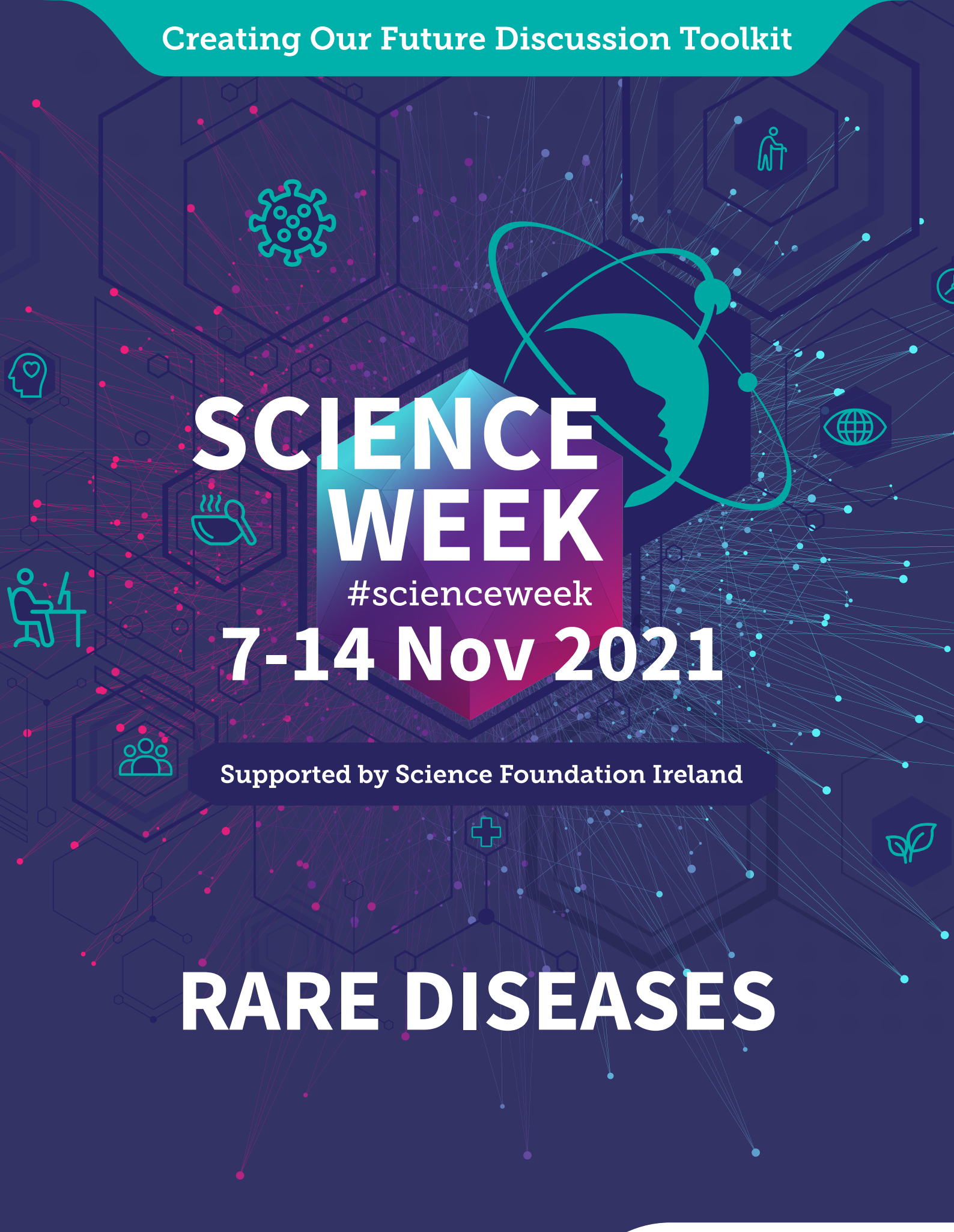
Creating Our Future Discussion Toolkit



RARE DISEASES

Science Foundation Ireland sfi For what's next

CREATING OUR FUTURE

ABOUT CREATING OUR FUTURE

Creating Our Future is a Government of Ireland campaign to stimulate a national conversation between everyone in Ireland on their ideas on how to make our country better for all. While we might not always think about it, research and innovation affect nearly every part of our lives. The COVID-19 pandemic in particular has shown the importance and value of research and innovation in our lives – medically, socially, politically, and economically. Ideas are the starting point for all research and innovation. Anyone, anywhere, can have an idea that inspires research and innovation. It could be based on an opportunity or challenge that someone has identified in their own life, for their community, for Ireland or for the world. Or it could be based on a topic that someone is curious or passionate about.

Creating Our Future wants to hear them all – in particular from our young people. All ideas will be captured and will help inform the future direction of research and innovation in Ireland.

Secondary school students are invited to take part in this national conversation on research. Research carried out in the present will have an impact on your future, and you have the opportunity to have your say on research that will help to make a better future for all in Ireland.

Some of us are excited to go back to exactly the way things were before the pandemic, others have found recent times to be a relief and want to keep the changes we've experienced, others want to hold on to some of the changes but not all of them. Our impact on the environment has improved in so many ways, such as air quality, and yet deteriorated in others, such as the use of single-use plastics. Remote working is the new norm for a significant number, this has directly impacted on quality of life in the positive and the negative for so many, for others it has resulted in a serious hit to livelihoods as footfall in office areas drop. Throughout this journey, the public has been largely united in turning to science for the data to inform decisions for today, and to bring hope for tomorrow.

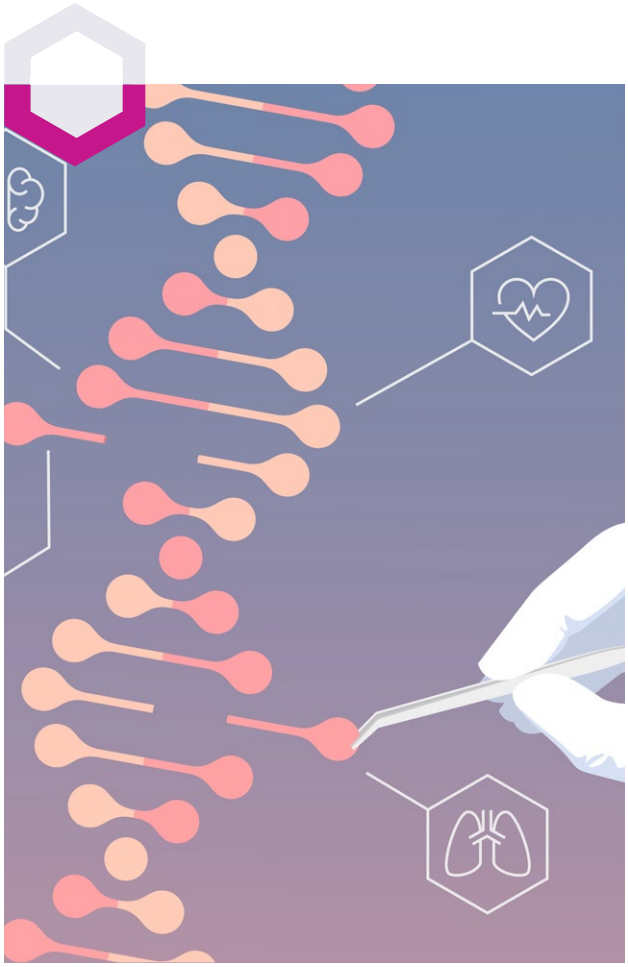
Use these discussion toolkits in the weeks leading up to Science Week to debate and discuss STEM topics with your class that will affect our future, from changes in the fashion industry to using technology to enhance our cities and communities.

During Science Week, submit your own idea to creatingourfuture.ie to help researchers in Ireland to create a better future.

This toolkit provides background and discussion stimulus on rare diseases, including what rare diseases are, what can cause them and some ways in which science and technology can help. You'll also read case studies from families in Ireland who are affected by rare diseases.

There are video and news sources linked throughout the document to help stimulate the conversation.

INTRODUCTION AND BACKGROUND



What is DNA?

Put simply, genomics is the study of an organism's genome – its DNA – and how the information encoded within DNA is used to build the organism. All living things, from single-celled bacteria, to multi-cellular plants, animals and humans, have DNA.

Genuity Science (2020)

Developed in collaboration with:

genuity science

SFI Centre for Research Training in GENOMICS DATA SCIENCE

What is a genome?

- All living things have a genome made up of DNA, which contains the instructions to build and repair our bodies. DNA has four building blocks, which we refer to by the letters A, T, C, and G and our genome has more than 3 billion pairs of these letters arranged in a very precise sequence
- Every person's genome is around 99.9% the same as everyone else's, but that 0.1% equates to around 3 million differences.
- Some differences in our DNA determine physical characteristics, such as eye colour. Others can influence our chance of developing a disease.
- We can now sequence DNA (determine what those DNA letters are and what order they appear in) and analyse genomic information to inform healthcare, helping to better diagnose, treat and even prevent disease

What are genes?

Genes are sections of DNA which are the basic units of inheritance. This means that genes determine what traits are passed down from a mother and father to their child.

Humans have around 20,000 'coding' genes, which contain the information to build and regulate proteins – essential for building and repairing our bodies. Some genes determine physical characteristics, such as eye colour. Others can influence the chance of developing a health condition, such as cystic fibrosis.

- Genes account for 2% of our genome. We are only staring to discover what the other 98% of the genome does.

What is a Rare Disease?

What do Rett Syndrome, Motor Neurone Disease (MND), Huntington's Disease and Genetic Parkinson's Disease have in common? They are all classified as rare diseases.

Rare diseases are characterised by their relatively low prevalence (less than 1 in 2,000 people in the EU). Collectively rare diseases affect around 6% of the population in the Republic of Ireland, accounting for at least 300,000 individuals. There are between 18-30 million people with a rare disease in the EU.

Despite major advances in research in recent decades, it is increasingly recognised that there are massive gaps between translating advances in rare disease knowledge into potential medication.

And the cost can be insurmountable, which can contribute to restricting or delaying the availability of new and innovative drugs worldwide, particularly in poorer countries, but also increasingly in richer countries that are struggling to meet these very high costs. Availability can also be restricted or delayed by the way new and innovative drugs are assessed.

Although rare diseases affect relatively small numbers of people, it is estimated that 350 million people globally have a rare disease, more than double the number of patients affected by AIDS and cancer combined.

Developed in collaboration with:

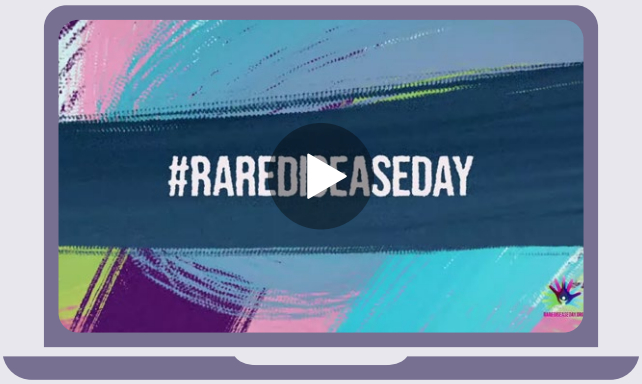
Rare Diseases Ireland

Rare Ireland Family Support Network

future neuro

DISCUSSION STIMULUS

Rare Disease Day takes place every year to raise global awareness. Every February, this global, patient-led awareness campaign brings together millions worldwide. This video profiles six people living with different rare diseases around the world. For more information, go to www.rarediseaseday.org



Source: YouTube - rarediseaseday

You can also have a look at the following video, which was filmed in Dublin with members of the Irish Rare Disease community!

Source: YouTube - eurordis

Epilepsy Lighthouse Project

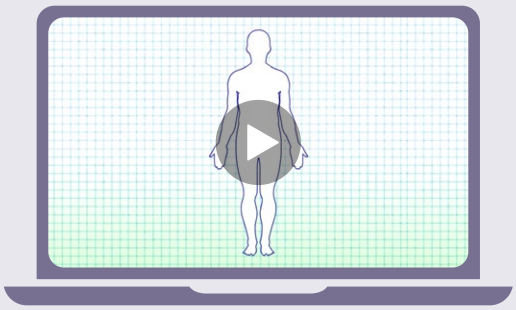
Katie Cooke is a 20-year-old competitive runner from Dublin. Due to an aggressive form of epilepsy, she experiences up to 12 seizures a day. Despite her condition, Cooke hasn't let anything stand in the way of her love for running. With the help of the Pisces phone app, both she and her treatment team can keep track of her healthcare and improve her diagnosis and treatment. Do you think technologies such as these could help people living with rare diseases?

Source: YouTube - Epilepsy Ireland

Understanding Chromosomal Translocation

This animation explains Chromosomal Translocation which occurs when one chromosome becomes attached to another, sometimes leading to genetic problems. This is one of a set of animations provided by a consultant Clinical Geneticist and University College Dublin.

Translocation is just one example of a chromosome disorder, others include duplication and deletion. There are videos detailing other examples available on the UCD School of Medicine YouTube Channel



Source: YouTube - UCD Medicine

Read the below case study about Mary, Isla and their family for a real life example of chromosomal translocation

I'm Mary, a member of Rare Ireland Family Support Network, an organisation set up in 2017 to support families of children living with rare conditions.

I have two brothers with special needs. It was suspected that one of my brothers had a genetic condition, so we went for genetic testing. It turns out that my mother and I have a balanced chromosome translocation. This means a piece from chromosome 4 has transferred or swapped places with a piece from chromosome 8. (You would have seen this in the animation above). There is no loss or gain of chromosome material but instead two pieces have swapped seats. There is another type of Chromosome Translocation, called unbalanced translocation. This is when the amount of chromosome material present is incorrect, meaning there is a gain or a loss of a chromosome.

Someone with a balanced translocation usually has no health or developmental problems, although they may face difficulties if they want to have children. As I have a balanced chromosome translocation I had a 30% chance of my child having an unbalanced chromosome translocation.

Mary's daugther Isla

When I was pregnant with Isla, I had a test which confirmed she had a normal 4 chromosome but her chromosome 8 has a leg with an extra bit from chromosome 4, meaning she had an unbalanced chromosome translocation.

When Isla was born she was checked daily by the doctors, and they found nothing out of the ordinary. Once home she fed and grew well, pretty much like any other new-born.

Isla's condition means she has moderate intellectual delay. She processes information slowly. She also has a severe language delay. She finds it hard to understand words. That said she is slowly improving. Last year she started in our local preschool. She has a 1:1 assistant. She has just started her second year and will go to special school next year. We hope this tailored environment will help Isla continue to progress.

Medically everything is going well, she has a yearly check up with her doctor and physically she is good too, although she can be a bit unbalanced.

Beyond my own family I've not met another family in Ireland with Isla's condition. Her doctors hadn't ever heard of it. This can make the journey a little lonely and hard going. Rare Ireland offers so much support in the day to day. There is always someone that can offer advice on the different journey through accessing services or just general support.

Why Do We Want to Know About Genomes?

Genomics and genomic medicine are very much in the spotlight. A genome is a complete set of genetic instructions. But what can genomics tell us about ourselves? And what are clinicians and healthcare scientists looking for when they analyse someone's genomic information? In this video, Dr Michelle Bishop from Health Education England's Genomics Education Programme introduces us to a range of reasons why different people and professions want to know about genomes.

Source: Vimeo - Genomics Education Programme

Rare Disease Case Study

I'm Laura, one of the founders of Rare Ireland Family Support Network, an organisation set up in 2017 to support families of children living with rare conditions.

In October 2000 my first child was born, knowing what I know now the signs of a chromosome disorder were there from the start, but she remained undiagnosed for almost 11 years. Alanna was born at 38 weeks 5 days, despite being full term she only weighed 5lb 4oz, she had very low muscle tone, so she was floppier than the average baby, she had no suck reflex so was unable to drink a bottle and needed to be tube fed. She stayed in the NICU for 2 weeks before being allowed home. Things for Alanna in her early years were very difficult, she failed to thrive and meet any of her developmental milestones.

In April 2011 Alanna was the second child in Ireland to be diagnosed with a rare chromosome disorder called Koolen de Vries syndrome – or KDVS for short. On the long arm of one chromosome Alanna is missing approximately 5 genes. 3 of her missing genes are not thought to be of much significance in regard to development.

One of the missing genes is responsible for healthy development. The deletion of this gene causes moderate-severe intellectual disability, developmental delay, epilepsy, low muscle tone, hypermobile joints, Autism, ADHD, significant speech and language delay, feeding difficulties in infants, low immune system, anxiety,

Laura and Alanna

sleep disorders, OCD, renal and heart defects, distinctive facial features, brittle hair and nails, foot deformities, dental abnormalities i.e. missing adult teeth, vision impairment.

Another gene deleted works to suppress neurological conditions like Parkinson's disease, Alzheimer's and Dementia. This gene is currently being researched in relation to the effects of it being deleted but it is believed that the absence of this gene increases the risk of KDVS individuals developing these neurological conditions in later life.

Laura and Alanna with President Michael D. Higgins

Having KVDS does not necessarily mean having all the characteristics of Koolen de Vries syndrome. People living with KDVS have extremely amiable personalities, they are a pleasure to be around. They love to entertain and make people laugh. Alanna who is almost 21 is no exception to this, she struggles on a daily basis with things we all take for granted and will continue to require support throughout her life, but this never defeats her. She is a ray of sunshine and is loved by everyone who meets her.

Being a parent of a child who is undiagnosed, like Alanna was for almost 11 years, is a lonely journey but nothing compares to the loneliness and uncertainty of being a parent of a child with a condition so rare it's unknown to most doctors. Not ticking any boxes in regard to the supports available is very daunting. Rare Ireland was set up to offer the support that's been lacking and pave the way for our children's siblings so that their future as advocates is more certain. Family to family support is invaluable, knowing you're not alone on your journey has been a saving grace to over 1.3k families across Ireland in the last 4 years.

Guiding Questions

- What is your understanding, having read the resource, of what a rare disease is?
- What are the challenges that you think scientists and researchers would face when looking for treatment for a rare disease?
- Should human gene editing be allowed?
- What are the ethical issues involved in editing out disabilities such as chromosomal disorders?
- How do you think technology can play a role in treating rare diseases? And/or how do you think technology can play a role in helping a person living with a rare disease?
- What do you think scientists could do to help people with rare diseases?

Additional Resources

Rare Diseases Ireland is a patient advocacy national alliance for voluntary groups representing people affected by or at risk of developing a rare disease.

Read more

Europe invested over €1.8 billion in more than 320 collaborative research and innovation projects through the framework programmes related to rare diseases.

Read more

EuroGEMs is the European Society of Human Genetics Education Materials website, providing a wide range of teaching and learning resources related to genetics and genomics: https://www.eurogems.org

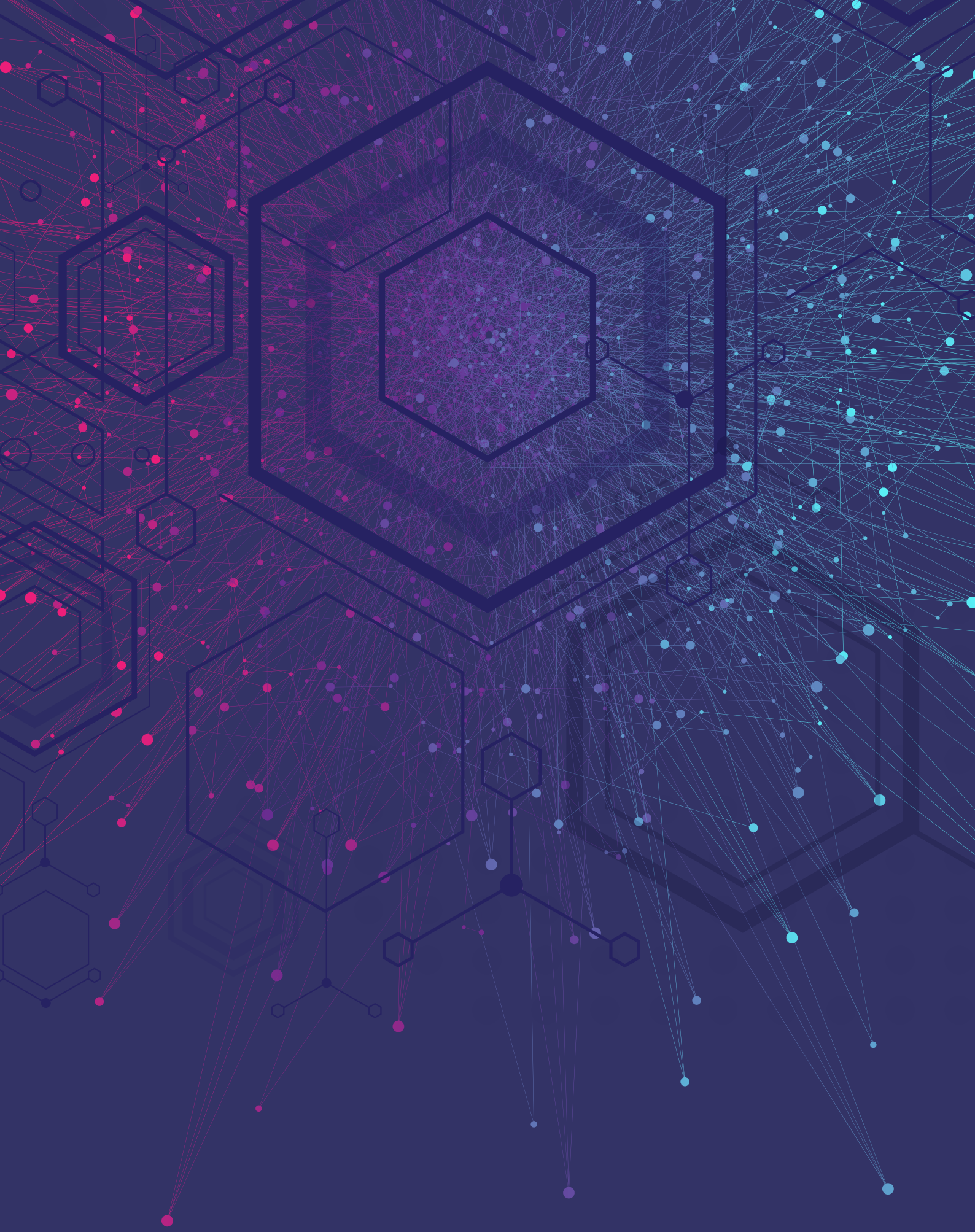
Health Research Charities Ireland - An Easy Guide to Rare Diseases in Ireland and Consensus for Action.

Read more

Ireland's Biggest Brainstorm - have your ideas heard!

Now that you have discussed rare diseases, take five minutes to think of an opportunity or challenge you see for yourself, your community, Ireland or the world on this topic.

These can be captured in the classroom using this template and emailed to creatingourfuture@sfi.ie or if you have access to a computer lab log on to creatingourfuture.ie and submit your idea directly on the website.



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