RAREDUCANDO DIGITAL EDUCATION

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E- Course -LESSON 2

RARE DISEASES: Database, BIBLIOGRAPHY AND BEST PRACTICES

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(TRANSCRIPTION -TEXT VIDEO)

Hello everyone, I am Giulia Volpato, contact person of patients within the European Reference Networks of ERN SKIN. President of p63 EEC International Syndrome. Expert in advocacy, privacy and bioethics.
Today we will discuss together the very briefly issue of the right to health and how this is related to the dignity of each individual and, consequently, of every child and every student.
Surely your children or your students have protected, well protected, their dignity, their freedom. But let us understand together how health is understood and protected internationally. We will also see a brief overview of what is the support network for you parents, for you teachers, for all the training and information that in my opinion, is fundamental to be able to approach the topic of rare diseases, the smaller ones. Let us therefore start from a brief presentation on the right to health at international level.

Then the introduction to the rare disease. As has been said, I will not only speak to you as chairman of the Association of Patient Representatives but also as a patient myself. To start dealing with our subject, let’s try to understand what health means, what the right to health is, and where we see the right to health protected.

First of all, we see here Article 1 and then Article 25 of the Universal Declaration of Human Rights, which quotes “all human beings are born free and equal in dignity and rights”, as I said earlier, “everyone has the right to a standard of living sufficient to guarantee health and well-being, with particular regard to the necessary medical care and social services, and gives the right to home safety, in the event of illness and disability”.

This is crucial.
Therefore, the Universal Declaration of Human Rights gives us the basis for the concepts that we will see shortly, namely dignity, freedom, security, protection, necessary social services, health and well-being.
We move to a further international scene and see how many documents are certainly not all, but they are only an example of the Constitution of the World Health Organisation of '46, Article 1 of the Universal Declaration of Human Rights that we have seen.
St. Josè’s Declaration, perhaps less known, but equally important, the Universal Declaration on the

Human Genome.

They're all records. Here’s the last one. It is therefore written to the UN declaration in 2022, which specifically mentions and protects the rare disease as such. As a result, we see that at the international level, at the European level, there are many legal bases that protect health.

Let us remember that a healthy person, a healthy teacher, a healthy parent, a healthy child, now we will see the definition of health, allows himself, and others, a higher quality of teaching and time spent in the classroom, consequently greater social integration.

Let us now see the Italian Constitution. The Italian Constitution will analyze it with regard to two articles: Article 13, therefore, is the one that speaks, and deals with personal freedom and Article 32. Let us therefore see, just like the Italian Constitution, protects the health of the individual in every aspect.

It is essential to remember how for the Italian State, but in general, for the European State, the State cannot violate the life of the person. This is in relation to no impact of freedom.

But let’s see now what sickness means. Is there a definition of disease? No. A widely shared definition of disease does not exist. There are interpretations like this. Therefore, disease means alteration of the physiological-psychological state of an organism that reduces or negatively alters its normal functions.

Here we see the concept of “normal”, functionality, the concept of “own”, its normal functionalities and above all I emphasize an alteration of the physiological and psychological state. So by illness we must not only mean colds, headaches, wheelchairs, but all the difficulties that our son or student faces on a physiological and psychological level.

So considering in the future, let’s see now if there is a definition of health for health. The opposite of illness is therefore understood as a state of complete physical, mental and social well-being from which it consists not only in the absence of illness or infirmity. Possession of the best possible fact of health is a fundamental right of every human being.

This is Article 1 of the World Health Organisation. So what does it tell us, what is health for health? We must understand complete physical, mental and social well-being. I would say that in this first statement the school and the family play a fundamental role. We spend, a good part of our time at school and family.

As a result, the most complete well-being, physical, mental and social balance are essential to guarantee the right to health. In fact, let’s see how it does not mean only the absence of disease or infirmity, being healthy. Physical, mental and social well-being must be ensured.

What brings our students, with difficulties or disabilities and problems with something we cannot give an answer to or for teachers because nobody explains it to us; or something that we can't give an answer to as parents, because no one tells us.

We report our child, our student to be a child or a student, no longer a symptom, illness, disability, someone to be teased, to be removed, to marginalize when we remember and when we protect two fundamental rights the right of dignity, where the patient, the student, the child should not be seen simply as an illness to be cured, but as a person with possibilities, abilities, rights and duties.

We learn together, therefore, as teachers and as parents to try to look the other way, to put themselves in the shoes, to take care of someone and something. Let’s try to look at the class from the point of view of those who live it from a wheelchair. Let’s try to think about the class from those who live it by listening and not being able to see, we try to think about the context class through the sensations of those who can only see, can't listen.

Try to think of a social familiar text of these children, of these students and try to understand, putting on the other side, what are their remote abilities and possibilities. We consider our students and children, as children and not as illness. Once we consider our child in their dignity, we also manage to protect them according to fundamental right, freedom. Freedom has the right of each individual to decide independently in relation to his or her own person or on behalf of the child and caregiver.
What does that mean? That if a child expresses his own malaise, his own difficulty, let’s listen to him, let’s put ourselves in his shoes. We try to give him his decision-making space, making him express what he has in his freedom. My freedom ends where yours begins. We do not limit the freedom of our children and students, because by protecting the principle of dignity and freedom we can guarantee them a daily social of well-being, equality and inclusion.

Once this very small premise I mention only for information, then we will see it in the next lesson, which can be within the world of rare diseases institutional networks to support you and reference.

Let’s see here the logo of the Italian Ministry of Health, obviously for each country and for each European State we will have to go and analyse what are the guidelines of your State. For example, for Greece, we will analyse what are the guidelines, directions and institutions present in Greece.
For Italy, we have a national network of rare diseases that can certainly be a great starting point if there is not really the starting point to have all the information, the support and the necessary indications both for your parents and for your teachers.
The national network for rare diseases is based in Rome. It was born in 1998, it is obviously coordinated by the Ministry of Health, assisted by the Higher Institute of Health, from which was born in ‘98 precisely the National Centre for Rare Diseases, born in’98 through national health plans that later developed with the pacts in the State-Region, then each Region to its own. Regional Coordination Centre for Rare Diseases, which can be contacted for medical, therapeutic and diagnostic information, including at the level of school integration. What can be useful tools for your students? What can be the aids? What are aids? How to recommend a parent to use aids for their child, to improve the educational quality of their child? All this information can be found at the regional centers for rare diseases that obviously collaborate with local health companies, the Ulss of the various districts that are also supporting you, then in the following elections, you will see in detail where to go and request information, obviously regarding the National Center for Rare Diseases and original coordination of rare diseases, if you enter the CNMR Centro Nazionale Centro Nazionale Rare Diseases or Disease.gov, or the Higher Institute of Health. Find all links. All the information that can be of support to you to support your child or student in turn.

A very small flash also on the European Reference Network for Rare Diseases. We need to know that 24 centers have been set up throughout Europe, in 2000 and in 2017. They are centers of HUB, specialized in symptoms and not for pathology therefore for evidence and not for disease and even if they may be of your support.

They are called European Reference Networks, so European reference networks if you type on Google European Reference Networks you find the 24 Ern within which you can find information related to diseases and contacts, even specialists.
I'll do a bit, the story, exactly of the ERNs are 24 and we will then see it in detail in the following

lessons.

I take this opportunity to talk to you, give you a very small hint of what can be the tools that can be a little more practical that you can have at a technological level, so on the Internet to find out the information that can help you in understanding how to organize a dedicated lesson, also aimed at students with rare illness such as approaching, addressing the topic of rare illness in the classroom at school, which we will deepen much more than the subsequent elections and the type of psychological impact, can have a rare disease than class and also in parents of the rest of the grace.

So I re-share the screen for a moment to show you in detail some databases that can definitely support you in this task,

Here we go. Let’s start with ORPHANET. This is the orpha.net address Written Orphanet with the “PH”. It is a French organization based in France that has now developed throughout Europe with its national headquarters, which has always carried out, carried out and coordinated a very widespread communication, very correct and very detailed on what is the reality of every single rare disease.
Epidemiological reality, hence where it originates; Symptomatic, therefore the symptoms without neglecting, however, what are the networks and networks fundamental for information and support to the family and school. I'll show you for a moment what can be the tools for you.

Here are all the activities proposed by orpha.net and followed by orpha.net, the one that interests us first; so, the list of rare diseases cataloged by pathology, symptom, description and everything else. Here on the left you can find search filters that can give you indications, for example classification, gender by disability.

To search for your disease in the page search bar you can type or the name of the disease, if you know it, I have some symptoms.
I write EEC in my case, my pathology. You see that typing the name, some indication, the portal automatically gives you all the results inherent in that pathology. Let us remember that we are talking about rare diseases, diagnosed and not diagnosed. So the results could be as many as in this case.

Eec syndrome and let’s look down, see we have four results. So we found four sources of certified information, because within the orpha.net site we only have certified information, but if we go to open we don't have all four, now we go to open the tabs.

We will see that in the case of EEC syndrome the information reported will be almost the same in terms of degrees. It is not the same syndrome that has palatoschisis or eec syndrome without the palatoschisis, syndrome and just, eec syndrome and related diseases. We have, for example, EEC syndrome, now we'll see something together.

Here is how we can imagine, in fact, that there is some information about categorisation that interests us relatively with regard to school, while there is below, in fact, a summary that can certainly help you to better identify the difficulties of your students or your children.

In this summary there is some medical information, but absolutely understandable to all. Remember that the portal is aimed at families, sick people and even doctors or specialists experienced in the field; so, the language used is a transversal language at all. Below, however, in addition to the summary of the pathology, there is also other information.

You see, for example, we can go and see what are associations, certainly in the case of very rare particular diseases a fundamental landing point to which you could and should get if the patient associations like we are, are able to give you all the indications related to the diseases you see here how the imported associations you see the flag are at an international level.

So all associations that have joined the Orphanet Project program within the Orphanet program are reported in correspondence to their own pathology. I saw before we were there and wait till I am, we are the second I saw. I'm coming back. Here we go. So if we click on P63 EEC.

Here they are, to which you can possibly write for directions and information, also because not to any particular teaching methods dedicated to those children and those diseases.
Another portal we see is the Eurordis portal.

It is the European Federation for Rare Diseases, recognised by the European Commission. Works in strong synergy for the advocacy of the patients themselves. So for the protection of the rights of rare patients, he was also a strong promoter of the interests of rare patients in the management, in the drafting of the statement of the UN report published in early 2022.
So now she's really doing a great deal of policy work on rare diseases. Here too, this portal offers you many topics, lots of ideas, of course, for example, what are rare diseases? What does it mean to live with a rare disease? And if any of you might be interested there is also the link services and training within which you can enter, registering leaving your contacts you can join many Eurordis training projects. EURORDIS works a lot with dedicated academy, both patients and family members or in this case teachers.

So it's very useful. The information is very clear. And certified. I'll show you the latest material that could be very useful in my opinion. You can find it in the bibliography. I'll show you the latest material that can be very useful. I found it in a biography. It is a kind of guidance guide and is written by the Higher Institute of Health, precisely by the National Center for Rare Diseases in 2013. Somewhat dated, but it is still deeply topical and in this case it is absolutely useful to teachers as it is "education for use and methods and materials to talk about rare diseases in primary school".

Okay, scrolling through the summary and I repeat, you find everything published in the bibliography, you will see that from page 14 there are these activity sheets. Okay, it's divided by class, class one, and second part. Find it. There it is. Just the educational material, useful to talk to your students about rare illnesses.
Obviously calibrated according to age and we will then see in subsequent lessons how these activities that can be the proposals also in a different way, are also present in our format rare-educating digital education. We'll explain a little later, but here it is not asking for the file. There are all indications suitable to properly treat the topic of rare diseases at school.
I thank you for your attention. We have really done a little excursus on what is the basic instrumentation at your service; those who can be the contacts at your service to give quality and correct information, special attention to your children, to your students.

For further materials, you will be provided in the course of the next lessons. Thank you.

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