org
(ESHG Genetic Educational Materials & Sources)


- A new website with >100 categorised & fully up-to-date links (& summaries) to carefully selected online resources from around the world, for education & reference

by Prof Edward Tobias
BSc MBChB PhD FRCP FHEA
Professor of Genetic Medicine &
Hon. NHS Consultant in Clinical Genetics,
Queen Elizabeth University Hospital
and University of Glasgow, Scotland, UK.

(Also: author of free Clinical Genomics smartphone apps, at:
www.genomicsapps.org)



The screenshot shows the EuroGEMS.org website interface. At the top, there are navigation tabs: HOME, FOR GENETICS PROFESSIONALS, FOR UNIVERSITIES & STUDENTS, FOR PATIENTS & FAMILIES, FOR SECONDARY SCHOOLS, and FOR PRIMARY SCHOOLS. Below the tabs is a header for 'ESHG Genetic Educational Materials & Sources (EuroGEMS.org)'. A paragraph of text explains the site's purpose: 'A large number of highly impressive educational resources have already been developed and made available online, internationally. The purpose of the pages provided here is to assist educators and individual learners in identifying sources of educational materials, related to genetics and genomics, that they may find useful. In addition to the many sources to which these pages link directly, many of the linked websites themselves include many further links to substantial educational resources.' Below this text is a grid of six resource categories, each with a representative image and a label: 'For genetics professionals' (OMIM), 'For universities & students' (a screenshot of a website), 'For patients & families' (a poster about cancer), 'For secondary/high schools' (a diagram of DNA), 'For primary schools' (a diagram of DNA), and 'Ethical, legal & social issues' (a photo of people in a meeting).

 University of Glasgow

Overview of presentation

1. Background to the creation of the EuroGEMS.org website
2. Website content (as screenshots)
3. Analytics – showing an exponential increase in use, internationally, over the past year
4. Testimonials/feedback from users
5. Thanks and request for assistance (final slide)


Prof Edward Tobias
Univ. of Glasgow,
Scotland, UK

Background: personal teaching responsibilities

TEACHING

IN U.K.

- **Clinical Director of 3 large postgraduate Masters (MSc) programmes at Glasgow University** (with >800 ex-students around the world)
 - MSc in Medical Genetics & Genomics (UK-award-winning)
 - MSc in Genetic & Genomic Counselling & MSc in Molec. Pathology.
- **Winner of 11 teaching awards (including 4 from students, team awards and two national awards)**
 - Lead for Clinical Genomics course with computer workshops for data analysis, training students and staff
 - Lead for Medical Genetics teaching to 300 medical students / year
 - External advisor & examiner at 4 other universities (1 overseas)



INTERNATIONAL CONTRIBUTIONS:


- **Member of exam. committee establishing the new UEMS European Diploma in Med. Genetics & Genomics (EDMGG exam: 14th June 2019)**
- **A presenter on international MOOC** ("Cancer in 21st Century: Genomic Revolution")
- **Author of 2 textbooks on Medical Genetics (with translations)**
- **Author of Clinical Genomics Apps** (expanded in Feb 2019 & released free on the App stores; see genomicsapps.org) Link: www.genomicsapps.org

Prof Edward Tobias
Univ. of Glasgow,
Scotland, UK

Background: other work-related roles

RESEARCH

- One of principal investigators on the Scottish Genomes Partnership whole genome sequencing (WGS) project (which now has received £10.2 million of funding)
- Supervising post-grad. Research on:
 - Whole exome sequence (WES) data analysis
 - Small ncRNA analyses and
 - Collaborations with: Deciphering Developmental Disorders (DDD) (Cambridge), MRC Harwell (Oxford) & Crick Institute (London)




University of Glasgow, Scotland, UK. Fourth-oldest university in English-speaking world.

CLINICS

(as Honorary NHS consultant)

- General clinical genetics clinics in South-west Scotland, UK.
- Multi-disciplinary team meetings discussing clinical cases and variants

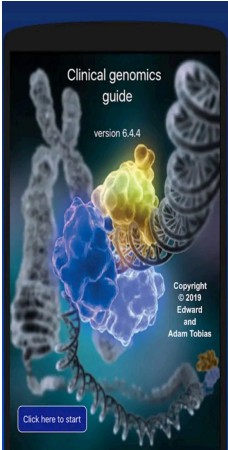
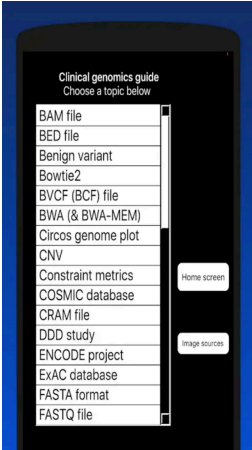
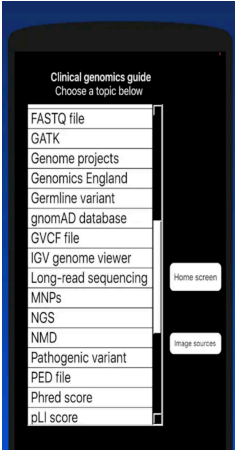
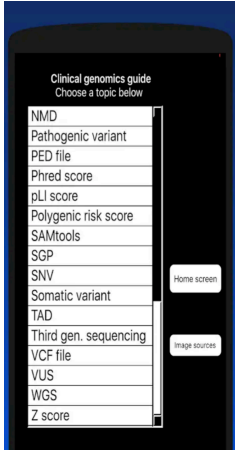


Queen Elizabeth University Hospital, Glasgow, UK. With 14 floors, is one of the largest UK acute hospitals.

Prof Edward Tobias
Univ. of Glasgow,
Scotland, UK

Clinical Genomics apps for smartphones

- **New expanded version of Clinical Genomics glossary app was launched in Feb 2019 on both App stores. Quickly explains complex genomics terminology. Free to download.**
- Accompanied by a self-assessment quiz app.
- **New version has over 40 definitions of e.g. BED, FASTQ & GVCF files, NMD & TADs**
- **See www.genomicsapps.org (or search App store for Edward Tobias)**

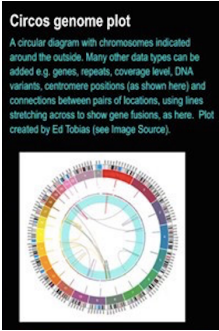





Prof Edward Tobias
Univ. of Glasgow,
Scotland, UK

Additional screenshots from App


Circos genome plot

A circular diagram with chromosomes indicated around the outside. Many other data types can be added (e.g. genes, repeats, coverage level, DNA variants, centromere positions (as shown here) and connections between pairs of locations, using lines stretching across to show gene fusions, as here. Plot created by Ed Tobias (see Image Source).




Third generation sequencing

Also known as "long-read sequencing". Performed, by different methods, on long size DNA molecules rather than on many small segments of DNA (as in second generation sequencing i.e. NGS). Image shows DNA (white) entering an alpha-haemolysin "nanopore". See "long-read sequencing".

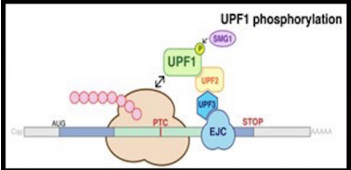


IGV

IGV (integrative genomics viewer) is a program for displaying e.g. BAM files, showing the sequence reads aligned to a reference sequence.



UPF1 phosphorylation



VCF header rows

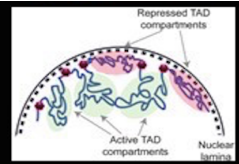
Chrom & position	Reference	Substitution
1 1000000	A	G
1 1000001	T	C
1 1000002	C	T
1 1000003	G	A

Quality scores

Repressed TAD compartments

Active TAD compartments

Nuclear lamina



Now used by approx. 5000 individuals, in >70 countries

Prof Edward Tobias
Univ. of Glasgow,
Scotland, UK

Creation of www.EuroGEMS.org website for ESHG

- **Criteria for inclusion**
 - Links to sites (approx. 100 to date) that provide useful, high-quality, readable free information
- **Content discussed with**
 - Many ESHG committee members
 - Local professional colleagues
 - Students
 - School teachers
- **Technical aspects of website**

include:

 - Unlimited bandwidth
 - Fast page-loading

ESHG The European Society
of Human Genetics

News About the ESHG Membership Meetings Public & Professional F

Genetic Education

**The EuroGEMS.org is accessible via the link below
from this ESHG web-page
or directly, by entering: eurogems.org**

Genetic Education

This section is intended to be an educational resource for the Human Genetics Community and contains information on Courses (co) organised by the ESHG, ESHG's webcast archive, the excellent EuroGEMS.org website dedicated to Educational Resources, Calls for collaboration, Job opportunities and more. Choose

Prof Edward Tobias
Univ. of Glasgow,
Scotland, UK

Website content: screenshots

The following slides contain screenshots of selected web-pages from EuroGEMS.org

Prof Edward Tobias
Univ. of Glasgow,
Scotland, UK

ESHG Genetic Educational Materials & Sources (EuroGEMS.org)

Educational resources for genetic & genomic education

A large number of highly impressive educational resources have already been developed and made available online, internationally. The purpose of the pages provided here is to assist educators and individual learners in identifying sources of educational materials, related to genetics and genomics, that they may find useful. In addition to the many sources to which these pages link directly, many of the linked websites themselves include many further links to substantial educational resources.

Screenshot of the EuroGEMS.org home-page



OMIM®
Online Mendelian Inheritance in Man®
An Online Catalog of Human Genes and Genetic Disorders



Genetic database interface showing search results and filters.



ARE YOU WORRIED ABOUT YOUR ASTHMA? ARE YOU WORRIED ABOUT YOUR CANCER?
Genetic testing and family history

For genetics professionals **For universities & students** **For patients & families**



When and how to refer



Secondary/high school educational resource with DNA diagram.



Primary school educational resource with DNA diagram.

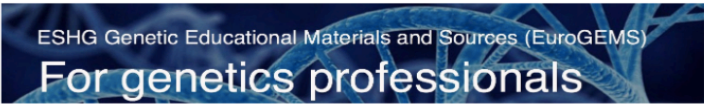
For primary care **For secondary/high schools** **For primary schools**



Ethical, legal & social issues resource showing people at a computer.

Ethical, legal & social issues


Screenshots from
EuroGEMS web-
page "For
Genetics
Professionals"



This page provides information that is likely to be useful to professionals in clinical genetics/genomics e.g. information about individual clinical conditions (GeneReviews, OMIM and Orphanet), individual genetic variants (DECIPHER, LOVD, ClinVar, HGMD, VarSome, CanVar & VEP), transcript identification (LRG) and variant nomenclature (HGVS). Professionals are also likely to find sources that are listed on other pages here to be of relevance and interest, particularly the [Universities page](#) (including the genome browsers) and the [Ethical, Legal and Social Implications page](#). On this page:


[CLINICAL INFO](#) | [GENOME BROWSERS](#) | [VARIANT ANALYSIS](#) | [GENERAL](#) | [LOCAL INITIATIVES](#)

CLINICAL INFORMATION




GeneReviews

This database contains a large number of reasonably comprehensive, disease-orientated, reviews. They are generally expert-authored and easy to read, containing clinical features, differential diagnoses and molecular details.



OMIM


An online, highly detailed, reference catalogue, listing human genetic conditions & their related genes. Initially compiled by the late Victor McKusick it now contains >24,000 entries & includes valuable links to other databases e.g. ClinVar.



Orphanet


A portal for rare diseases & orphan drugs. Database of numerous short articles regarding rare diseases and disease genes. Organisation is based in Paris.

GENOME BROWSERS




DECIPHER

An interactive database with a suite of tools to aid interpretation of genomic variants. It retrieves information from a variety of bioinformatics resources. To help interpretation, a patient's variant is displayed together with levels of normal variation and pathogenic variation at that locus.



Ensembl Genome Browser


A powerful genome browser, based at the Wellcome Trust Sanger Institute, UK. Contains a link to Variant Effect Predictor (VEP), for analysis of likely consequences of DNA variants.



UCSC Genome Browser


The Genome Browser of the University of California Santa Cruz. An easy-to-use website that displays all the options on a single configurable web page.

VARIANT ANALYSIS & INTERPRETATION




DECIPHER

An interactive database with a suite of tools to aid interpretation of genomic variants. It retrieves information from a variety of bioinformatics resources. To help



LOVD

Leiden Open Variation Database (LOVD): an online gene-centered collection and display of DNA variations. Contains a large body of variant data plus valuable updated



ClinVar

A database of genetic variants, based at the NCBI in USA, containing in a single database, hundreds of thousands of records, regarding specific gene variants.



gnomAD (genome Aggregation Database)

The genome Aggregation Database (gnomAD) contains over 120,000 exome sequences plus over 15,000 whole-genome sequences. It is far larger than ExAC (containing approximately 60,000 exomes), which preceded gnomAD.



Human Genome Variation Society

The HGVS provides helpful guidelines for describing specific variants in DNA, RNA and proteins, according to internationally agreed standards. A relatively brief "simple" summary can be found [here](#).



Human Gene Mutation Database

The HGMD at Cardiff in Wales, UK, is an attempt to collate all published gene lesions responsible for human inherited disease. The full (professional) database requires a subscription but academics can access the less up-to-date but still useful, version.

Screenshots (continued) from EuroGEMS web-page "For Genetics Professionals"



VarSome DNA variant analysis

VarSome provides analysis of individual genetic variants, one at a time. Permitted input formats include HGVS nomenclature for DNA-level and protein (amino acid) level variants (but not protein-level frameshift variants entries, as not sufficiently specific). Also provides automated variant interpretation using ACMG-guidelines.



Cancer predisposition gene Variant Database


A resource for cancer predisposition gene variant interpretation. Uses multiple data sources to classify variants, including shared data from NHS diagnostic laboratories on BRCA variants to enable case:control comparisons with population data. Includes both automated and curated classifications. Contains links to functional analyses eg saturation genome editing BRCA1 haploid cell assay.



Variant Effect Predictor (VEP)


Variant Effect Predictor (VEP), part of the Ensembl Genome Browser, provides automated interpretation of single or multiple genetic variants. It provides Polyphen and SIFT analysis scores and also provides frequency data from ExAC and gnomAD.

GENERAL INFORMATION




Locus Reference Genomic

A database, supported by EMBL and NCBI, providing non-changing gene and transcript identifiers. Increasingly used by laboratories to determine which specific transcript is to be used when reporting gene variants.



Glossary & Quiz Apps for Clinical Genomics

Free glossary app, containing brief illustrated explanations of >40 frequently-used terms e.g. "FASTQ, BAM, VCF, gVCF, Phred, BED, BWA & GATK". Also an accompanying self-assessment quiz app. Both apps function offline & are used as reminders by students & professionals, internationally.



European Reference Networks

Virtual networks involving Reference Centers across Europe. They aim to tackle complex or rare conditions by convening a "virtual" advisory board of medical specialists to review a patient's diagnosis & treatment. Provided in MULTIPLE LANGUAGES.

Screenshots (continued) from EuroGEMS web-page "For Genetics Professionals"

LOCALISED/NATIONAL INITIATIVES

Please note that the Orphanet and European Reference Networks websites (above) are provided in multiple languages.

Other resources:


England
[Health Education England: Genomics Education Programme](#)

France
[France Genomique: training](#)


<p>Screenshots (continued) from EuroGEMS web-page “For Genetics Professionals”</p>	<p>LOCALISED/NATIONAL INITIATIVES</p>	<p>EuroGEMS.org includes additional links to several excellent localised/ foreign-language online resources</p>
	<p>Please note that the Orphanet and European Reference Networks websites (above) are provided in multiple languages.</p> <p>Other resources:</p> <p>England Health Education England: Genomics Education Programme</p> <p>France France Genomique: training</p> <p>Germany Mediathek Humangenetik</p> <p>First German Academy for Further Medical Training on Rare Diseases (FAKSE)</p> <p>Netherlands Cancer genetics referral (Dutch guidelines)</p> <p>Scotland Scottish Genetics Education Network (ScotGEN)</p> <p>Scottish Genomes Partnership</p>	

ETHICAL, LEGAL & SOCIAL IMPLICATIONS

Ethical, legal, social implications of genetics & genomics




This page contains websites that provide information or discussion, relating to the complex ethical, legal and social implications of the latest technological developments.




**Public & Professional
Policy Committee**

This page contains details and links for several recommendations of this expert committee of the European Society of Human Genetics.









**Progress
Educational Trust**

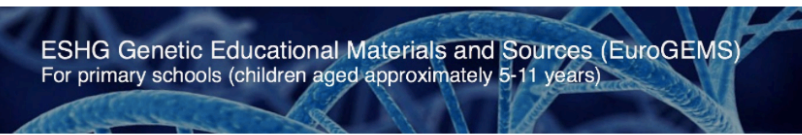


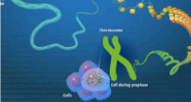
Informing debate on assisted conception and genetics, the Progress Educational Trust's website contains downloadable documents on topics such as genome editing & mitochondrial donation.



**Human Variome Project:
ethics resources**

This page provides many resources relating to ethical, legal and social issues related to the storage and sharing of human genetic data.

 <p>Nuffield Council on Bioethics</p> <p>An independent body that examines and reports on ethical issues in biology & medicine. Downloadable ethical reviews, reports & briefings on e.g. NIPT, genome editing & mitochondrial DNA disorders.</p>	 <p>Clin. Ethics & Law Unit Southampton</p> <p>Several highly thought-provoking pages, including: Ethics of Genomics. Research at the Clinical Ethics and Law Unit at Southampton (CELS) is bridging the gap between current clinical practice and emerging ethical dilemmas.</p>	 <p>HFEA UK</p> <p>The Human Fertilisation and Embryology Authority (HFEA) is the UK Government's independent regulator overseeing fertility treatment and research</p> <p style="text-align: right;">ELSI (continued)</p>
 <p>ASHG Policy & Advocacy Overview</p> <p>A large set of policy documents relating to ethical & legal issues in e.g. Conduct in Genetic Research & Societal Uses of Genetics. Pages provided by the American Society of Human Genetics.</p>	 <p>Global Alliance for Genomics & Healthcare</p> <p>Regulatory and ethics "toolkit" - a set of ethics documents related primarily to genomic data sharing, that are said to be "ready-to-use". Downloadable PDFs with over 12 foreign language translations.</p>	 <p>ELSI program at NIH, USA</p> <p>The Ethical, Legal and Social Implications research program information page at the National Institutes for Health in the USA.</p>

 <p>ESHG Genetic Educational Materials and Sources (EuroGEMS) For primary schools (children aged approximately 5-11 years)</p>			<p>Primary schools page</p>
<p>This page contains useful sources of teaching materials for primary school teachers, involving many innovative and enjoyable interactive activities in connection with several topics relating to DNA, inheritance and cell biology.</p>			<p>A range of exciting & innovative resources</p>
 <p>Science Education Hub - Radboud University</p>	 <p>BBSRC Bioscience for the Future</p>	 <p>NIH - NHGRI Education</p>	<p>Downloadable teaching plans and interactive activities</p>
<p>Based in The Netherlands, this provides innovative, exciting teaching materials, interactive activities, videos & a book series- for primary schools & teachers, encouraging inquisitive & explorative thinking, e.g. on DNA, replication & inheritance. In English & Dutch.</p>	<p>The BBSRC Bioscience for the Future website contains a section (Resources for Primary Schools) that includes downloadable activities, including Discovering DNA, DNA in the Garden and Build your own cell</p>	<p>This Education section of the NIH National Human Genome Research Institute contains a wealth of relevant online resources, including DNA Day primary school activities, fact sheets, teaching plans for teachers and an online education kit.</p>	<p>e.g. "Build your own cell ! " and "DNA Origami "</p>

Primary schools page (continued)

Videos & factsheets



Jeans for Genes - educational resources

This fund-raising but also educational UK website provides professionally recorded videos of children (of different ages), each talking about their rare condition. It also includes general videos on genes and inheritance, with accompanying downloadable factsheets for teachers.

Animated videos



Learn Genetics University of Utah

A range of resources for the classroom with a set of very attractive animated videos (within the section "Tour of Basic Genetics"), including: "What are traits?", "What are DNA and genes?" and "What are proteins?"

Includes "DNA Origami"



Classroom - Genetics Home Reference (NIH)

A large mixed list of resource links for teachers, including a few suitable for primary schools (including "Origami DNA", "Yummy Gummy DNA" i.e. making an edible double helix with sweets and also "Sequence Bracelets"). The list is alphabetical but sub-divided by topic to make it easier to find relevant resources.



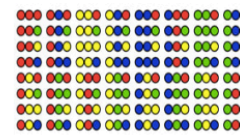
Unlocking Life's Code & the Animated Genome

This site (Unlocking Life's Code) was created in association with the National Institutes of Health, USA. In addition to the film entitled The Animated Genome, it contains other animations and resources for school teachers.



DNA & chromosomes explanatory animation

Relatively simple but wide-ranging animated video commissioned by the BBC, discussing the structure of DNA, chromosomes and even a little about the mysteries of intergenic DNA. Suitable for older primary school children.



Teacher's toolbox Arizona State University

A range of activities, explanations, podcasts, word-search sheets & crosswords, relating to biology, including DNA, genes & cells. It includes DNA pages encouraging children to think about e.g. triplet combinations. Resources are searchable.

Localized/national initiatives

Nederland
[LeveDNA!](#)

Primary schools
Page
(continued)

ESHG Genetic Educational Materials and Sources (EuroGEMS)

For secondary (high) schools (pupils aged approximately 12-18)

On this page, a wide range of useful sources of teaching materials are provided. The numerous information sources that are accessible via the pages described below include many that are highly suitable for use by secondary school teachers. Please click [here](#) for information regarding the European DNA Day Essay Contest.

Page for secondary / high schools



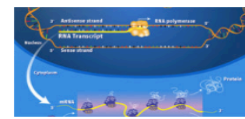
University of Leicester (UK) VGEC

The Virtual Genetics Education Centre (VGEC) provides many topic summaries and lists of linked teaching resources. Resources can be selected by age-range (e.g 11-14, 14-16 & 16-18).



Wellcome Campus: Your genome

This is a large educational website that includes multiple educational 3D animations on DNA replication, gene expression, NGS, sequence data interpretation, gene editing & ethical issues.



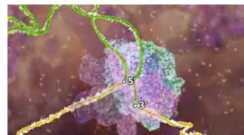
NHGRI-Education (secondary)

The NIH National Human Genome Research Institute's Education Section contains many resources that are suitable for secondary school education including materials on transcriptome, translation, PCR, cloning, DNA sequencing and GWAS.



Genetics Education Centre Univ. of Kansas

An impressively long list of links to genetics educational resources. A varied collection that includes videos and is displayed largely alphabetically.



DNA Learning Center Cold Spring Harbor Labs

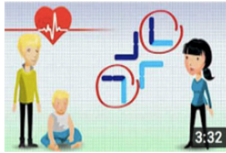
This website includes 3D-animations that relate to transcription, translation and DNA packaging plus the "Gene Screen" app for learning about recessive disorders



BBSRC - secondary schools

This section of the BBSRC Bioscience for the Future website, designed for older school pupils, includes DNA in the Garden (ages 14-16 and post-16) and Stem Cells- Science and Ethics (14-16 years)

**Secondary /
high schools
Page
(continued)**



Understanding genetics and rare diseases

A set of carefully-prepared animations explaining genetics concepts including inheritance mechanisms and various types of chromosome abnormalities. The link provided here is to the full list of animations. Provided by a consultant Clinical Geneticist and University College Dublin, in MULTIPLE LANGUAGES (& most extensively in Maltese).



University of Utah: Epigenetics

Source of information and teaching resources relating to epigenetics, including a downloadable self-assembly model of DNA with histones



BigPicture Education Resources

Wide variety of articles, debates, quizzes, animations and videos relating to genetics (& other fields). High-quality articles. Can filter by resource type & educational level. Funded by Wellcome.

**Secondary /
high schools
Page
(continued)**



Jeans for genes - including videos

This website includes professionally recorded videos of older children with rare conditions, with accompanying downloadable factsheets and classroom activities. It includes resources for KS3 and KS4 (ages approximately 11-16).



Classroom - Genetics Home Reference

A varied collection of resource links, especially for high school teachers categorised by topic (e.g. "DNA Structure", "RNA Basics", "Precision Medicine", "Gene Therapy & Gene Editing" and "Epigenetics"). Many US resources e.g. at Cold Spring Harbor.

**Secondary /
high schools
Page
(continued)**

Localised/national initiatives

Please note that the "Understanding genetics and rare diseases" animations/videos (above) are provided in multiple languages.

Other resources:

Nederland

LeveDNA!

Malta

Understanding genetics - animations (Maltese narrations)

Wales

Wales Gene Park: Genetic Education

Secondary / high schools
Page (continued)

Educational videos on High-schools page

Luke's Story - a Genetic Disorders UK / Jeans for Genes Day film



MORE VIDEOS
🕒 ➔


HOME
FOR PRIMARY SCHOOLS
FOR SECONDARY SCHOOLS
FOR PATIENTS & FAMILIES
FOR UNIVERSITIES & STUDENTS

FOR GENETICS PROFESSIONALS ETHICAL, LEGAL & SOCIAL IMPLICATIONS

ESHG Genetic Educational Materials and Sources (EuroGEMS)


Information for patients and families

This page provides links to sources of information that are primarily designed for patients and their relatives although many will also be useful to senior school pupils and some professionals. The information includes comprehensive A-Z directories of summarised information for numerous genetic conditions (including local contact details) as well as educational leaflets that can be downloaded and printed.




EuroGentest

A series of 15 leaflets providing general information for patients and families about genetics and genetic testing, in multiple languages. Based in Leuven, Belgium.



Contact

A UK charity supporting families, providing an A-Z directory of many genetic conditions and full contact details of support groups.




Orphanet

A consortium of 40 countries, providing summarised info on rare diseases & patient organisations (with contact details in many countries, especially in Europe).

For patients & families


HOME FOR PRIMARY SCHOOLS FOR SECONDARY SCHOOLS **FOR PATIENTS & FAMILIES** FOR UNIVERSITIES & STUDENTS
FOR GENETICS PROFESSIONALS ETHICAL, LEGAL & SOCIAL IMPLICATIONS

For patients & families (continued)




Antenatal Results & Choices

This organisation offers non-directive information & support to parents, before, during & after antenatal screening.



Unique support group

A support group especially for chromosome disorders but also for single gene disorders. Numerous family-friendly, medically-verified, disorder-specific information guides, in several languages.




Genetics Home Reference

A large guide to genetic conditions hosted by the US National Library of Medicine. See the section: "Health Conditions A-Z"


HOME FOR PRIMARY SCHOOLS FOR SECONDARY SCHOOLS **FOR PATIENTS & FAMILIES** FOR UNIVERSITIES & STUDENTS FOR GENETICS PROFESSIONALS ETHICAL, LEGAL & SOCIAL IMPLICATIONS

For patients & families (continued)




Leaflets on cancer genetics

Source of hospital leaflets on cancer genetics with referral guidelines, from the West of Scotland Genetic Services.




Genetic Alliance

This UK charity provides information for patients, families and other individuals who may be interested in genetic conditions.




Clinical genetics leaflets

Source of many leaflets on clinical genetic conditions and genetic testing, from ScotGEN and the South-East Scotland Genetic Service.




Understanding genetic disorders

Animations explaining different types of inheritance patterns e.g. autosomal dominant, autosomal recessive, consanguinity (cousin marriages), X-linked recessive and chromosomal rearrangements. Produced by National Centre for Medical Genetics and compiled by Univ. College Dublin. The explanations are provided in MULTIPLE LANGUAGES.



EGAN - patient network

The "Patients Network for Medical Research and Health", EGAN is an alliance of European disease-specific patient organisations and National Genetic Alliances. Working for a voice in research & health policy, it seeks to improve understanding, effective treatment, & prevention of genetic conditions & also support for the people affected.



EURORDIS Rare Diseases Europe

This is a non-governmental patient-driven alliance of patient organisations representing 862 rare disease patient organisations in 70 countries. They aim to build a strong pan-European community of patient organisations and people living with rare diseases. Provided in MULTIPLE LANGUAGES.

For patients & families (continued)

Localized/national initiatives

Please note that the EuroGentest, Orphanet, Understanding genetic disorders and EURORDIS websites (above) are provided in multiple languages.

Other resources:

Scotland

www.scotgen.org.uk

Scottish Genomes Partnership (SGP)


Nederland

www.erfelijkheid.nl

Prof Edward Tobias
Univ. of Glasgow,
Scotland, UK

HOME FOR PRIMARY SCHOOLS FOR SECONDARY SCHOOLS FOR PATIENTS & FAMILIES FOR UNIVERSITIES & STUDENTS


ETHICAL, LEGAL & SOCIAL IMPLICATIONS



ESHG Genetic Educational Materials and Sources (EuroGEMS)
For universities & students (including genome browsers)


For universities & students

This page contains several powerful websites allowing a user to search for a gene, protein or genomic region, without cost, providing instant access to an enormous amount of up-to-date scientific data from around the world. University students and lecturers are also likely to find other sources to be of interest - particularly those listed within the pages on [Ethical, Legal and Social Implications](#) and information for [Genetics Professionals](#).




Ensembl

A powerful genome browser, based at the Wellcome Trust Sanger Institute, UK. Contains a link to Variant Effect Predictor (VEP), for analysis of likely consequences of DNA variants.



UCSC Genome Browser

The Genome Browser of the University of California Santa Cruz. An easy-to-use website that displays all the options on a single configurable web page.



NCBI

The large collection of linked genome databases at the National Centre for Biotechnology Information, based at the National Institutes of Health, Bethesda.

For universities & students (continued)

HOME FOR PRIMARY SCHOOLS FOR SECONDARY SCHOOLS FOR PATIENTS & FAMILIES

FOR UNIVERSITIES & STUDENTS FOR GENETICS PROFESSIONALS ETHICAL, LEGAL & SOCIAL IMPLICATIONS

For universities & students (continued)

HGVS society

Human Genome Variation Society. Provides helpful guidelines for describing specific variants in DNA, RNA and proteins, according to internationally agreed standards. A relatively brief "simple" summary can be found [here](#).

Genecards


Easy-to-use, provides single-page summary of any gene's location, expression levels, protein function and disease associations.

Orphanet

Gene search page at Orphanet, which provides summarised info on rare diseases and patient organisations.


HOME FOR PRIMARY SCHOOLS FOR SECONDARY SCHOOLS FOR PATIENTS & FAMILIES
FOR UNIVERSITIES & STUDENTS FOR GENETICS PROFESSIONALS ETHICAL, LEGAL & SOCIAL IMPLICATIONS

**For universities & students
(continued)**




HGMD

The Human Gene Mutation Database at Cardiff in Wales, UK, is an attempt to collate all published gene lesions causing inherited disease. The full (professional) database requires a subscription but academics can access a useful but less up-to-date version.



Locus Reference Genomic

A database, supported by EMBL and NCBI, providing non-changing gene and transcript identifiers. Increasingly used by laboratories to determine which specific transcript is to be used when reporting gene variants.



Genome Browser

DECIPHER

DECIPHER, based at the Wellcome Trust Sanger Institute, at Cambridge, UK, provides access to data regarding genes, genomic regions and also disease-related variants that they contain.


Prof Edward Tobias
Univ. of Glasgow,
Scotland, UK

Now:

- Re-checking all links regularly (& updating as required)
- Adding new content e.g. new page for primary care & non-genetic specialists, sub-headings in professionals' page (both now done), & continuing to add more links
- Hoping to add translations of website in the future


New page:

For primary care and non-genetics specialists




ESHG Genetic Educational Materials and Sources (EuroGEMS)
For primary care health practitioners


This page provides links to sources of information that are selected for usefulness to primary care health practitioners. The information includes educational modules (e.g. in Gen-Equip), comprehensive A-Z directories of summarised information for numerous genetic conditions, including local contact details (e.g. in Contact, Orphanet and, in more detail, in GeneReviews) as well as educational leaflets that can be downloaded and printed and apps to assist with cancer genetics referrals.



Gen-Equip




Contact




Orphanet

For Primary Care & non-genetics specialists (continued)




GeneReviews

This database contains a large number of reasonably comprehensive, disease-orientated, reviews. They are generally expert-authored and easy to read, containing clinical features, differential diagnoses and molecular details.




Unique

A support group especially for chromosome disorders but also for single gene disorders. Numerous family-friendly, medically-verified, disorder-specific information guides, in several languages.



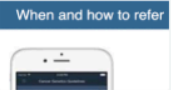
Antenatal results & choices

This organisation offers non-directive information & support to parents, before, during & after antenatal screening.




Inherited cancers

Information on genetic conditions and inherited cancers, provided by Macmillan. It includes information relating to the causes, the risk factors and the possible consequences of test results.



Cancer genetics referrals

Information, web interface and free smartphone apps from Guy's & St Thomas' Hospital, UK designed to assist primary care doctors (GPs) in deciding which individuals should be referred to the hospital Clinical Genetics service.



Clinical genetics leaflets

Source of many leaflets on clinical genetic conditions and genetic testing, from ScotGEN and the South-East Scotland Genetic Service.

Prof Edward Tobias
Univ. of Glasgow,
Scotland, UK

Additional links for non-genetics specialists

Localized/national initiatives

Please note that the Gen-Equip and Orphanet websites (above) are provided in multiple languages.

Other resources:

England
[Health Education England: Genomics Education Programme](#)

Nederland
www.erfelijkheid.nl

[Huisarts en Genetica](#)

Scotland
[Scottish Genetics Education Network \(ScotGEN\)](#)

For Primary Care & non-genetics specialists (continued)

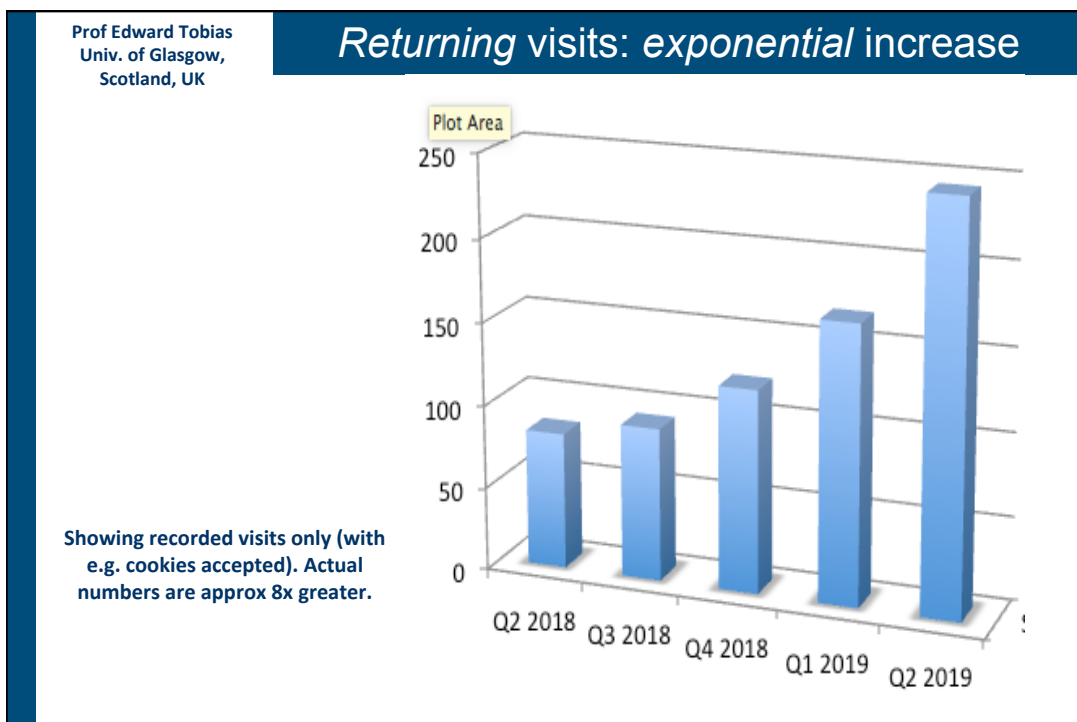
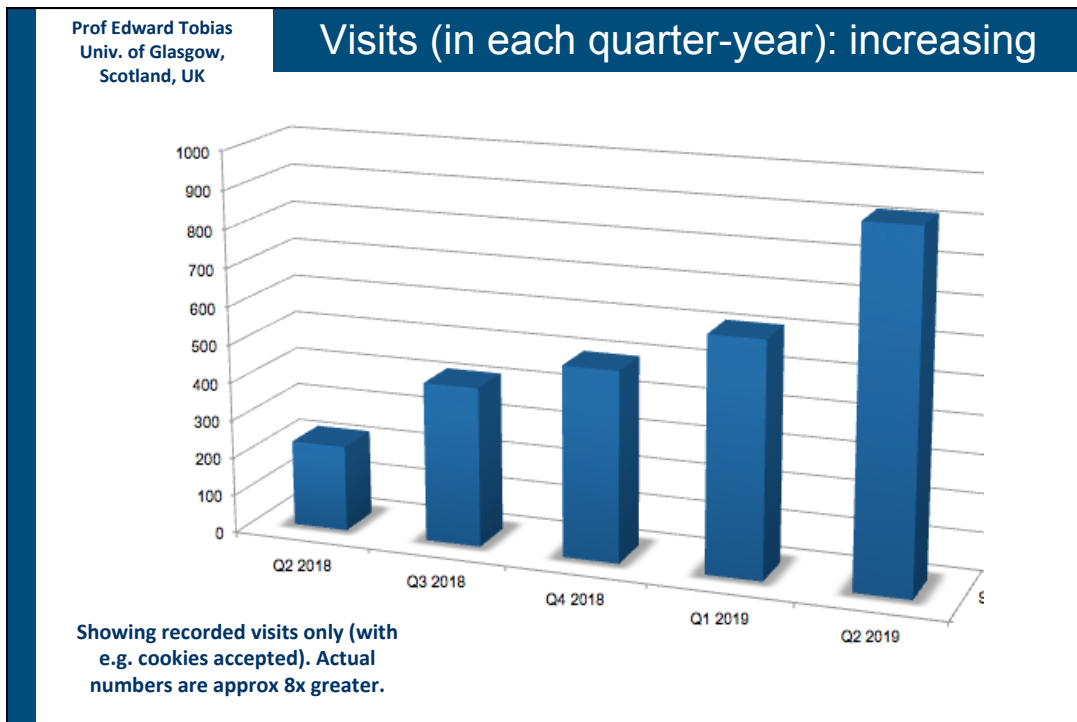
Prof Edward Tobias
Univ. of Glasgow,
Scotland, UK

VISITS

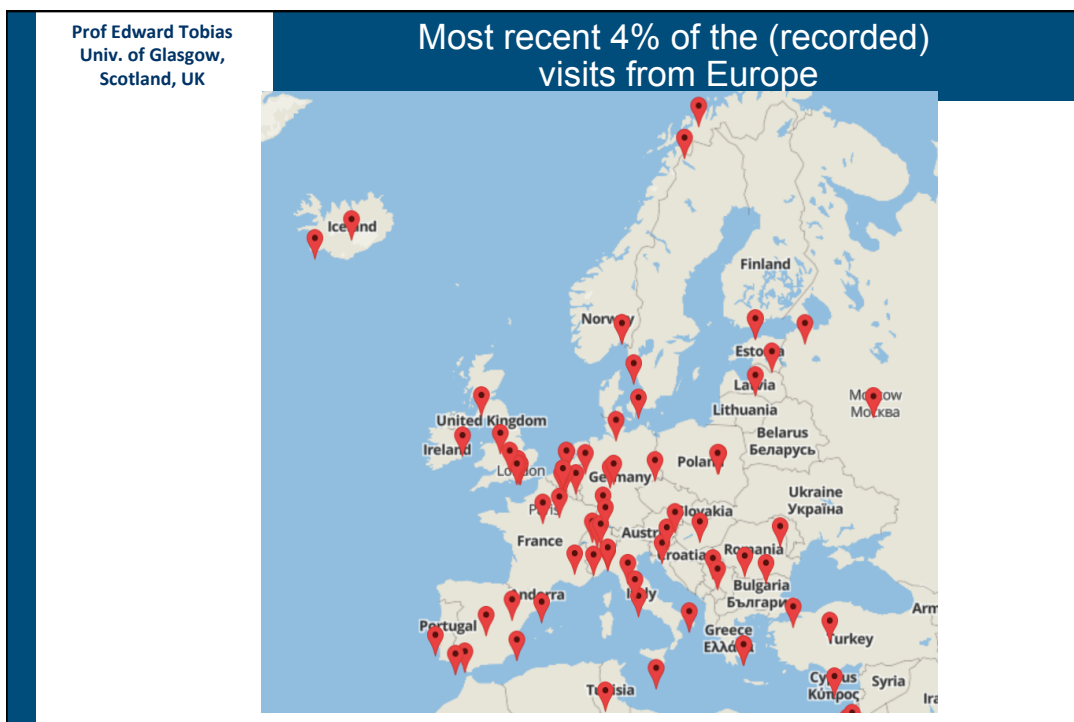
Thousands of visits to website, so far (in 1st year)...
from **92 countries**



A world map where countries are shaded in various shades of blue to represent the number of visits from each country. The map shows a high density of visits from North America, Europe, and parts of Asia and Africa. Greenland and several countries in Africa and South America are shown in light grey, indicating no visits were recorded from those regions.



Prof Edward Tobias Univ. of Glasgow, Scotland, UK			92 countries. Only 12% of total visits are shown
United Kingdom (285 visitors)	Hong Kong (14 visitors)	Mexico (5 visitors)	
United States (128 visitors)	Egypt (13 visitors)	Slovakia (5 visitors)	
Italy (113 visitors)	Austria (12 visitors)	United Arab Emirates (5 visitors)	
France (75 visitors)	Croatia (10 visitors)	Georgia (4 visitors)	
Spain (74 visitors)	Sweden (10 visitors)	Macedonia (4 visitors)	
Belgium (70 visitors)	Saudi Arabia (10 visitors)	Japan (4 visitors)	
Portugal (64 visitors)	Iceland (9 visitors)	New Zealand (4 visitors)	
Germany (63 visitors)	Israel (9 visitors)	Philippines (4 visitors)	
Turkey (56 visitors)	South Africa (8 visitors)	Chile (3 visitors)	
Norway (53 visitors)	Bosnia and Herzegovina (8 visitors)	Algeria (3 visitors)	
Canada (53 visitors)	Serbia (8 visitors)	Taiwan (3 visitors)	
Romania (44 visitors)	Russian Federation (7 visitors)	Vietnam (3 visitors)	
Greece (36 visitors)	Cyprus (7 visitors)	Moldova, Republic of (2 visitors)	
Netherlands (32 visitors)	Slovenia (7 visitors)	Peru (2 visitors)	
Lithuania (29 visitors)	Hungary (7 visitors)	Kenya (2 visitors)	
China (27 visitors)	Latvia (7 visitors)	Iran, Islamic Republic of (2 visitors)	
Poland (26 visitors)	Nigeria (7 visitors)	Tunisia (2 visitors)	
Switzerland (25 visitors)	Denmark (7 visitors)	Qatar (2 visitors)	
Australia (23 visitors)	Argentina (6 visitors)	** (2 visitors)	
Bulgaria (22 visitors)	Estonia (6 visitors)	Europe (2 visitors)	
Ireland (20 visitors)	Pakistan (6 visitors)	Reunion (2 visitors)	
India (19 visitors)	Korea, Republic of (6 visitors)	Ecuador (2 visitors)	
Brazil (17 visitors)	Thailand (6 visitors)	Colombia (1 visitor)	
Finland (17 visitors)	Czech Republic (6 visitors)	Oman (1 visitor)	
		Malaysia (1 visitor)	
		Angola (1 visitor)	







Prof Edward Tobias
Univ. of Glasgow,
Scotland, UK

Examples of university students' (anonymous) feedback comments 2018-2019

- "The website was wonderful for accessing genetics databases and looking up relevant genetic conditions."
- "The EuroGEMS website was particularly useful."
- "We were all very grateful for all of the additional resources"
- "Thank you for igniting my passion for genetics."
- "Excellent. All links have been really useful."
- "If more topics were taught like this then it would make a big difference"
- "It has made me consider pursuing further learning opportunities in this area of medicine."
- "Informative, amazing ... sparked my interest in genetics"
- "Haven't considered it as a career before. Certainly do now"

Written feedback from a secondary school's head of biology department:

- "This website looks terrific. I will certainly share the website within the department and I am confident that it will be very useful indeed."

Prof Edward Tobias
Univ. of Glasgow,
Scotland, UK

Examples of emails from professionals (on ESHG Board & also representatives of National Hum. Genet. Societies)

- “I am really impressed by your terrific work.”
- “Congratulations on making EuroGEMS.org truly beautiful, and comprehensive. I can clearly see how much effort has gone into this.”
- “Thank you once again for the excellent web resource overview you have made!”
- “EuroGEMS.org is great! I think this is absolutely brilliant.”
- “I was amazed! ...How much we can profit from your amazing site.
- “Congratulations on creating an excellent, **comprehensive and highly useful resource for education** on the ESHG webpage!”

Prof Edward Tobias
Univ. of Glasgow,
Scotland, UK

and finally....

EuroGEMS.org is a completely free resource (as are the Clinical Genomics apps)

Thank you to (my son) Adam Tobias, to colleagues on the ESHG Education Committee and to Jerome del Picchia & his colleagues who helped with the website's creation, thank you to the many members of the ESHG Board and the National Human Genetics Societies Committee who contributed to the website's further development and thanks to all NHS & university colleagues in Glasgow (UK) for all of their help and support.

Many individuals on the committees mentioned above have stated that it would be very helpful to raise awareness of these resources. **Therefore please, if possible, mention eurogems.org (& also genomicsapps.org)**

- **to other members of your own national societies,**
- **to other educational organisations**
- **and to colleagues and students.**

Thank you.

edward.tobias@glasgow.ac.uk