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Thalassemia

By Salvatore Stira, ABE Italy

ABE Master Teacher Fellowship Program

AMGEN® Biotech Experience

Scientific Discovery for the Classroom

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If you have questions about any of the curriculum pieces, please reach out to us at ABEInfo@edc.org, and we will be happy to connect you with the author and provide any assistance needed.

AMGEN® Biotech Experience

Scientific Discovery for the Classroom

Italy

ABE Master Teacher Fellowship

Thalassemia

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SUMMARY OF PROJECT IDEA

Students will deepen relationship between scientific research in modern biology and medicine by studying the genetic basis of thalassemia, a form of hereditary anemia quite widespread in Sicily, where I live, and in *all Mediterranean countries.*

After studying the structure and function of the different types of hemoglobin and the genetic mechanisms that lead to the synthesis of the chains of this important protein, students will be asked to investigate through active teaching methods the gene mutations that cause alpha and beta thalassemia. To achieve this goal, bioinformatics tools and LabXchange platform will be used and practical activities will be designed with active participation of students, through the inquiry-based methods.

Then we will study the molecular biology methods used to detect this blood disease. At this stage, in addition to using bioinformatics tools, it will also be possible to use molecular biology equipment such as PCR to simulate mutation identification techniques.

Finally, evolutionary aspects of this disease and potential gene therapy techniques for thalassemia care will be investigated, reflecting also on ethical issues.

Estimated Project Duration: three weeks (9–10 hours)

Student Understandings/Big Ideas:

1) Recent advances in molecular biology have stimulated research and progress in almost all the disciplines of life science, with positive impact also on medicine
2) Bioinformatics, that use specific software and databases, is essential for management of data in modern biology and medicine
3) Understanding modern biology and medicine requires an interdisciplinary approach
4) Small changes in biomolecules makes a big difference in living organisms
5) Diagnosis of many genetic and infectious diseases is now carried out through molecular biology techniques (e.g., PCR)
6) Biological evolution influences the spread of genetic diseases
7) Human DNA modification techniques can potentially cure genetic diseases, but they involve ethical problems

Students will be able to understand:

- the inherited mechanisms of genetic diseases
- effects of DNA mutations on protein synthesis
- structure and functions of different types of hemoglobin molecules
- biology and genetics of thalassemia

Students will be able to:

- compare the effects of different types of mutation on hemoglobin chains synthesis
- assess the importance of molecular biology in the characterization of thalassemia
- explain the main techniques of molecular biology to detect thalassemia
- design an experiment (even virtual) to study thalassemia genetics
- link the geographical distribution of thalassemia to biological evolution
- explain potential application of gene therapy in the care of thalassemia
- explain and assess the ethical issues surrounding scientific research, diagnosis, and gene therapy
- use bioinformatics resources

Assessments:	**Standards:**
Pre-assessments Entry test through computer quiz (es. Kahoot) **Formative assessment** - LabXchange assessment - Observation grids - Work diary - Laboratory reports **Summative assessment** Final test	

Opening "Hook":

1) TV Interview with a thalassemia patient complaining about the reduction of blood donations due to the pandemic by SARS-COV-2

   https://m.facebook.com/watch/?v=872607213211928&__rdr

2) Video call of thalassemia patients during COVID-19 pandemic to invite people to not forget them

   https://www.youtube.com/watch?v=NOHD_c5YJBo&t=3s

Prior Knowledge and Skills:

1) DNA structure and function
2) Protein structure
3) Genetic code
4) Red blood cells
5) Hemoglobin structure and function

Cultural Relevancy and Personal Connections:

Have you ever known a thalassemia patient or a healthy carrier for thalassemia ?

Do you know how many healthy carriers for thalassemia are there in Sicily ?

To encourage students to reflect on the cultural and social aspects of thalassemia. I've included in the project a narrative medicine article, where the author dwells on the social and cultural dimension of thalassemia focusing on the experience of the person and his needs. The article analyzes the biographical narratives of twenty interviewees from which emerges the issue of stigma and how the social representations of thalassemia influenced the labeling process, with important consequences on the quality of life.

https://www.academia.edu/37793810/Io_che_sento_io_che_narro_Talassemia_stigma_e_rappresentazioni_sociali

Moreover, I've included in the project an interview with Dr. Gian Luca Forni, member of the board of the Scientific Society for the Study of Thalassemia and Hemoglobinopathies, and with Marco Bianchi, president of UNITED Onlus, about emotional and personal life sphere of thalassemia patients.

https://www.medicinanarrativa.eu/burden-illness-survey-care-paths-betathalassemia-major-interview-gian-luca-forni-marco-bianchi

Lastly, videos about thalassemia patients' stories have also been included in the project:

https://littlestars.tv/short-films/thriving-with-thalassemia-taabishs-story/

https://rarediseases.org/videos/thalassemia-namithas-story/

https://www.youtube.com/watch?v=NyZgZ3mHd-Q

Learning Activities at a Glance:	
Activity: **Search for information on globin genes and globin gene mutations using bioinformatics tools**	Materials and Resources Needed: • Computer • Internet • Ensembl bioinformatic tools • NIH National Library of Medicine • Article S.L. Thein "The Molecular Basis of β-Thalassemia" https://pubmed.ncbi.nlm.nih.gov/23637309/
Activity: **LabXchange interactive "How Do Restriction Enzymes Cut Plasmids?"**	Materials and Resources Needed: • Computer • Internet • LabXchange • Interactive: *"How Do Restriction Enzymes Cut Plasmids?"* https://www.labxchange.org/library/items/lb:LabXchange:783397ff:lx_simulation:1
Activity: **LabXchange interactive "Application of PCR"**	Materials and Resources Needed: • Computer • LabXchange • Interactive "Applications of PCR" https://www.labxchange.org/library/items/lb:LabXchange:1fb8b9d5:lx_simulation:1
Activity: **LabXchange interactive "PCR primer design"**	Materials and Resources Needed: • Computer • LabXchange • Interactive "PCR primer design"
Activity: **Extraction of human DNA**	Materials and Resources Needed: • Micropipettes • Tips • Microtubes • Centrifuge • Thermostat

Learning Activities at a Glance:	
	• Lucozade HAFW (or similar soft drinks) • Tris-EDTA • SDS • Proteinase K • Sodium chloride • Ethanol • Test tube (5-10 ml) • https://iubmb.onlinelibrary.wiley.com/doi/pdf/10.1002/bmb.20351
Activity: **LabXchange simulation: restriction enzyme digest**	Materials and Resources Needed: • Computer • LabXchange • Simulation "restriction enzyme digest" • https://www.labxchange.org/library/items/lb:LabXchange:1fb8b9d5:lx_simulation:1
Activity: **A practical activity with paper sheets and scissors about use of restriction enzymes to detect mutations**	Materials and Resources Needed: • Computer • Paper sheet with globin gene partial sequences • Pen (or pencil) • Scissors • List of restriction enzyme with digestion sites
Activity: **Laboratory: PCR amplification of beta-globin gene**	Materials and Resources Needed: • Micropipettes • Tips • PCR microtubes • Specific primers • Taq polymerase • Polymerase buffer • Human DNA • Thermocycler • PCR software

Learning Activities at a Glance:	
Activity: **Laboratory: Restriction enzyme digestion of beta globin gene and electrophoresis**	Materials and Resources Needed: • Micropipettes • Tips • Microtubes • Centrifuge • Microwave (or thermostat) • Amplified human beta globine gene DNA • Restriction enzymes SfaNI, MaeI and OxaNI • Restriction enzyme buffer • Agarose • TBE buffer • GelGreen DNA stain • Electrophoresis system
Activity: **Mapping Gene Therapy Concepts**	Materials and Resources Needed: • Internet • Computer (or smartphone) • Poster • CMap software
Activity: **Debate: bioethics implications of gene therapy**	Materials and Resources Needed: • Internet • Computer (or smartphone) • Poster • Markers
Activity: **Creation of maps on spread of beta-thalassemia**	Materials and Resources Needed: • Poster • Printed maps of world, Mediterranean region, and Italy • Transparent paper • Markers • Data on the spread of thalassemia • Data on the spread of malaria

Day 1 (2 hours)

Introduction to thalassemia

In this lesson, the teacher recovers the fundamental concepts of genetics and biochemistry and introduces the students to thalassemia.

Learning Goals:

- Know the structure and function of nucleic acids
- Know the structure and function of protein
- Relate nucleic acids and proteins
- Acquire general information about thalassemia

Assessed Outcome:

- Understand the relationship between the sequence of nucleotides in DNA and the sequence of amino acids in proteins (genetic code)
- Know that thalassemia is a genetic and hereditary disease spread in Sicily

Key Vocabulary: DNA, RNA, proteins, genetic code, transcription, translation, gene, hemoglobin, red blood cells, thalassemia

Materials and LabXchange Pathway(s):

- Interactive Whiteboard
- Videos
- Interactive quizzes (Kahoot)
- LabXchange interactives and simulations

Teacher Preparation: The teacher makes interactive quizzes with Kahoot to test student knowledges about DNA, RNA, proteins, and genetic code and choose LabXchange activities

Sequence

Activity Description	*Time*	*Materials*
1. Interactive quizzes (Kahoot) about DNA, RNA, proteins and genetic code	30 min	• Interactive Whiteboard • Student's individual smartphones Internet
2. What is DNA ? Revision about DNA	15 min	• LabXchange interactive "What is DNA?" https://www.labxchange.org/library/items/lb:LabXchange:1e964d56:lx_simulation:1
3. From DNA to protein Revision about transcription, translation, genetic code	25 min	• LabXchange simulation "From DNA to protein" https://www.labxchange.org/library/items/lb:LabXchange:ae81d54a:lx_simulation:1
4. Hemoglobin structure	15 min	• LabXchange interactive "Protein folding" https://www.labxchange.org/library/items/lb:LabXchange:f93a9e87:lx_simulation:1
5. Opening Hook "What is thalassemia?"	20 min	• TV Interview with a thalassemia patient complaining about the reduction of blood donations due to the pandemic by SARS-COV-2 https://m.facebook.com/watch/?v=872607213211928&_rdr • Video call of thalassemia patients during COVID-19 pandemic to invite people to not forget them https://www.youtube.com/watch?v=NOHD_c5YJBo&t=3s
6. Discussion about videos contents	15 min	

Day 2 (2 hours)

Social and cultural aspects of thalassemia

In this activity students are encouraged to reflect on the social aspects of thalassemia disease through watching videos and reading of an article of medicine narrative

Learning Goals:

- Thinking about social aspects of thalassemia

Assessed Outcome:

- Know social implications of thalassemia
- To become aware about importance of blood transfusions

Key Vocabulary: thalassemia, blood transfusion

Materials and LabXchange Pathway(s):

- Interactive Whiteboard
- Videos
- Smartphone (or computers)
- Printer
- Mentimeter
- Poster
- Markers and glue
- Copies of a narrative medicine article

Teacher Preparation: teacher chooses videos and make copies of narrative medicine article

Sequence

Activity Description	*Time*	*Materials*
1. Reading of a narrative medicine article about social and cultural dimension of thalassemia	40 min	• Research paper about social aspects of thalassemia: *Raffa, V. Salute e Società, XVI, suppl. 3/2017 "Io che sento, io che narro.. Talassemia, stigma e rappresentazioni sociali" (I feel, I teel. Thalassemia, stigma and social representations)* https://www.academia.edu/37793810/Io_che_sento_io_che_narro_Talassemia_stigma_e_rappresentazioni_sociali
2. Watching videos: 1) interview to Dr. Forni about emotional and personal life sphere of thalassemia patients; 2) videos about thalassemia patients stories	30 min	• Interactive Whiteboard • Videos: Interview to Dr. Forni https://www.medicinanarrativa.eu/burden-illness-survey-care-paths-betathalassemia-major-interview-gian-luca-forni-marco-bianchi Videos about thalassemia patients' stories https://littlestars.tv/short-films/thriving-with-thalassemia-taabishs-story/ https://rarediseases.org/videos/thalassemia-namithas-story/ https://www.youtube.com/watch?v=NyZgZ3mHd-Q
3. Implementation of shared wordcloud about social and cultural dimension of thalassemia	10 min	Smartphones (or computers) Mentimeter

Activity Description	*Time*	*Materials*
4. Creation of a poster about social and cultural dimension of thalassemia	40 min	Poster, printer, glue, markers

Day 3 (1,5 hours)

Mutation of Beta-globin gene

In this lesson, the effects of mutations on the beta-globin gene, that cause beta-thalassemia, are studied.

Learning Goals:

- Knowledge of various types of mutations
- Use bioinformatics resources
- Understand the effect of various types of mutations on expression of beta-globin gene

Assessed Outcome:

- Finding through bioinformatic resources beta-globin sequence
- Knowing how to read the beta-globin gene sequence
- Compare the effects of different types of mutation on hemoglobin chains synthesis

Key Vocabulary: mutation, genetic code

Materials and LabXchange Pathway(s):

- Smartphone (or computers)
- Internet
- Printer
- Protein synthesis interactive model
- LabXchange simulation: mutation

Teacher Preparation:

- Download and print protein synthesis interactive model sheets
- Choose some beta-globin mutations for activity
- Prepare and print thalassemia-sickle cell anemia comparison table

Sequence

Activity Description	*Time*	*Materials*
1. Learn effect of mutation on protein synthesis through a LabXchange simulation	30 min	• Computer • Internet • LabXchange simulation: mutations https://www.labxchange.org/library/items/lb:LabXchange:f1ec1b5b:lx_simulation:1
2. Search for information on beta-globin gene and globin gene mutations that cause thalassemia using bioinformatics tools	30 min	• Ensembl bioinformatic tools • NIH National Library of Medicine • Article: *S. L. Thein "The Molecular Basis of β-Thalassemia"* https://pubmed.ncbi.nlm.nih.gov/23637309/
3. Research of most common beta-globin gene mutations in Sicily	10 min	• Smartphones (or computers) • Internet
4. Study of effect on protein synthesis of various mutations in beta-globin gene	30 min	• Protein synthesis interactive model (print or digital version) es. https://biology-roots.com/store/Protein-Synthesis-Model-p289254034)
5. comparison between thalassemia and sickle cell anemia through a comparison table written by students	20 min	• Textbook • Comparison table

Day 4 (2 hours)

Preparatory activity to amplification of human beta.globin gene

In this lesson the students learn how to amplify genes through PCR technique

Learning Goals:

- Understand PCR technique
- Know potential applications of PCR technique
- Project beta-globin gene amplification

Assessed Outcome:

- Realizing that PCR is now one of the most widespread method of analyzing deoxyribonucleic acid (DNA)
- Knowing to describe the principal applications of PCR in life science
- To be being able to project (through IBSE method) an experiment to amplify beta-globin gene

Key Vocabulary: DNA, PCR, amplification, primers, Taq polymerase, diagnostic

Materials and LabXchange Pathway(s):

- Interactive Whiteboard
- Internet
- LabXchange interactives
- Beta-globin gene sequence
- Free PCR Primer Design Program

Teacher Preparation: organize IBSE activity (project of an experiment to amplify beta-globin gene)

Sequence

Activity Description	*Time*	*Materials*
1. Study of PCR tecnique and its applications	40 min	• LabXchange interactive "PCR mechanism" https://www.labxchange.org/library/items/lb:LabXchange:f7f6962a:lx_simulation:1 • LabXchange interactive "PCR applications" https://www.labxchange.org/library/items/lb:LabXchange:bc4846e2:lx_simulation:1
2. Project, using IBSE method, an experiment to amplify human beta-globin gene	40 min	• School notebook • Poster (or blackboard)
3. Design of human beta-globin gene PCR primers: a) learn a general method to design PCR primers; b) use a free PCR Primer Design Program to design human beta-globin PCR primers	40 min	• LabXchange interactive PCR primer design https://www.labxchange.org/library/items/lb:LabXchange:2960e770:lx_simulation:1 • PCR primer design program https://primer3.ut.ee/

Day 5 (1 hour)

Extraction of human DNA

In this lesson students extract their DNA using a simple method

Learning Goals:

- Understand physical and chemical properties on which it is based dna extraction

Assessed Outcome:

- Obtain own DNA with a sufficient degree of purity

Key Vocabulary: DNA, protein, extraction

Materials and LabXchange Pathway(s):

- Micropipettes
- Tips
- Microtubes
- Centrifuge
- Thermostat
- Lucozade HAFW (or similar soft drinks)
- Tris-EDTA
- SDS
- Proteinase K
- Sodium chloride
- Ethanol
- Test tubes (5-10 ml)

Teacher Preparation: Organize lab activities

Lab safety: Students must use lab coat, latex gloves, and disposable materials

Sequence

Activity Description	*Time*	*Materials*
1. Extraction of human DNA Lab protocol: https://iubmb.onlinelibrary.wiley.com/doi/pdf/10.1002/bmb.20351	60 min	• Micropipettes • Tips • Microtubes • Centrifuge • Thermostat • Lucozade HAFW (or similar soft drinks) • Tris-EDTA • SDS • Proteinase K • Sodium chloride • Ethanol • Test tubes (5-10 ml) • Scientific article about human DNA extraction *Hearn, R. P., & Arblaster, K. E. (2010). DNA Extraction Techniques for Use in Education. Biochemistry and Molecular Biology Education, 3, 161-166.* https://iubmb.onlinelibrary.wiley.com/doi/pdf/10.1002/bmb.20351

Day 6 (2 hours)

Polymerase chain reaction

In this lab activity human beta-globin gene is amplified through Polymerase chain reaction technique. Then, PCR products are analyzed through DNA electrophoresis.

Learning Goals:

- Know how to apply the PCR technique
- Analyze experimental data

Assessed Outcome:

- amplify human beta-globin gene through PCR
- identify whether the amplification has been successful through DNA electrophoresis

Key Vocabulary: DNA, PCR, amplification, electrophoresis

Materials and LabXchange Pathway(s):

- Micropipettes
- Tips
- PCR microtubes
- Specific primers
- Taq polymerase
- Polymerase buffer
- Human DNA
- Thermocycler
- PCR software
- Agarose
- TBE buffer
- GelGreen DNA stain
- Electrophoresis system

Teacher Preparation: prepare laboratory experiences

Lab safety: students must use lab coat, latex gloves and disposable materials

Sequence

Activity Description	*Time*	*Materials*
1. Human Beta-globin gene amplification through Polymerase chain reaction	70 min	• Micropipettes • Tips • PCR microtubes • Specific primers • Taq polymerase • Polymerase buffer • Human DNA • Thermocycler • PCR software
2. Prediction of PCR products size (group activity)	10 min	
3. Analysis of PCR products through DNA electrophoresis	40 min	• Agarose • TBE buffer • GelGreen DNA stain • Electrophoresis system

Day 7 (1 hour)

Restriction enzyme and mutations

In this lesson, through a practical activity with paper sheets and scissors, the students learn how restriction enzyme can be used to detect mutations

Learning Goals:

- Know restriction enzyme mechanism
- Understand how restriction enzyme can be used to detect mutations

Assessed Outcome:

- Predict how many DNA fragments are formed after digestion with one or more restriction enzymes

Key Vocabulary: DNA, restriction enzyme

Materials and LabXchange Pathway(s):

- Interactive Whiteboard
- Internet
- LabXchange interactives
- Paper sheet with globin gene partial sequences
- Pen (or pencil)
- Scissors
- List of restriction enzyme with digestion sites

Teacher Preparation: print copies of beta-globin gene sequence and copies of restriction enzyme list

Sequence

Activity Description	*Time*	*Materials*
1. Study how restriction enzymes cut DNA	20 min	• LabXchange interactive "how do restriction enzymes cut plasmids? https://www.labxchange.org/library/items/lb:LabXchange:783397ff:lx_simulation:1
2. A practical activity with paper sheets and scissors about use of restriction enzymes to detect mutations	40 min	• Computer • Paper sheet with globin gene partial sequences • Pen (or pencil) • Scissors • List of restriction enzyme with digestion sites

Day 8 (2 hours)

Analyze mutations in beta-globin gene

In this lab activity students learn how restriction enzymes can be used to detect mutations in human beta-globin gene through lab experiments

Learning Goals:

- Understand how restriction enzymes can be used to detect mutations

Assessed Outcome:

- Predict how many DNA fragments are formed after digestion with one or more restriction enzymes in wild-type and mutate

Key Vocabulary: DNA, restriction enzyme, mutation, electrophoresis

Materials and LabXchange Pathway(s):

- Micropipettes
- Tips
- Microtubes
- Centrifuge
- Microwave (or thermostat)
- Amplified human beta globine gene DNA
- Restriction enzyme SfaNI
- Restriction enzyme buffer
- Agarose
- TBE buffer
- GelGreen DNA stain
- Electrophoresis system
- Scientific articles about restriction enzymes digestion of amplified human beta-globin DNA:
  1) *Atweh, G. F., Forget B.G. (1986). Identification of a beta-thalassemia mutation associated with a novel haplotype of RFLPs. American Journal of Human Genetics, 38(6):855-9.* https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1684858/pdf/ajhg00155-0065.pdf

2) *Pirastu, M., Ristaldi M. S. and Cao, A. (1989). Prenatal diagnosis of beta thalassaemia based on restriction endonuclease analysis of amplified fetal DNA. Journal of Medical Genetics 26, 363-367.* https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1015619/pdf/jmedgene00056-0011.pdf

Teacher Preparation: set up experiments; find an alternative activity if there are difficulty in experiments

Sequence

Activity Description	*Time*	*Materials*
1. Digestion with some restriction enzyme of amplified human beta-globin DNA. Choice of restriction enzyme is based on following scientific articles: a) https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1015619/pdf/jmedgene00056-0011.pdf b) https://pubmed.ncbi.nlm.nih.gov/3014869/	60 min	• Micropipettes • Tips • Microtubes • Centrifuge • Thermostat • Amplified human beta globine gene DNA • Restriction enzyme SfaNI, MaeI and OxaNI • Restriction enzyme buffer • Scientific articles about restriction enzymes digestion of amplified human beta-globin DNA: *1) Atweh, G. F., Forget B. G. (1986). Identification of a beta-thalassemia mutation associated with a novel haplotype of RFLPs. American Journal of Human Genetics, 38(6):855-9.* https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1684858/pdf/ajhg00155-0065.pdf *2) Pirastu, M., Ristaldi M.S. and Cao, A. (1989). Prenatal diagnosis of beta thalassaemia based on restriction endonuclease analysis of amplified fetal DNA. Journal of Medical Genetics 26, 363-367* https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1015619/pdf/jmedgene00056-0011.pdf

Activity Description	*Time*	*Materials*
2. Electrophoresis of digested with restriction enzymes and undigested amplified beta-globin DNA samples. During electrophoresis run students predict experimental results	40 min	• Agarose • TBE buffer • GelGreen DNA stain • Electrophoresis system • Microwave • Micropipettes • Tips • Microtubes

Day 9 (2 hours)

Gene therapy

In this lesson the students learn gene therapy techniques and their potential applications

Learning Goals:

- Understand principal gene therapy techniques
- Know potential applications of gene therapy on genetic diseases

Assessed Outcome:

- Knowing to describe gene therapy and principal applications of this technique in medicine

Key Vocabulary: DNA, gene therapy, virus, genetic disease

Materials and LabXchange Pathway(s):

- Internet
- Computer (or smartphone)
- Interactive Whiteboard
- Poster
- Markers
- CMap software

Teacher Preparation: organize groups activities; make copies of text about gene therapy

Sequence

Activity Description	*Time*	*Materials*
1. Watching videos about gene therapy	15 min	• Interactive Whiteboard • Video1 “cos’è la terapia genica?” (what is gene therapy?) https://www.youtube.com/watch?v=nFF714BWBYk • Video 2 “Un viaggio alla scoperta della terapia genica” (A voyage into gene therapy) https://www.youtube.com/watch?v=nUhOZf1_l_k

Activity Description	*Time*	*Materials*
2. Reading text about gene therapy from Fondazione Mutagens site	25 min	• Review on gene therapy by Fondazione Mutagens https://mutagens.it/informati/terapia-genica/
3. Group activity: the class is divided in four groups, each of them write a list of words about gene therapy; then, with a guidance of teacher, the class choose a shared word list.	20 min	• Review on gene therapy • Notebooks • Blackboard
4a. A group of student make a conceptual map about gene therapy using C-Map program	60 min	• Computer • Internet • C-map program
4b. A group of student draw a conceptual map about gene therapy in a poster	60 min	• Poster • Markers

Day 10 (1 hour)

Conference on gene therapy

A researcher from an excellence medical center in the city where the school is located will be invited to talk about a trial on thalassemia gene therapy treatment.

Learning Goals:

- Understand how gene therapy can be applied to thalassemia cure.

Assessed Outcome:

- Students must be able to explain expectations and limitations of thalassemia gene therapy treatment

Key Vocabulary: DNA, gene therapy, thalassemia

Teacher Preparation: set up experiments; find an alternative activity if there are difficulty in experiments

Sequence

Activity Description	*Time*	*Materials*
1. Conference of a doctor from "Piera Cutino research institute" of Palermo, who talks about a trial on thalassemia gene therapy treatment.	50 min	
2. Questions from students to the speaker	10 min	

Day 11 (2 hours)

Gene therapy bioethics

In this lesson the students debate about bioethics implications of gene therapy

Learning Goals:

- Understand bioethics implications of gene therapy

Assessed Outcome:

Students will be able to explore a bioethical issue by learning about the risks and potential outcomes involved in actual gene therapy trials.

Key Vocabulary: gene therapy, bioethics

Materials and LabXchange Pathway(s):

- Internet
- Interactive Whiteboard
- Copies of a study case
- Computers (or smarthpones)

Teacher Preparation: make copies of case study; prepare a list of sites about gene therapy bioethics

Sequence

Activity Description	*Time*	*Materials*
1. Watch clips of a video about gene therapy	30 min	Video "Ethical challenges in novel gene therapies for sickle cell disease" https://elsihub.org/video/ethical-challenges-novel-gene-therapies-sickle-cell-disease

Activity Description	*Time*	*Materials*
2. Read case study on gene therapy research	25 min	Learning Guide "*Gene Therapy Research & the Case of Jesse Gelsinger*" created by NYU Langone's High School Bioethics Project https://med.nyu.edu/departments-institutes/population-health/divisions-sections-centers/medical-ethics/education/high-school-bioethics-project/learning-scenarios/jesse-gelsinger-case
3. Debate: bioethics implications of gene therapy Using Debate methodology students discuss about bioethics implications of gene therapy. Students are divided in two groups, pro e cons gene therapy.	20 min	Smartphones Internet

Day 12 (1 hour)

Evolutionary aspects of beta thalassemia spread

In this lesson the students create maps thalassemia spread in the world and study evolutionary aspects of beta thalassemia spread

Learning Goals:

- Learn evolutionary aspects of genetic diseases spread

Assessed Outcome:

Students will learn that thalassemia is spread only in some areas and that this genetic disease is related to the spread of malaria

Key Vocabulary: evolution, natural selection, epidemiology, malaria

Materials and LabXchange Pathway(s):

- Printed maps of world, Mediterranean region, and Italy
- Transparent paper
- Markers
- Data on the spread of thalassemia
- Data on the spread of malaria

Teacher Preparation: search data on the spread of thalassemia and malaria; prepare copies of maps

Sequence

Activity Description	*Time*	*Materials*
1. Divide class in two groups: one draws a map of the spread of thalassemia on paper and the other draws a map of the spread of malaria on transparent paper	40 min	• Printed maps of world, Mediterranean region, and Italy • Transparent paper • Markers • Data on the spread of thalassemia • Data on the spread of malaria
2. The two groups overlap the two maps and determine if there is a correlation between the spread of malaria and thalassemia	20 min	Maps of the spread of thalassemia and malaria
3. Analysis of results and discussion	20 min	

ABE Master Teacher Fellowship: Curriculum Project Presentations

17-19/20 May 2022

AMGEN® Biotech Experience

Scientific Discovery for the Classroom

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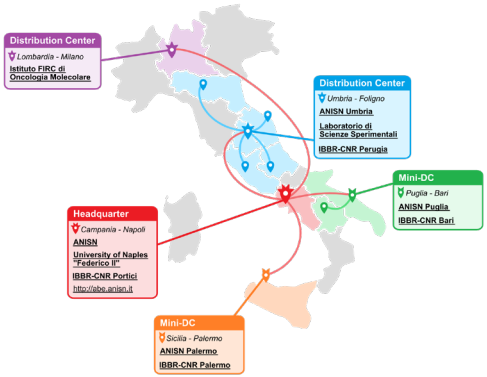
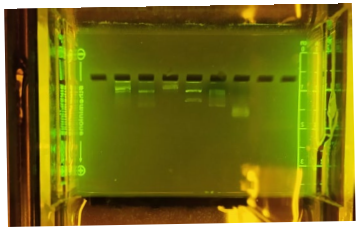
Natural Sciences and Chemistry teacher
Benedetto Croce Scientific High School - Palermo, Italy



Lifelong learner
Inquiry educator
Trekking
Nature

ABE Italy



ABE teacher since 2020

My students: 99 (7 classes: 1th to 5th)

ABE students: 33

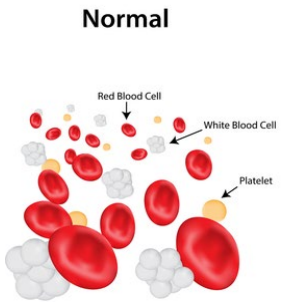
AMGEN® Biotech Experience
Scientific Discovery for the Classroom

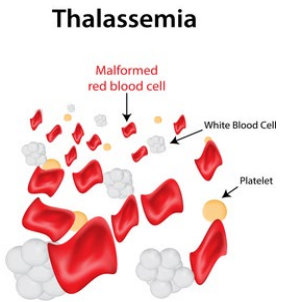
Thalassemia

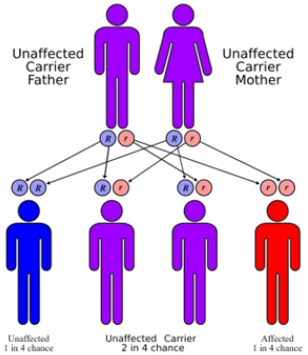
In my opinion, it is essential that students fully understand the relationship between basic research and medicine.

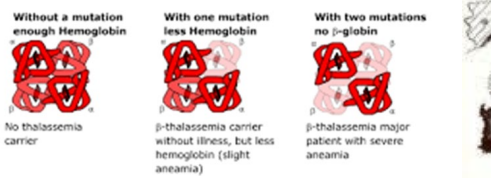
To achieve this goal, I decided to choose as a topic the **genetics of thalassemia**, a form of hereditary anemia quite widespread in my region.

People with thalassemia produce either no or too little hemoglobin, the protein molecule in red blood cells that carries oxygen. The disorder results in excessive destruction of red blood cells, which leads to excessive tiredness and fatigue.









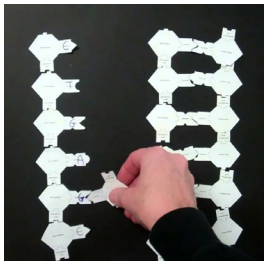
Project goals and activities

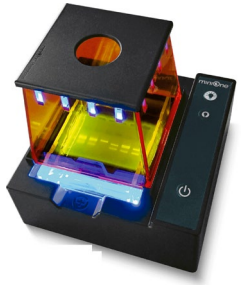
Students will be understand:

- inherited mechanisms of genetic diseases
- effects of DNA mutations on protein synthesis
- relationship between protein structure and function (especially in hemoglobins)
- biology and genetics of thalassemia

Students will be able to:

- explain the main techniques of molecular biology to detect and characterize genetic diseases
- design an experiment (even virtual) to study thalassemia genetics
- link the geographical distribution of thalassemia to biological evolution
- explain potential application of gene therapy in the care of thalassemia and other genetic diseases
- explain and assess the ethical issues surrounding scientific research, diagnosis and gene therapy
- use bioinformatics resources





Activities:

- practical activities (inquiry-based methods)
- bioinformatic tools and virtual simulations (LabXchange platform)
- molecular biology techniques (electrophoresis, PCR, etc.)
- debate on bioethical issues

AMGEN® Biotech Experience

Scientific Discovery for the Classroom

Learnings from research

What new ideas or information have you learned about this topic by working on this curriculum project?

- Study of thalassemia is an excellent model of the fact that different mutations can lead to the same phenotype (unlike sickle cell anemia, often used as an example in school textbooks)
- Thalassemia provide varying levels of resistance to malaria and are proposed to have emerged as an adaptive response to malaria in Africa and Mediterranean regions.
- Gene therapy is a viable cure for beta thalassemia. Adding a functional gene to defective blood stem cells is a successful therapy for patients with severe beta thalassemia

Describe any help you have received from others (Fellows, Amgen staff, other experts, Program Office).

- It was very helpful to compare myself to other Fellows to improve my project. It was a good idea to split the Fellows group into nests.
- My fellowship advisor (Dr. Chip Stark) has been valuable in giving me suggestions on specific aspects of my project, such as bioinformatics resources, clinical trials and bioethics.
- Amgen staff provided useful materials (videos, articles, links, etc.) to develop my project

Overall, how has the Fellowship helped you develop this curriculum?

Fellowships allowed me to deepen my knowledge and skills in science education, making my project more effective from an educational point of view