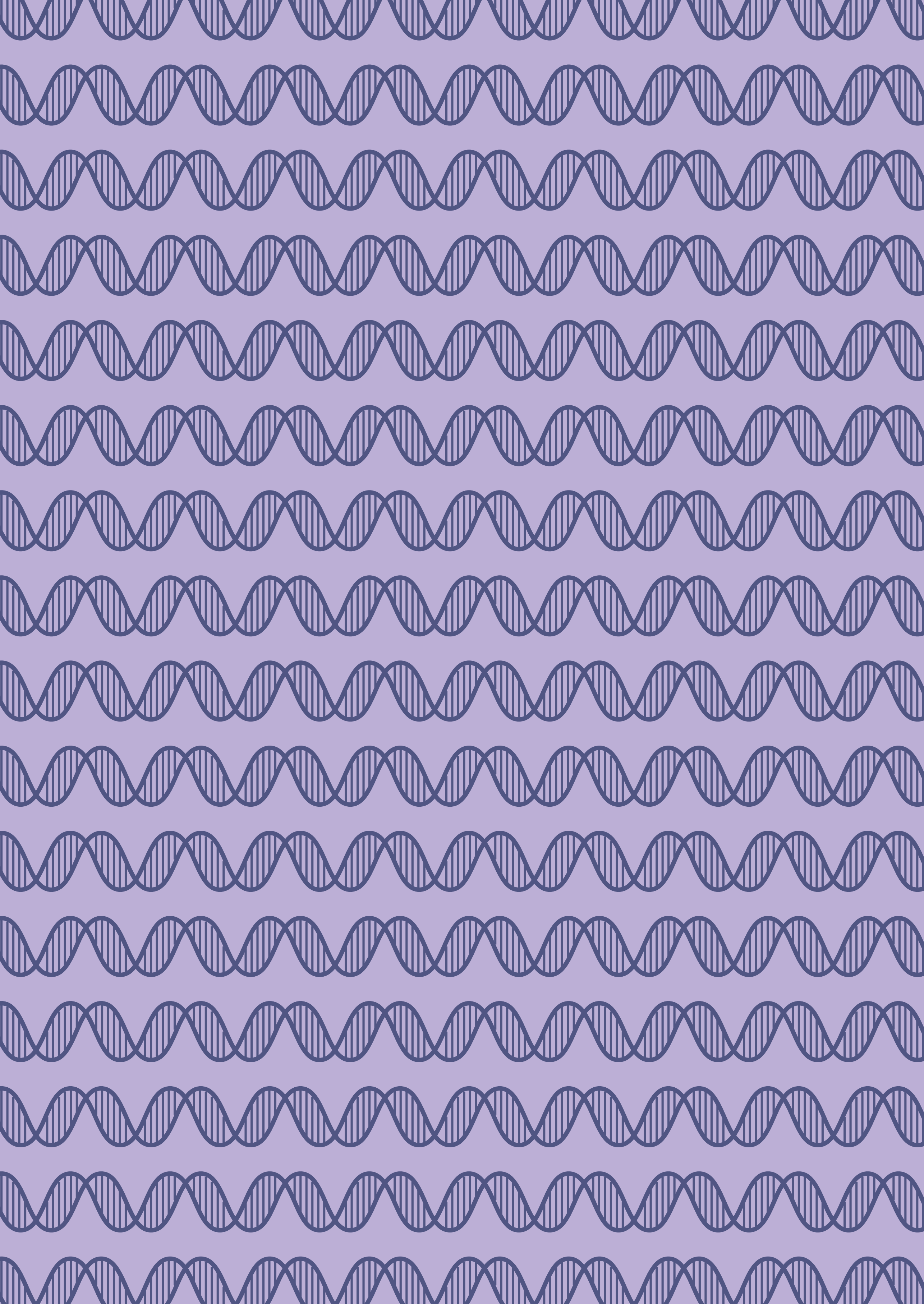
Albinism

– just a gene!

Gudrun A. Rappold



Albinism
–
just a gene!

Gudrun A. Rappold

Foreword

When I was born in Nigeria, my parents were very surprised to see me. My colour was in stark contrast to their rich brown skin and black hair. They were quickly informed by the doctor who helped to give birth to me, that I probably had albinism. The doctor wrote in my first hospital card: “albino?” You see, even the doctor herself questioned whether I had albinism. Like my parents, she knew few people with albinism and was not aware of the variety that exists even within albinism. Years later, in my twenties, I was the one who finally explained the genetics of albinism to my parents, and only because someone had published some information about albinism on the internet.

My parents did an excellent job loving, educating, and raising me. However, I wish they had more support. I wish my parents, family and friends had a book like Albinism: just a gene, when I was born. They would have learned details about the genetics of albinism in a simplified yet comprehensive way. Perhaps they would have been better prepared to support my visual impairment which was a major issue for them and for me. Maybe they would have been more informed to explain my condition to those who harassed me for being black but white in appearance.

However, I am happy and highly encouraged that this book will equip many people with albinism on how to move the topic of albinism from the realm of rumors, false information, and superstition into the realm of facts, science, and genetics. This is particularly important in many parts of Africa where false information about albinism and why it occurs, remain widespread, threating the peace and security of people with albinism and their families. Therefore, it is my wish that this book will reach many people across Africa. This way, we will continue to stem the tide of ignorance surrounding this beautiful genetic condition.

Finally, I wish to encourage all people with albinism and their family members to see the publication and dissemination of books like this as another helpful facilitator of the new dawn of meaningful inclusion of people with albinism and their family members in African culture and society at large.

Ikponwosa “I.K.” Ero

Lawyer, Human Rights Advocate, and the First
United Nations Independent Expert on Albinism (2015-2021)

I was deeply moved by this book, written by Professor Rappold, as I have been dealing with the topic of albinism in Africa for many years. I am, like Professor Rappold, repeatedly shocked by the futility of violence – particularly the mutilation and murder of children and adults with albinism in the southern countries of Africa. It is hard to believe that superstition has spread so widely. Even today, babies born with albinism are murdered and hundreds of people cannot live in safety – all because of a genetic defect they are born with. However, that the origin of this violence is in superstition strengthens my hope that early education can combat this and save affected people from this cruel fate in the future.

In the book “Albinism – just a gene!”, the origin of albinism and how it develops are vividly described and, in the course of it, how inheritance works is explained. Genetics determines our appearance. But as human beings, we learn from our actions. The book also talks about working together, respecting each other, and being there for each other. Professor Rappold emphasizes the valuable support that self-help groups can give to affected people. As the patron of an umbrella organization comprising many hundreds of self-help groups, I can only confirm this. If we unite, we can make a big difference – and this is how this cross-border friendship project was born. On my travels through various African countries, I have met many amazing and committed people, who stand up for the rights and medical needs of their fellow human beings, including people with albinism and their families. The initiative described in this book is another building block to helping people with albinism.

I wish the initiative every success and very much hope that it will make an important contribution to educating and clarifying what albinism is and preventing crimes in the future.

Eva Luise Köhler

Patron of the Alliance of Chronic Rare Diseases (ACHSE) and Chairwoman of the Board of Trustees of the Eva Luise and Horst Köhler Foundation for People with Rare Diseases

**In Europe, a disease is considered rare if no more than 5 in 10,000 people show the same clinical symptoms. 80 percent of rare diseases are inherited.*

1

All people are equal. No matter if you are young or old, thick or thin, black or white – all people are equal. No matter how you look, everybody has the same right to be treated with respect and tolerance. All people are equal and yet they are different. Diversity is good. Diversity makes our life colourful and interesting.

2

Everybody looks a bit different. Some people are short, others are tall. Some have fair skin, others have dark skin. If you have parents with fair skin, you will likely end up as a child with fair skin, too. If your parents are black, you will likely be black, too. If one of your parents is white and the other black, you will end up as a mix in between. You will look a little bit like your mom and a little bit like your dad. This is a basic rule in Genetics.

3

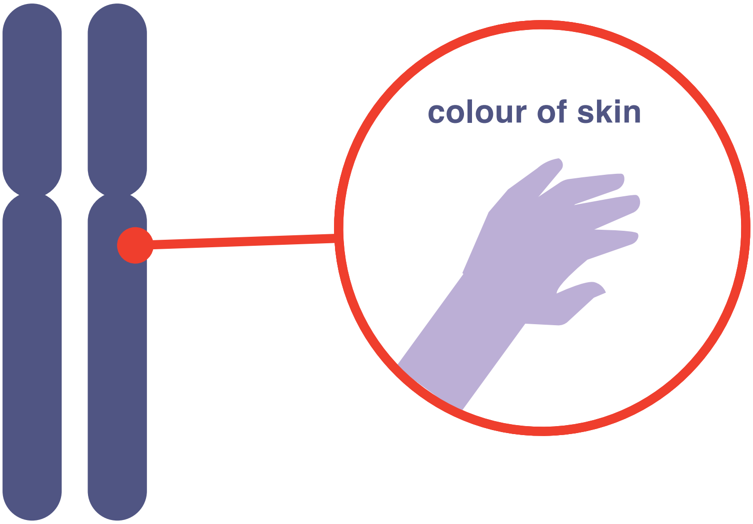
It was a monk, Gregor Mendel, who had a lot of spare time and found all this out about inheritance when experimenting with different sorts of peas in his monastery garden. The rules of inheritance that he found work out for all living things, not only peas or other plants, but also for animals and humans.

All families have some special traits that run in the family. They may have a special talent for composing music or playing a certain sport. Or they may have a higher probability of having a hot temper, going insane or having a heart attack. There are many different traits that can run in a family.

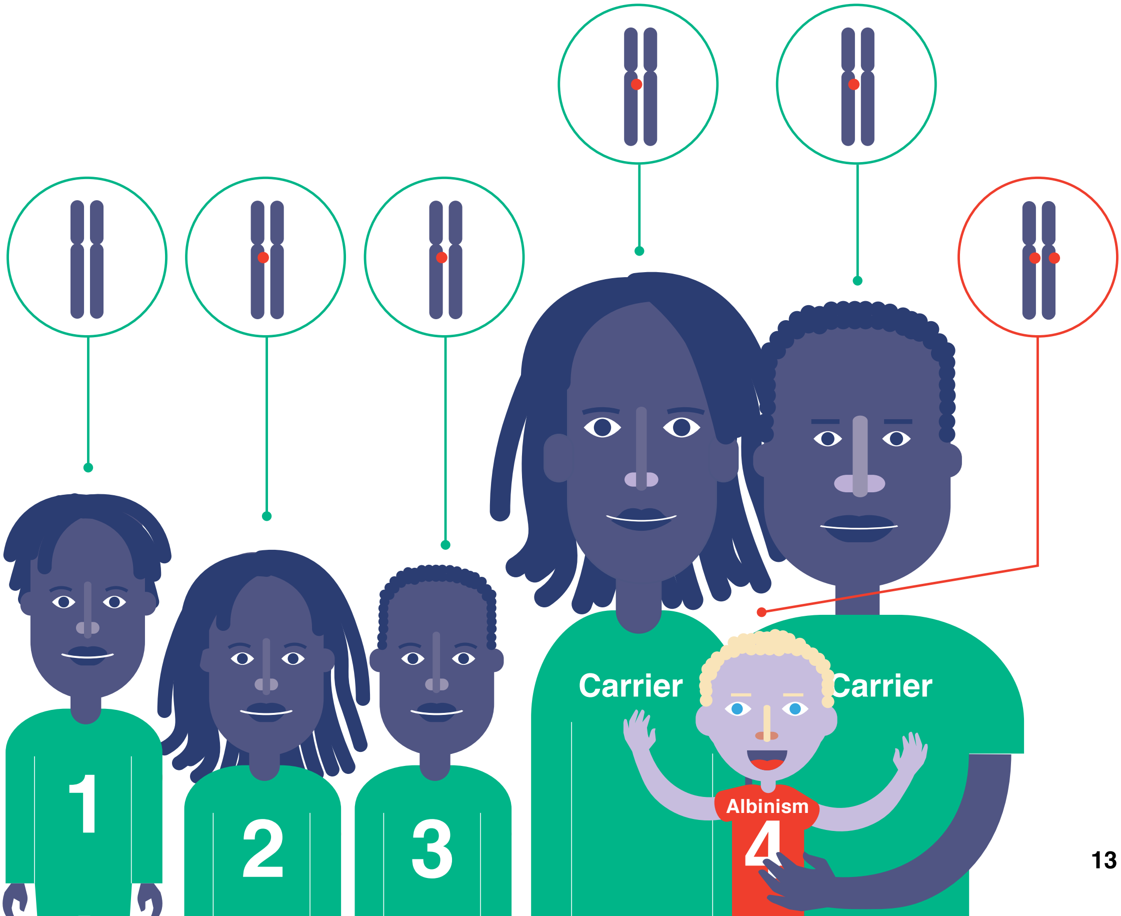
Traits, which are passed on by your parents, are called dominant if they already show in one of your parents. You have a 1 in 2 (50%) chance of getting a dominant trait or dominant disease from either your affected mom or dad.

4

But there are also recessive traits. For recessive traits, both parents carry a small change in a gene. Genes are the genetic information passed on to you from your parents. Genes give our bodies instructions on how to function. From the colour of your skin and eyes to how tall you are, the way you look – all of this is determined in your genes.



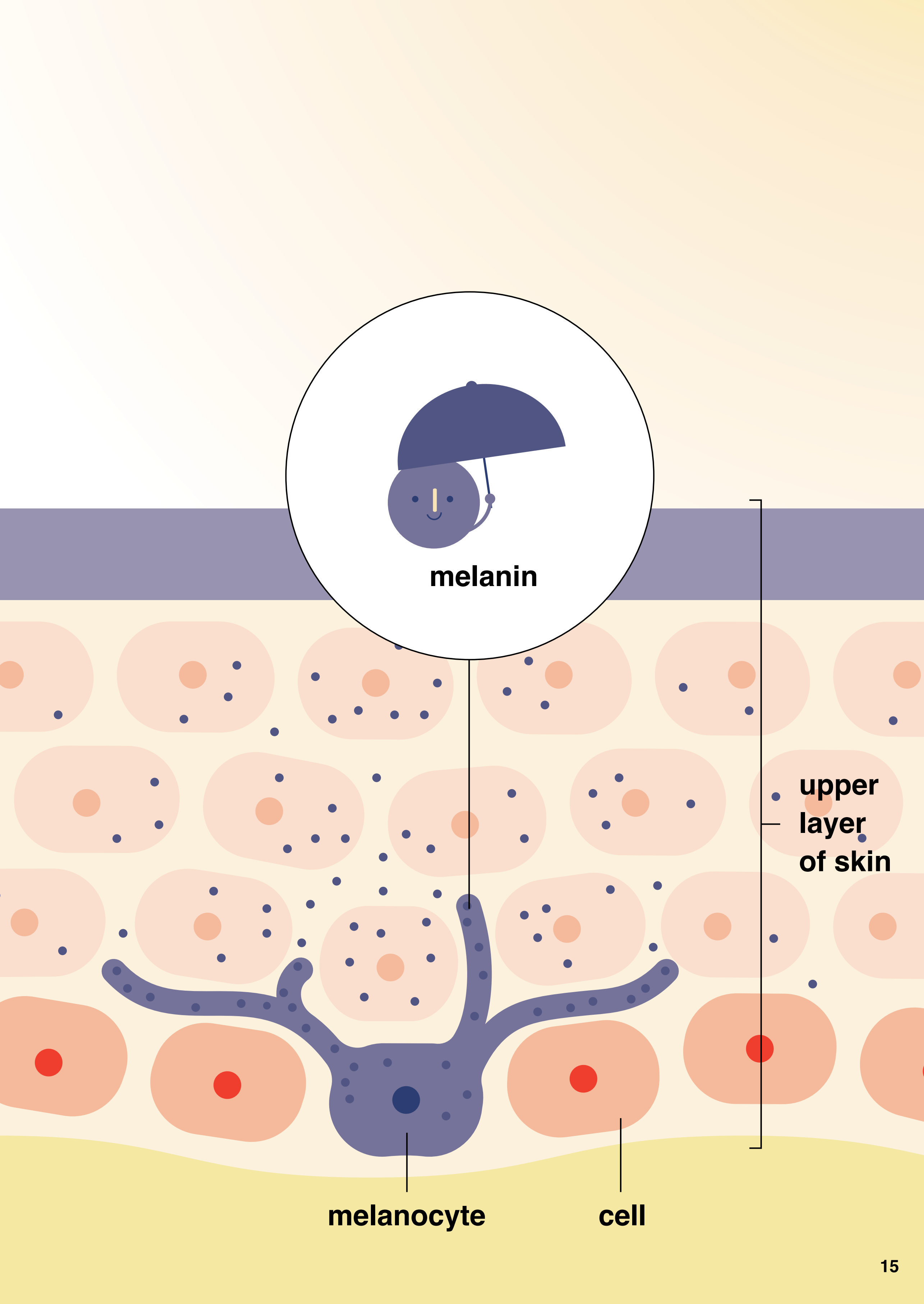
Recessive traits don't show; they are invisible. A parent who does not show but has the gene alteration is called a carrier. But when two carriers have a child together, there is a 1 in 4 (25%) chance in each pregnancy that this child will have this disease. One of these recessive disorders affects the eyes, the skin and the hair and is called albinism.



5

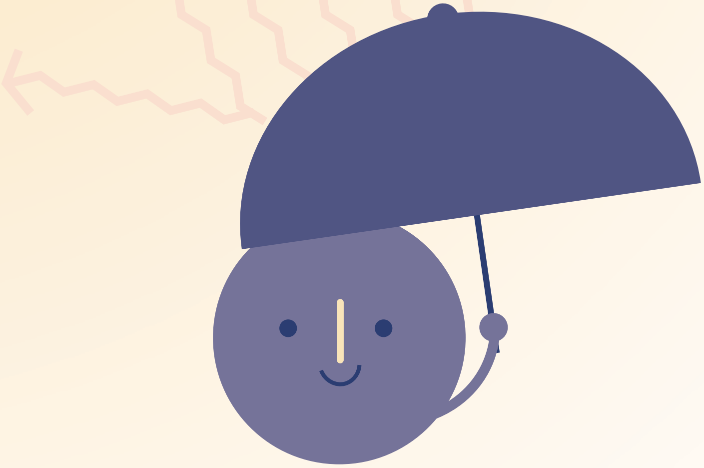
Your skin covers your entire body and helps you to maintain a consistent body temperature. It is a barrier between your body and the outside world.

Your skin has different layers and many different functions. In the upper layer, there are nerve cells, which help you to sense pressure and pain. Some cells help you to keep out germs. Other cells, called melanocytes, protect your skin from too much sun.

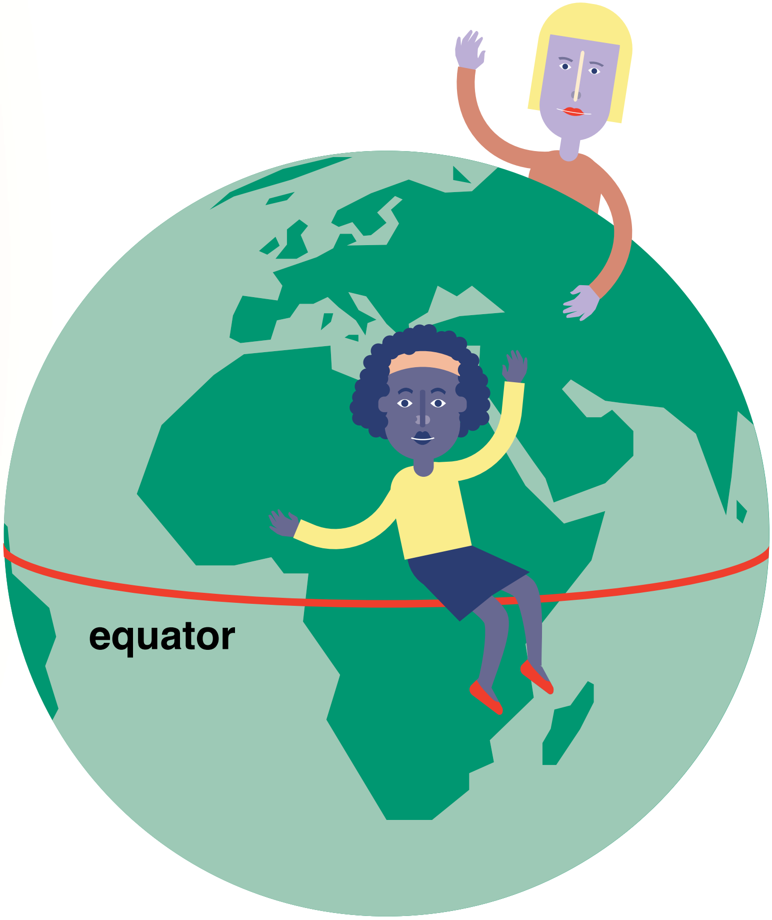

melanin
upper layer of skin
melanocyte
cell

6

Melanocytes store the dark pigment melanin in the outer protective layer of the skin. In the sunshine, your skin produces more melanin, which turns your skin darker. Darkening your skin is a good way to protect it from the dangerous UV rays of the sun. Some people with fair skin can easily get a sunburn – black people don't get one as easily.



melanin



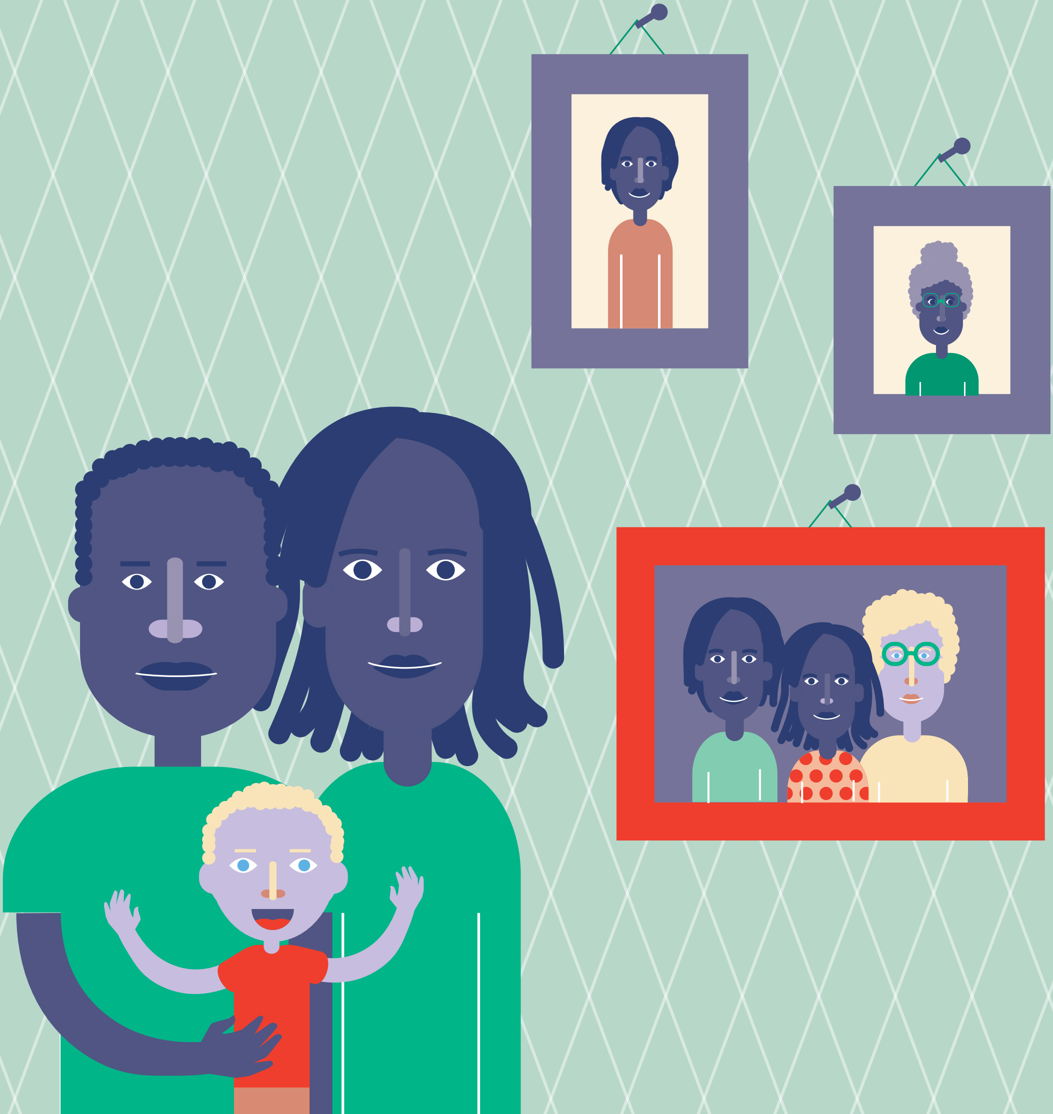
In some regions of the world, most people are white, for example in the countries in the north with less intensive sunshine. In other regions of the world nearer the equator, most people are black. Black people are better prepared to cope with the sun.

7

Sometimes a couple can have a child with very light skin and hair, as well as lighter coloured eyes. This can come as a surprise if it happens for the first time in both families. It is particularly striking if this happens to a black couple. Unexpected things attract a lot of attention. Quite likely, however, it may have happened in the families before. It is particularly frequent when relatives or even distant relatives

marry one another. Maybe an aunt or a great-grandfather already looked like this. In all cases both parents have contributed to this. Geneticists then say that this trait is recessive and runs in both families.

They have given it the name albinism, as alba means white.



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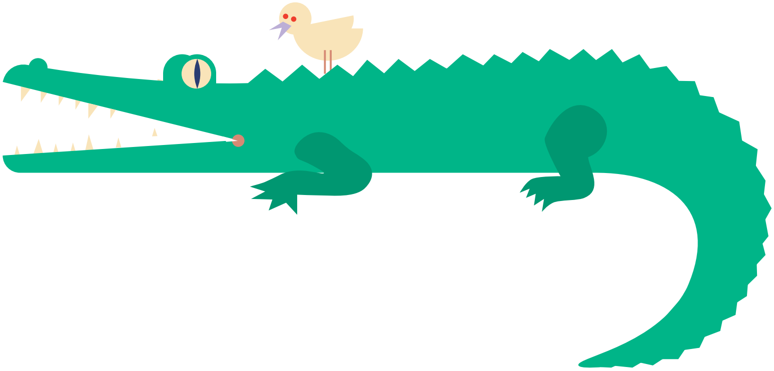
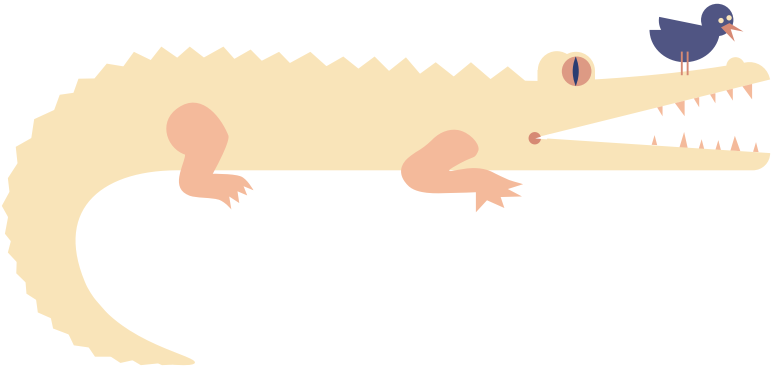
Animals can also have albinism. They are then considered especially attractive.

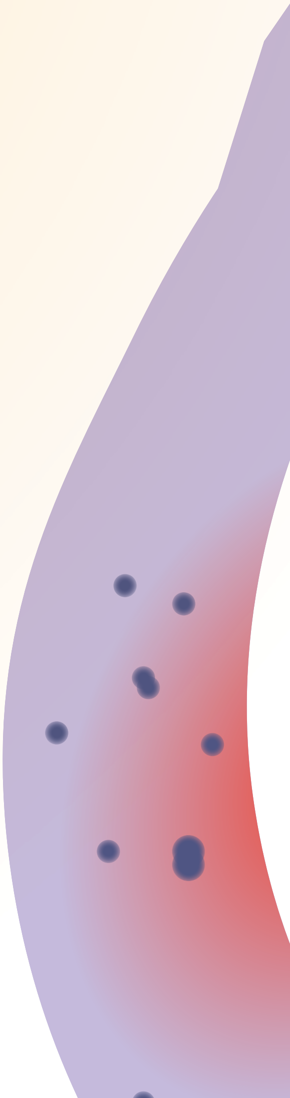


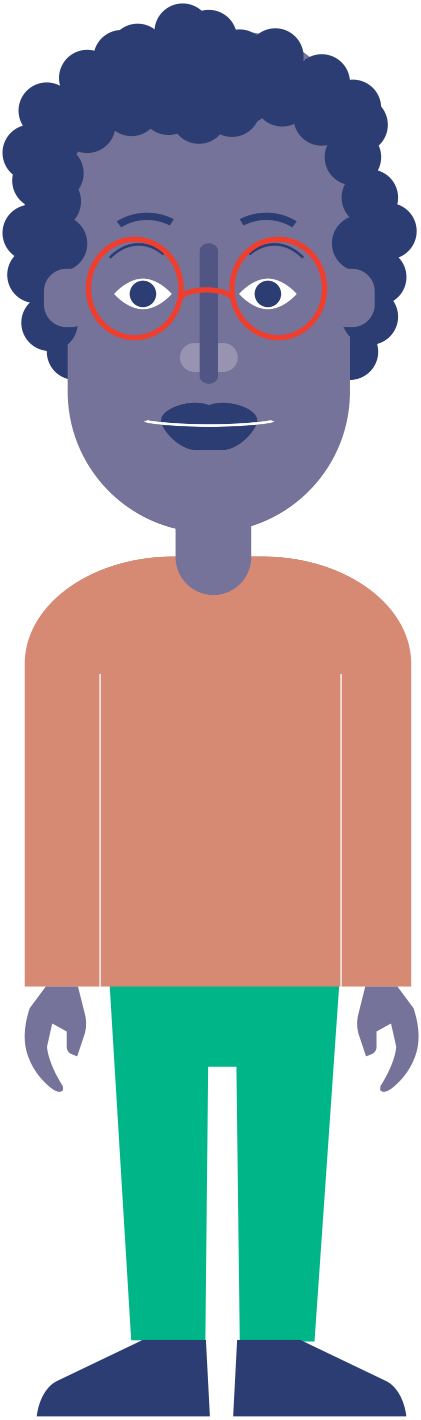


9

People with albinism have low vision. This is due to the misrouting of nerve fibers in their eyes. This makes it harder for them to read and catch a ball, but they are able to do a lot of the same work that you can. They also benefit from sunglasses to protect their eyes, and a hat and sun cream to protect their skin from UV rays of too much sun. Their skin does not have enough melanin to protect them from sunburn. That can lead to very badly damaged skin and eventually to skin cancer.

Otherwise, these people are just like you: they are clever and like music and sports.





20,000 genes

20 genes

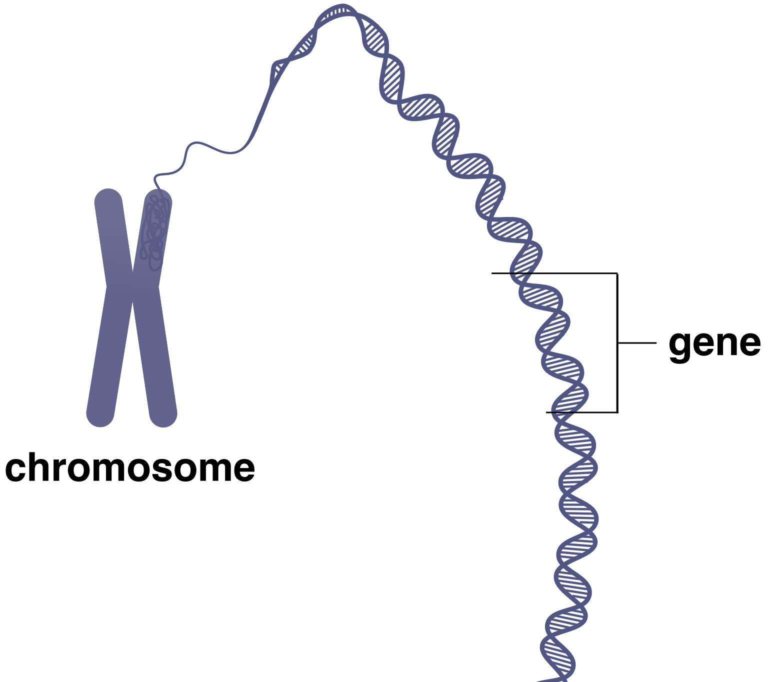
produce or
distribute melanin

10

All humans have roughly twenty thousand genes. In the case of albinism, a small change happens in one of 20 different known genes that produce or distribute melanin. One of these genes is called *OCA2* and its gene product is a protein in the membrane of the melanosomes. Changes in this gene are commonly found in people with albinism in Africa. But there are people with albinism all over the world.

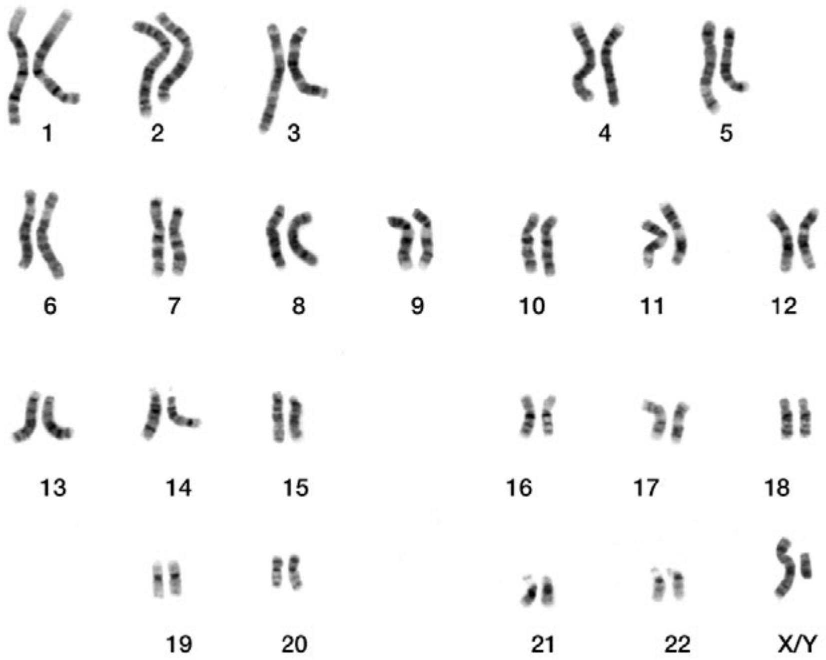
Another gene is called *OCA1* and produces an enzyme called tyrosinase. Most people with albinism in the north have a change in this gene. Normally this gene is used to produce enough melanin.

But in people with albinism, these genes no longer function properly.

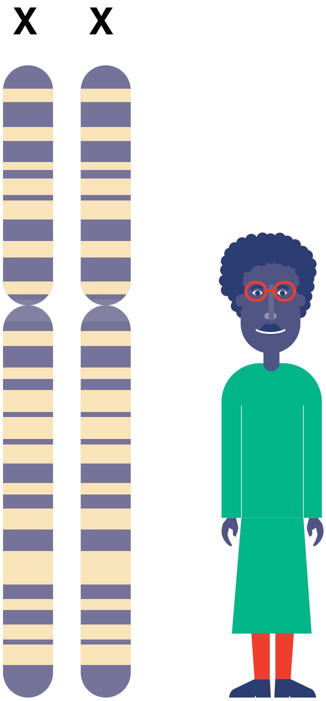


11

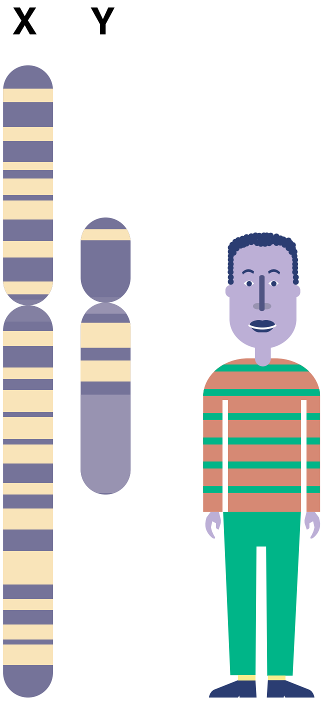
Genes reside on chromosomes. There are 23 chromosome pairs in every cell. Twenty-two chromosome pairs look pretty identical in all humans. But there is one exception: boys carry a Y chromosome in every cell which does not have a perfect match to another chromosome. Boys carry one X chromosome and a Y chromosome, while girls carry two X chromosomes. X chromosomes are much longer than Y chromosomes and carry many more genes.



X and Y chromosomes are also called sex chromosomes. The other chromosomes are called autosomes. If there is a defect on one of the X chromosomes in girls, it does not show as the second X chromosome can compensate for it. Girls have a clear advantage here.



females



males

12

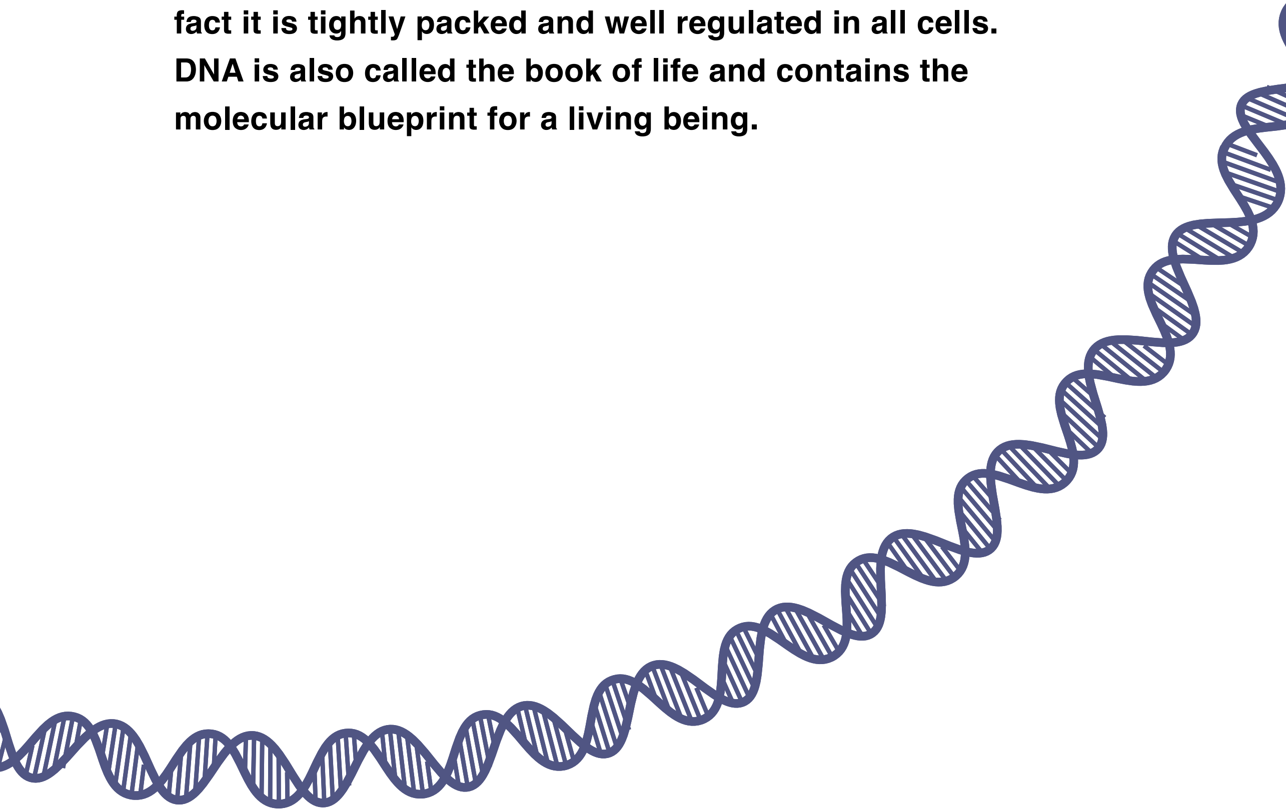
Genes that produce or distribute melanin reside on different chromosomes. One type of Ocular albinism, affecting only the eyes, is caused by a small change in a gene residing on the X chromosome. When there is a change in this gene, it affects only males, as females with two X chromosomes can compensate for the loss. Geneticists call this X chromosome-linked recessive inheritance.

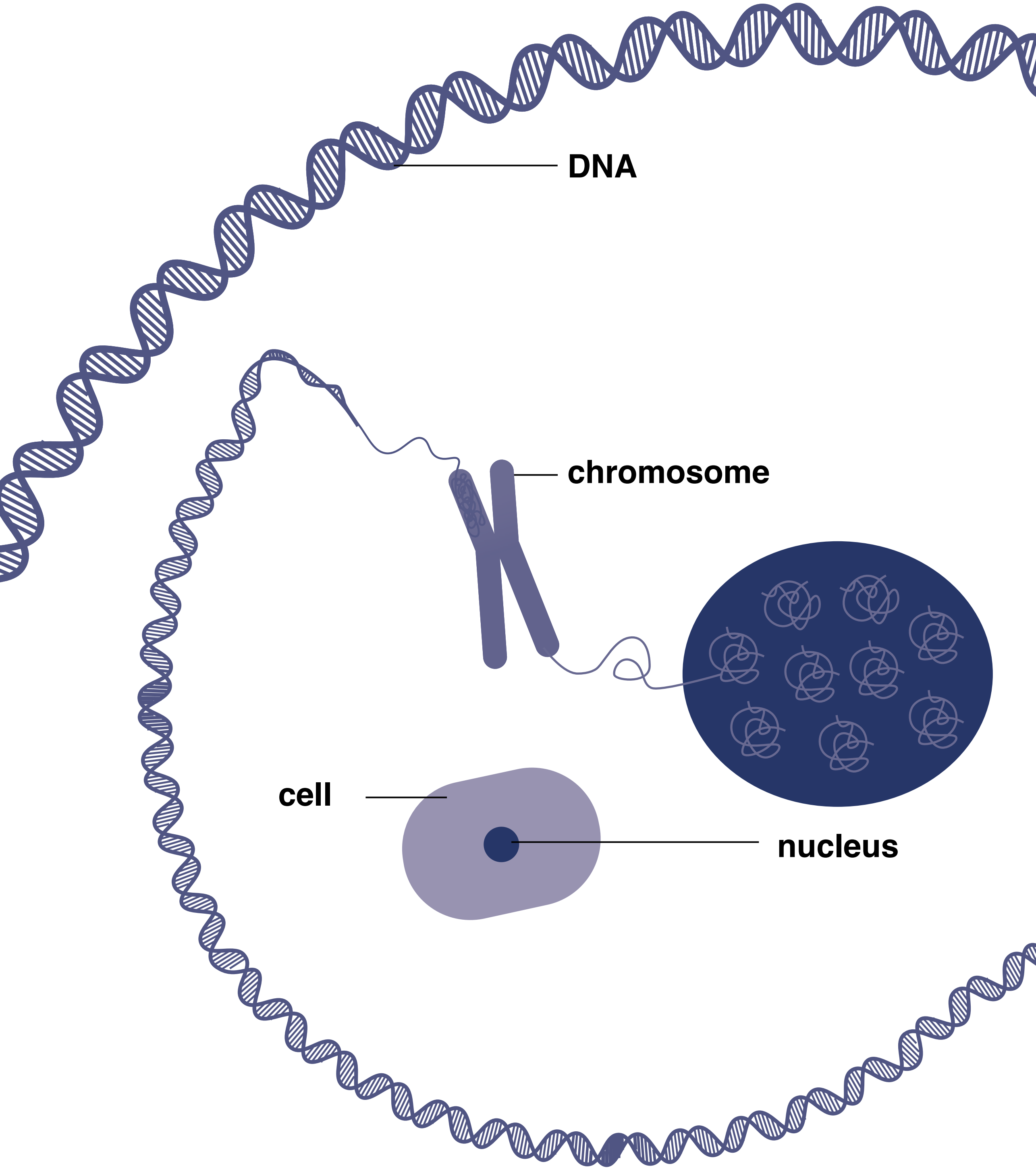
Different types of albinism are therefore passed on in different ways. All other gene variants that provide too little or no melanin reside on other chromosomes, not on the X chromosome. The most common form of albinism in Africa, caused by a change in the

OCA2 gene, for example, resides on chromosome 15. The most common form of albinism in the north affecting the skin, hair and the eyes, is caused by a small change in the *OCA1* tyrosinase gene residing on chromosome 11. If the gene defect resides on autosomes, the situation is different; girls and boys have the same probability of getting albinism. Geneticists call this autosomal recessive inheritance.

13

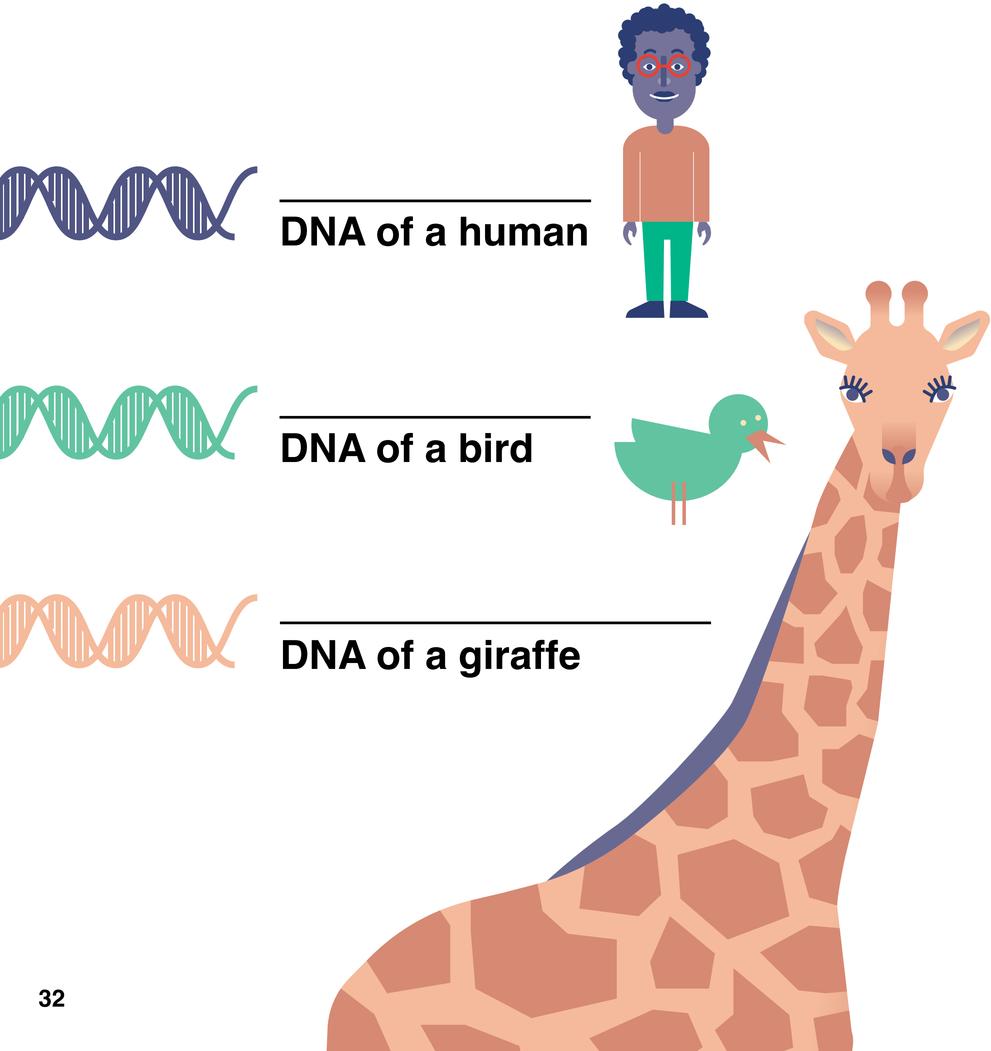
We talked already about cells and the 23 pairs of chromosomes that are found within each cell of your body and about the 20,000 genes that reside on these chromosomes. Now DNA comes into play. DNA stands for deoxyribonucleic acid, a long and complicated word, so it was decided to just use the initials. DNA is a large molecule in the shape of a double helix, which looks a little bit like a ladder that is twisted many times. It is an extremely thin and long thread of about one meter when stretched out, but in fact it is tightly packed and well regulated in all cells. DNA is also called the book of life and contains the molecular blueprint for a living being.

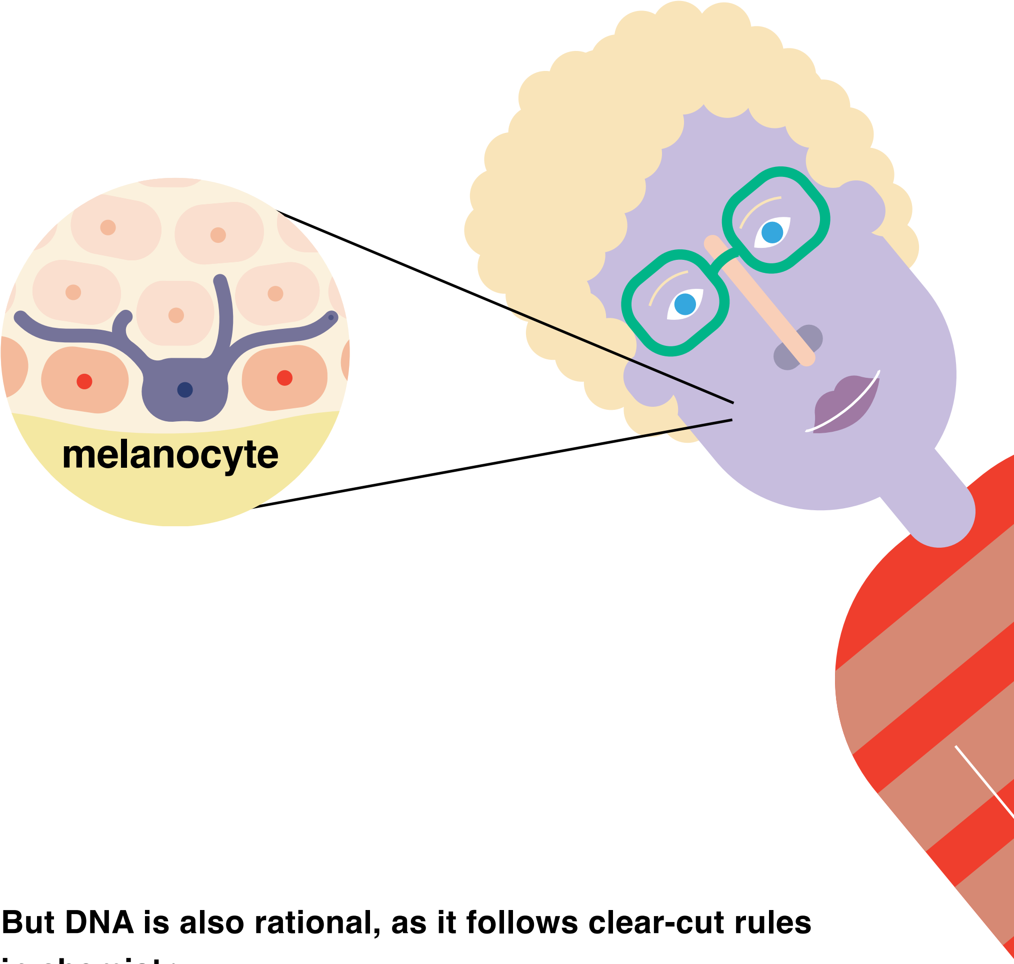



DNA
chromosome
cell
nucleus

14

DNA is truly amazing. It is amazing because it carries all the genetic information and thus the secret of life for a living being. If you are a human being, it carries the information for a human being. If you are a giraffe, it carries all the information to make a giraffe. If you are a bird, it carries all the information to make a bird.





But DNA is also rational, as it follows clear-cut rules in chemistry.

DNA is contained in all your cells. But if you have a gene defect in the tyrosinase gene or in one of the genes of this functional pathway, it only shows in the retina cells of the eye and the melanocyte cells in people with albinism.

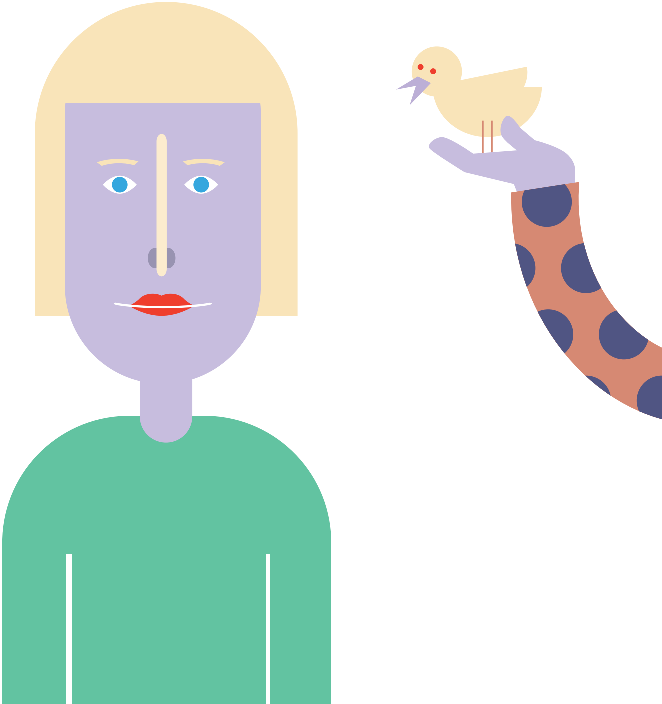
15

You also need to know that people do have traits that are not necessarily genetically inherited. If you exercise a lot, for instance, your muscles will grow and get stronger because of your own efforts. If you like to read, the nerve cells in your brain will connect faster, and maybe you will not only be able to read faster, but also become smarter. Therefore, you are not a "slave to your genes", because there are still a lot of things that you can do and decide for yourself.

16

People with albinism live in every country on Earth. In Europe, you will find one person with albinism among approximately 10,000 people. Albinism is therefore a rare condition. However, in other countries – mainly in those countries where it is more likely that people with a common ancestor may marry each other, this number is a lot higher. In some African countries you will find one person among 1,000 to 2,000 people.

If you add together all people with albinism, you will come up with a number of more than a million people living on our planet.

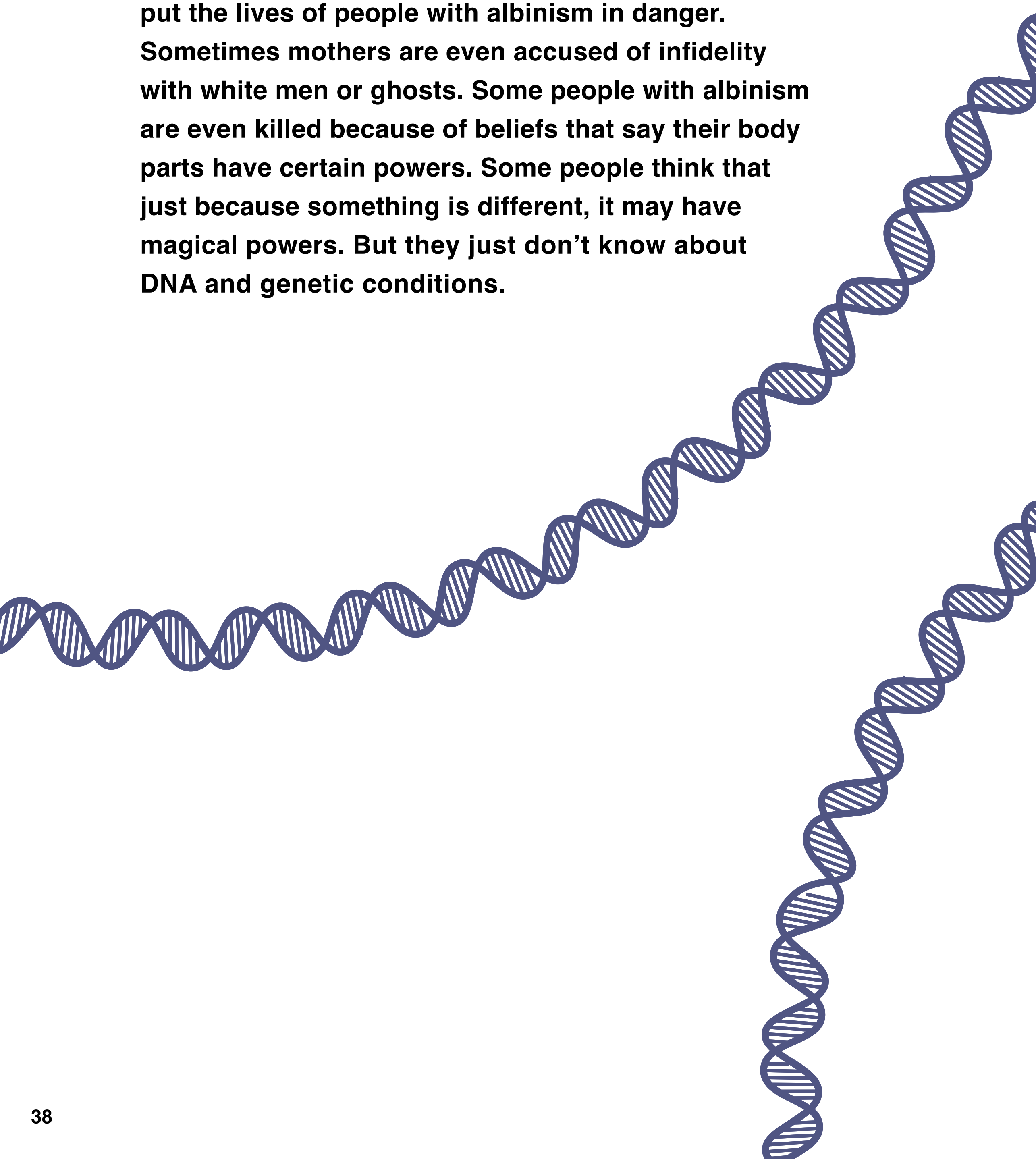


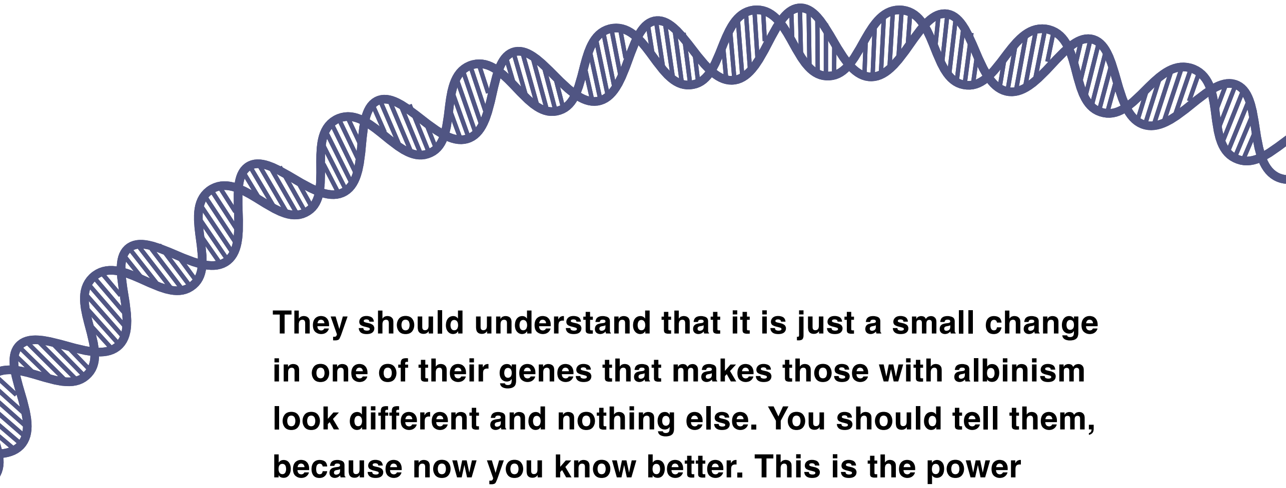
Today people can connect with each other through the internet or social media and support one another in self-help groups.

This will show them that they are not alone with their special needs and they can exchange advice.

17

In some areas of Africa, folklore and superstition put the lives of people with albinism in danger. Sometimes mothers are even accused of infidelity with white men or ghosts. Some people with albinism are even killed because of beliefs that say their body parts have certain powers. Some people think that just because something is different, it may have magical powers. But they just don’t know about DNA and genetic conditions.





They should understand that it is just a small change in one of their genes that makes those with albinism look different and nothing else. You should tell them, because now you know better. This is the power of knowledge.

Epilog

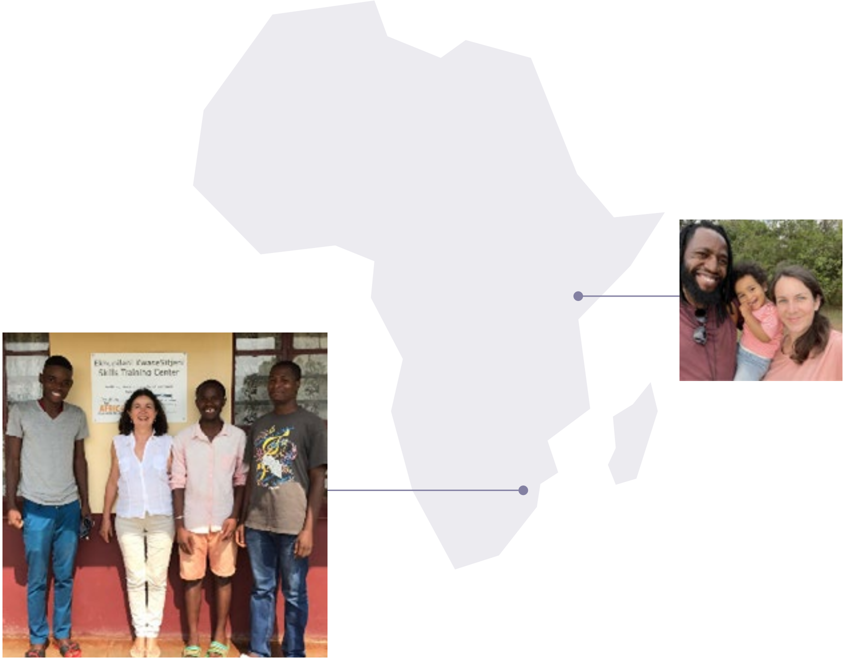
This is an international friendship project. It was triggered while travelling with my choir “Imbongi – Voices for Africa” in Eswatini (Swaziland) and talking to students at the skills training center in eSitjeni. There, I heard that newborns with albinism have “an association to the water” – meaning they get frequently drowned. In other regions of Africa, people with albinism get hunted, mutilated, or killed. These people are often children. The epicentre of violence against people with albinism is in Tanzania and Kenya, where the belief in spirits and witchcraft is deeply rooted. Witchdoctors claim that the body parts of people with albinism have magic powers and sell these, disassembled into small parts, as lucky amulets. Many hundreds of people have already been killed and countless people have been attacked and had limbs cut off.

Science contra superstition. I am convinced that scientific education has to begin at school. Albinism is a recessive pigment disorder, a genetic disorder, and genetics follows clear-cut rules. There is no superstition involved. All this provided the impetus to set up this small booklet for kids and young people.

I wrote a first draft of the text while staying for a week at the Institute of Wendy Bickmore in Edinburgh, Scotland. Rolf Sprengel in Heidelberg connected me with authors Jill und Giulia Enders, who, in turn, established the contact with Viola Kup, a communication designer living in Nairobi, Kenya. All the wonderful illustrations in this booklet were done by Viola. She was excited about the project from the very beginning and took time off for it, despite having just given birth to a daughter and being a working mother at the UN. Without her, the text would lack liveliness. Viola’s friend Sam Achola, a programmer, offered to set up an interactive website. This will allow us to also distribute the text via the Internet. We also thank Dawn Lee in Singapore, Susanne Loeben in Berlin, Benoit Arveiler in Bordeaux and Patricia Lund in Coventry for helpful comments.

All the translators who support this project with passion are thanked in advance with great gratitude. I was only the snowball that set the avalanche in motion.

Gudrun A. Rappold
Institute of Human Genetics
University of Heidelberg
December 2022



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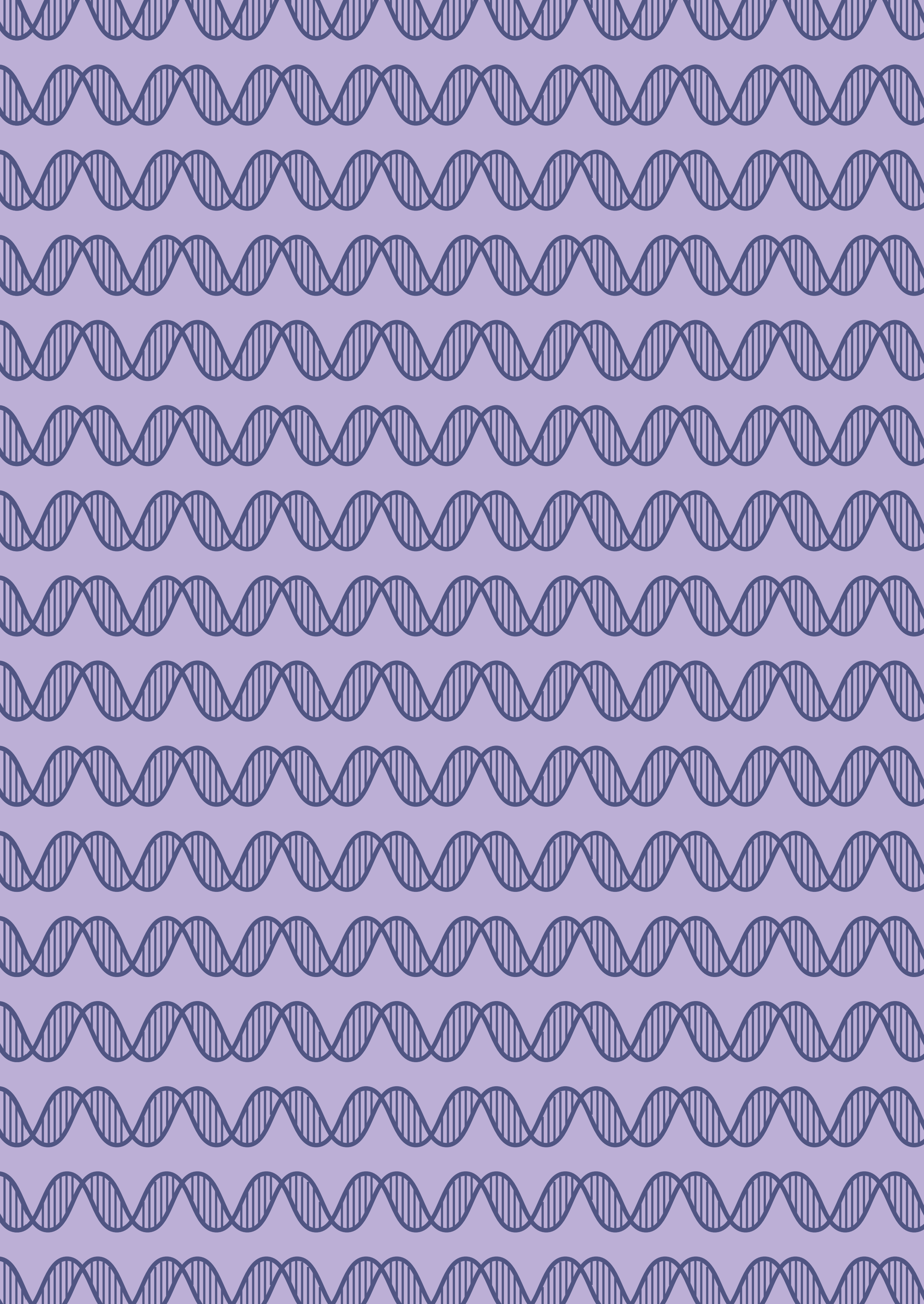

Why do we look the way we look? Why are some people black and others white? “Albinism – just a gene” highlights the genetic principles of pigmentation in people, including those with albinism, a recessive pigment condition resulting in very light skin, as well as lighter coloured eyes. The text clears up with beliefs in Africa and puts forward rational scientific explanations. School children and young adults can learn the basic principles of inheritance, genes, and DNA thanks to the simple language and clear illustrations that enliven the text and engage the reader.

albinism-justagene.com