21 QUESTIONS ABOUT TRISOMY 21

RT21

Regroupement
pour la Trisomie 21

SUMMARY

INTRODUCTION

As a society, it is possible to change our perceptions about people with trisomy 21, open ourselves up to their differences and help them take their place within our community. You may be surprised to see just how much they have to offer and how much we can learn from them!

With the help of numerous contributors, the team at RT21 (Regroupement pour la Trisomie 21) seeks to answer 21 of the most frequently asked questions about trisomy 21 and the people who live with the condition. Keep in mind that each person living with trisomy 21 is unique – developing according to his or her own capabilities, preferences and interests – and therefore the answers here are meant to be of a general nature.

RT21 is a Quebec non-profit organization whose mission is to foster the full development of people with trisomy 21, promote their contribution to society and defend their rights. RT21 also help the families, caregivers and professionals who support them.

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In 2014, Fabien Toulmé, known for his beautiful graphic novels, published "Ce n'est pas toi que j'attendais", in which he describes his first meeting with his daughter Julia, born with trisomy 21. He has contributed all the artwork included in this pamphlet, and we are grateful for his generosity.

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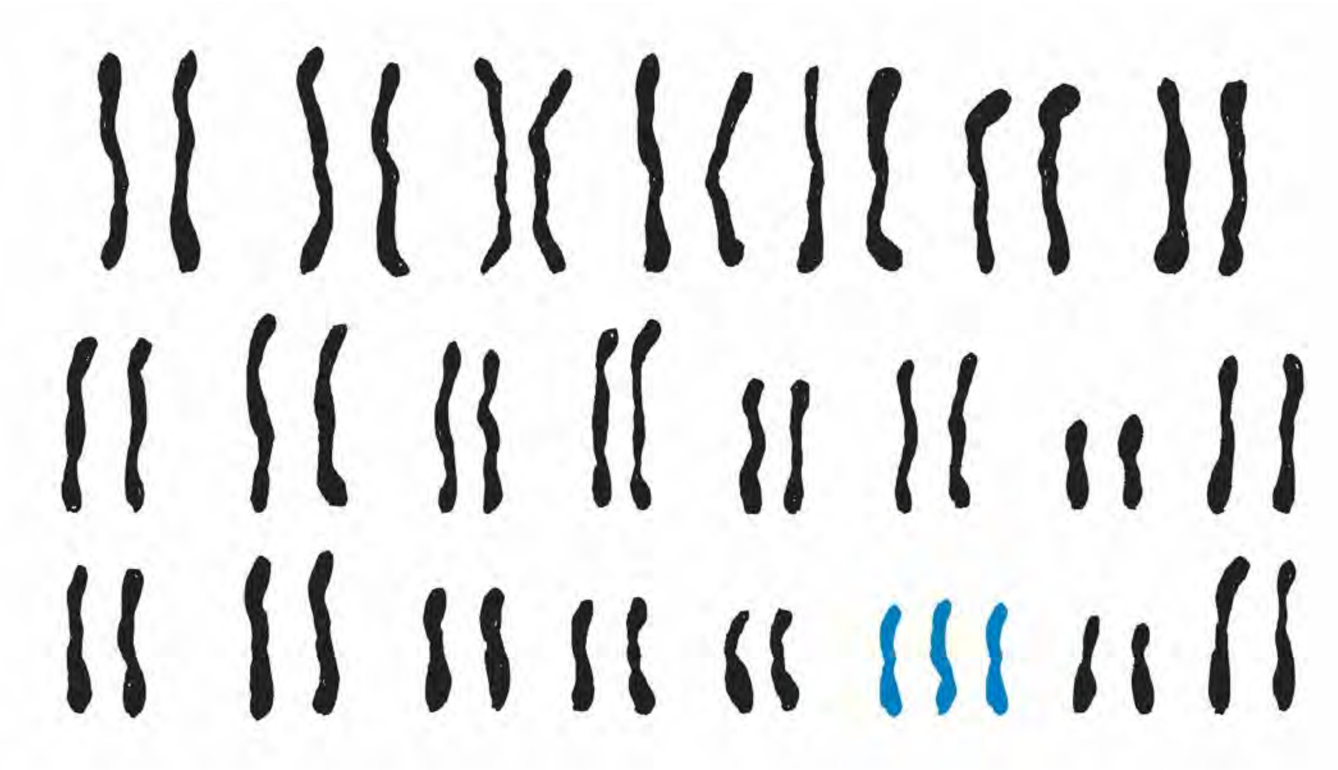
1. WHAT IS TRISOMY 21?

Trisomy 21, also known as Down syndrome, is a genetic disorder caused by the presence of a third chromosome on the 21st pair of chromosomes. It is not a disease, you can't catch it, and it can't be cured.

People with trisomy 21 exhibit cognitive delays and distinctive physical features that vary from one individual to the next. The condition is present in approximately one out of every 770 births, and its incidence increases as maternal age increases. Generally, trisomy 21 is not hereditary but rather occurs randomly.



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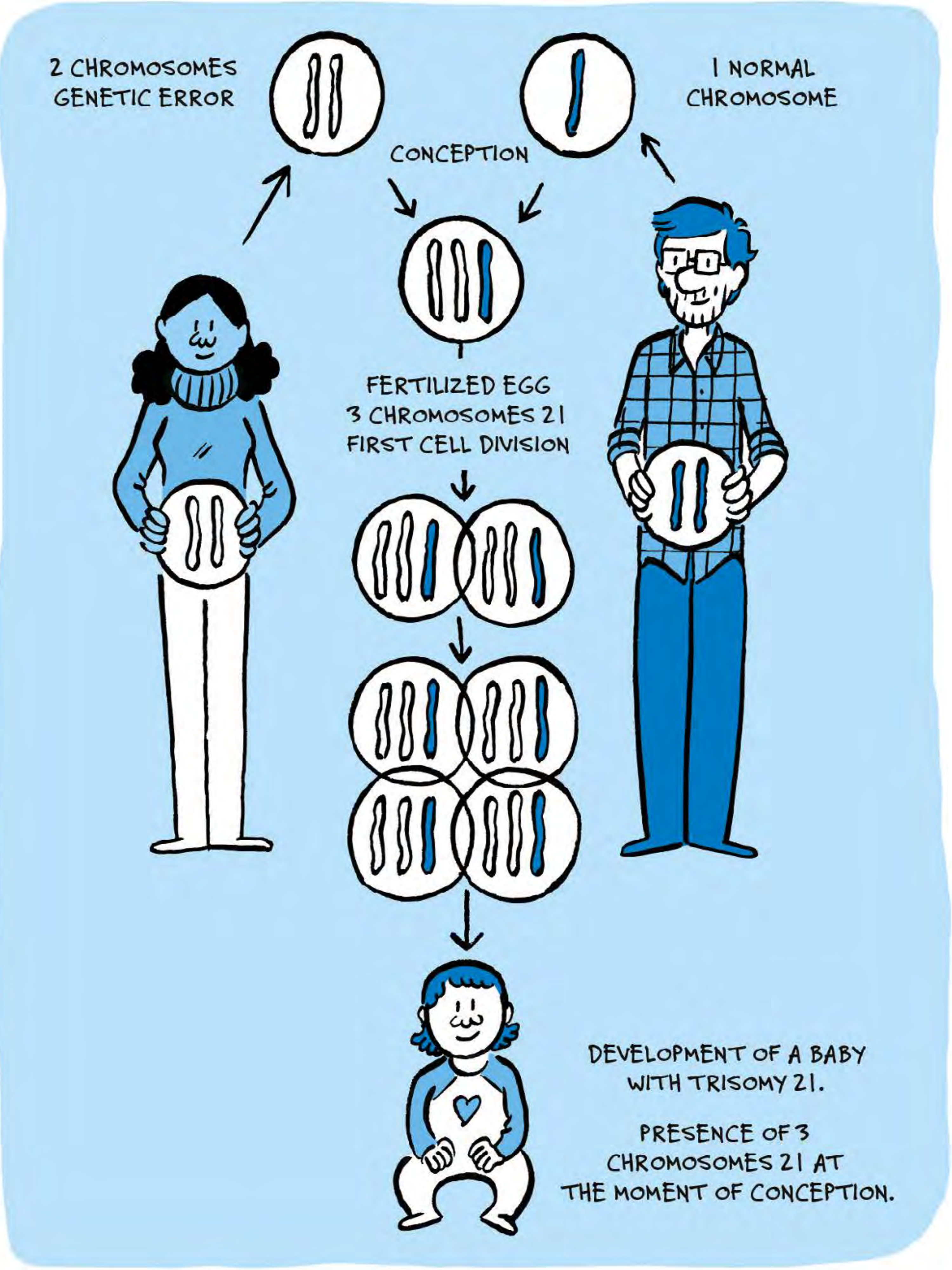
TRISOMY 21*



* Representation of a standard trisomy 21

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2. IS THERE MORE THAN ONE TYPE OF TRISOMY 21?

Three types have been identified so far.

1. Approximately 95% of people with trisomy 21 have **standard trisomy 21**, which is where every cell in the body is affected. It is the result of an error in the distribution of chromosomes (nondisjunction) that takes place during the first cell division *(as illustrated on page 4)*.
2. Approximately 2% have **mosaic trisomy 21** (mosaicism), which is where the error in the distribution of chromosomes happens at the second cell division. This results in cells with 46 chromosomes and cells with 47 chromosomes. People with mosaicism may sometimes have a milder intellectual disability and less pronounced physical features compared to people with standard trisomy 21.
3. Approximately 3% have **translocation trisomy 21**, which is where a part of chromosome 21 breaks off and attaches itself to another chromosome. One of the parents, him or herself a carrier without being affected, transmits the translocated chromosome to the child. This is the only type of trisomy 21 that can be inherited.

3. ARE THERE OTHER TYPES OF TRISOMY?

Humans have 23 pairs of chromosomes. A trisomy, the presence of a third chromosome, can happen on any pair, from the first to the 23rd. Trisomy 21 is the best known of all. Depending on the affected chromosome and the type of trisomy, the fetus may or may not be viable. In humans, most cases of trisomy lead to a miscarriage, but several allow for live births, including trisomies 8, 13, 18, and 21.

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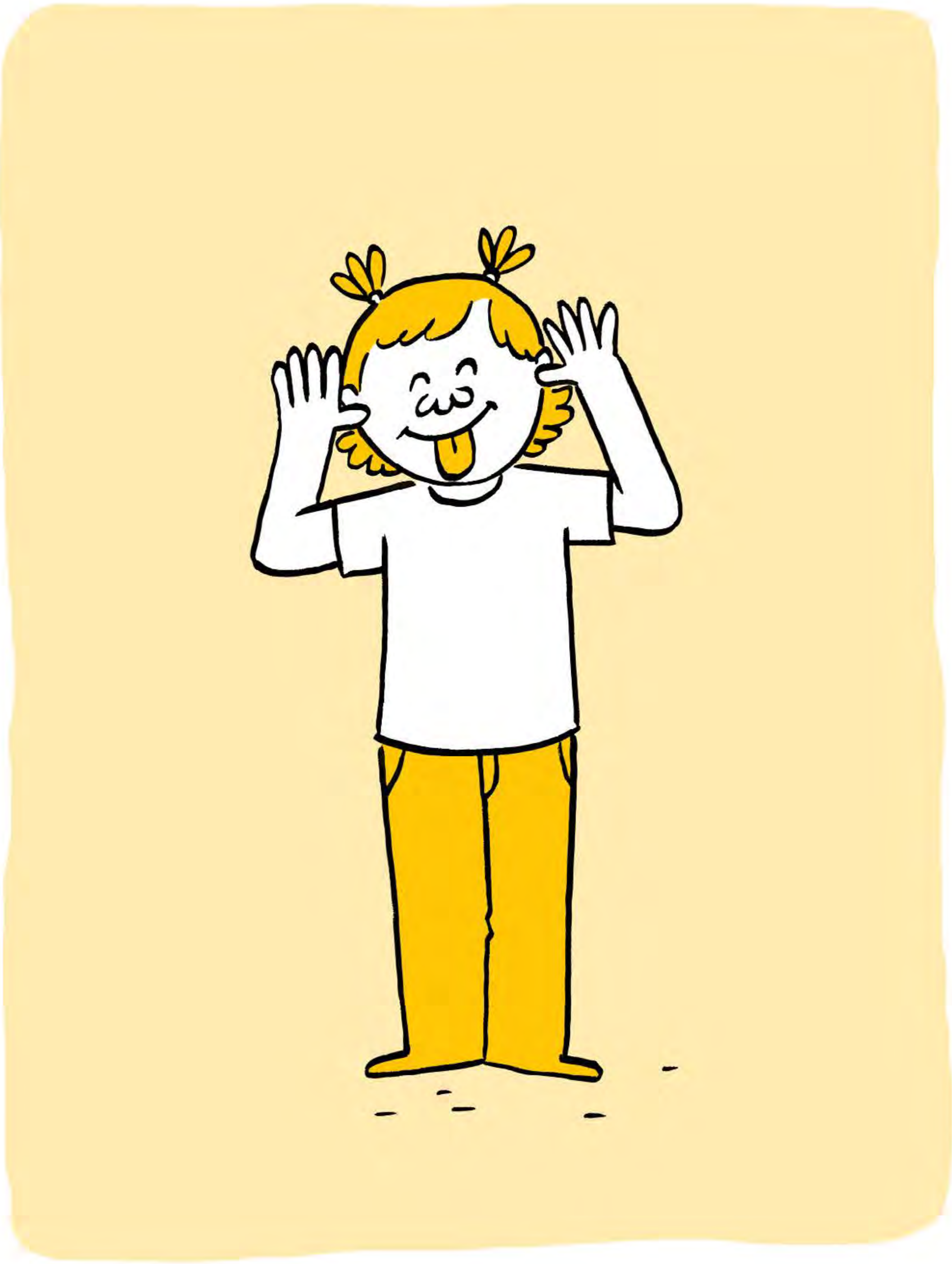

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4. WHAT ARE ITS PHYSICAL CHARACTERISTICS?

People with trisomy 21 often have slanted eyes, a round face, a small head, low-set ears, a short nose with a flattened base, a small mouth, and very thin lips. Their arms and legs are fairly short. They have a single crease across the palms of their hands and generally low muscle tone.

These characteristics are more or less pronounced depending on the individual. And just like all of us, the features of people with trisomy 21 are also shaped by heredity, which gives each individual their personal, distinguishable look.

5. WHY DO PEOPLE WITH TRISOMY 21 OFTEN STICK OUT THEIR TONGUE?

People with trisomy 21 generally have a smaller than average mouth, palate, and jaw. Their tongue has less room to move and so it often sticks out. The muscles of the mouth are also weaker, due to the overall low muscle tone. All this makes it harder for them to keep their mouth closed.

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DO ALL PEOPLE WITH TRISOMY 21 HAVE AN INTELLECTUAL DISABILITY?

The effects of trisomy 21 on development vary from one person to another and depend on several factors. The level of intellectual disability generally ranges from mild to moderate, though it can sometimes be severe. A psychological evaluation around the age of seven will determine the level of intellectual disability. Most people with trisomy 21 are able to integrate into society as autonomous adults.

7.

WHAT IS THE MENTAL AGE OF PEOPLE WITH TRISOMY 21?

The notion of mental age is not relevant since the range of experience and skills is variable. Depending on their interest, people with trisomy 21 can develop considerable skills in a given field, regardless of their age. We must also take into consideration the experience they gain over the years. A 30-year-old with the skills of an 8-year-old still has 22 years more life experience, which is certainly not negligible!

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8. CAN THEY GO TO DAYCARE AND SCHOOL LIKE OTHER CHILDREN?

Like their peers, children with trisomy 21 have a right to fully access services such as daycare and school. They are, whenever possible, included in regular classrooms or receive schooling in whatever environment is best for them. All children are part of the community and inclusive education should be preferred.

If inclusion in a regular classroom is not possible, they may attend specialized classes or schools. These environments support children throughout their schooling by providing smaller class sizes and specialized services.

9. CAN THEY READ, WRITE, AND COUNT?

Yes, people with trisomy 21 can learn to read, write, and count. Success is possible provided their capabilities, limits, needs, and interests are taken into account. To reach their objective, they are generally supported by a team that believes in them and spares no effort. This includes school staff, other professionals, and above all, the parents, who are the key to their child's development.

People with trisomy 21 acquire core skills in stages. While one core competency is being acquired, other skills are generally acquired more slowly.

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10. CAN PEOPLE WITH TRISOMY 21 LEARN MORE THAN ONE LANGUAGE?

Some studies have shown that being exposed to two languages has no negative effect on language development in children with trisomy 21. They can therefore learn two languages as long as they are sufficiently exposed to them. Known language difficulties will manifest themselves in both languages but will not be exacerbated by bilingualism.

11. CAN THEY FIND WORK AND LIVE AUTONOMOUSLY?

With the help of professionals, as well as friends and family, a large percentage of people with trisomy 21 manage to become autonomous, complete adequate schooling, integrate into the workforce, and actively participate in society. Of course this all depends on the level of intellectual disability and the preferences and interests of each individual!

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12. CAN THEY HAVE CHILDREN, AND IF SO, WILL THESE CHILDREN HAVE TRISOMY 21?

It is possible for people with trisomy 21 to have children. Many studies show that the men are, for the most part, sterile, but not the women. As to whether their children will also have trisomy 21, the probability is 50%.

13. CAN PEOPLE WITH TRISOMY 21 HAVE A LOVE LIFE AND BE SEXUALLY ACTIVE?

Yes. We sometimes think that because they have an intellectual disability they will also have an emotional disability, which is not the case. Sexual expression is linked to physiological, physical, psychological and intellectual development. Some people with trisomy 21 may only develop up to a basic relational stage, expressing little interest in relationships beyond those with family and friends. Based on individual needs and level of interest, they should be informed about different aspects of a healthy affective and sexual life, including understanding and expressing emotions, interpersonal and sexual relations, personal affirmation and self-esteem, as well as physical changes during puberty.

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14. DO THEY HAVE THE SAME RIGHTS?

People with trisomy 21 have the same rights as all citizens, rights that are guaranteed by the Canadian Charter of Rights and Freedoms. For example, educational inclusion is based on the right of every child to attend daycare and school without discrimination due to a handicap. Children with trisomy 21 can therefore learn at their pace and according to their abilities. The school board or school officials must consider all possible accommodation measures to compensate for known disabilities. This includes not discriminating on the basis of personal characteristics such as the intellectual, physical, sensory or psychological disabilities due to trisomy 21.

15. ARE THEY AWARE THAT THEY HAVE TRISOMY 21?

A large proportion of people with trisomy 21 eventually become aware of their condition and cope relatively well with this knowledge. They come to understand the challenges they face, in contrast to those of siblings and other peers. Some individuals can be negatively impacted and develop a mood disorder. Those with a mild intellectual disability often struggle the most, being more fully aware of their limits and of rejection on the part of society.

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16. WHAT IS THE LIFE EXPECTANCY OF PEOPLE WITH TRISOMY 21?

Life expectancy is influenced by the different medical conditions associated with trisomy 21. However, medical advances have increased the average lifespan so that today, people with trisomy 21 live an average of 60 years. It is expected that the average will fall in line with that of the general population in the foreseeable future.

17. WHAT MEDICAL CONDITIONS ARE MOST OFTEN ASSOCIATED WITH TRISOMY 21?

People with trisomy 21 often present malformations such as congenital heart defects and musculoskeletal abnormalities, such as loss of normal bone alignment at a joint (luxation), ligaments that are too loose (ligament laxity), low muscle tone (hypotonia), and joint instability. They are also more prone to develop Alzheimer’s and thyroid disease. However, it is important to note that most of them lead a healthy life.

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18. WHY IS IT DIFFICULT TO UNDERSTAND PEOPLE WITH TRISOMY 21 WHEN THEY TALK?

To correctly articulate sounds, you must first be able to hear them properly. Some people with trisomy 21 have hearing problems, which leads to difficulty producing sounds correctly. They also have low muscle tone and coordinating the many movements necessary for speech – movements of the lips and tongue, breathing, and vibration of the vocal cords – becomes a challenge. Additionally, it is often difficult for them to structure sentences correctly and use appropriate vocabulary. It is therefore not unusual for people with trisomy 21 to express themselves less clearly and to use simple language. However, their social skills allow them to be well understood.

19. DO PEOPLE WITH TRISOMY 21 HAVE A SOCIAL LIFE?

Yes, just like everyone else! A person's social life is encouraged and strengthened by family, friends, colleagues, and the bonds that are created through shared activities and hobbies. As with most people, their social circle is important. People with trisomy 21 continue to develop good social skills and appropriate social behaviour throughout their lives.

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20. HOW SHOULD I ACT AROUND PEOPLE WITH TRISOMY 21?

You should treat them like any other person of their age. It's important not to pretend like they don't exist by only talking to the people that are accompanying them. Use simple language, without resorting to baby talk. Just remember to adapt your vocabulary to their level of comprehension, establish eye contact, make short sentences, give clear instructions, use gestures to provide examples, and use visual cues.

21. HOW CAN WE HELP PEOPLE WITH TRISOMY 21 REACH THEIR FULL POTENTIAL?

Experience has shown that early stimulation helps develop motor skills, intellect, and language. It is therefore essential to intervene from an early age, remembering that stimulating the development of a child with trisomy 21 is not much different from stimulating any other child.

In addition to parental involvement, guidance from specialists can support development in children with trisomy 21 so that they can grow and achieve their full potential. It is important to believe in their capabilities and avoid placing limits on them. In time, they will discover their own limits and develop their strengths.


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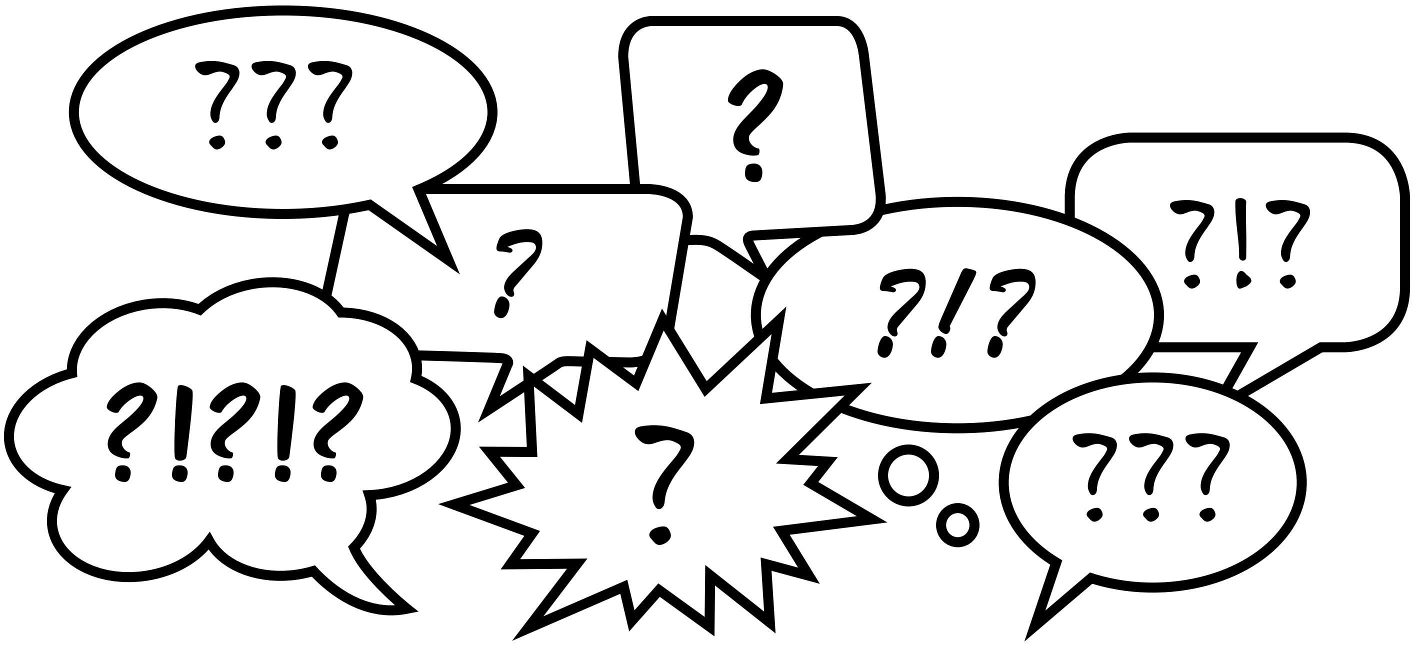

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