



Jake, 'Teddy Blue', Raffi the
Bumble Bee and the Mystical Yellow Flowers



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More information about neurotransmitters disorders and amino acid decarboxylase deficiency:



<http://www.aadcresearch.org>



<http://www.guiametabolica.org>



Jake lived in a tepee. He wore feathers on his head, a bow with arrows on his back and a small blanket in his red bag. Jake was a normal Indian child, but something made him special, super-special.

Jake had a friend who was a blue-coloured bear everyone knew as ‘Teddy Blue’; the townspeople said that he had blue sparkly fur because he was born on the night of a shooting star and that star was his Mom.



Jake and ‘Teddy Blue’ also had another friend called Raffi, he was a loud and very funny bumble bee who liked to sit on ‘Teddy Blue’ nose. Raffi went everywhere with Jake and ‘Teddy Blue’ and they were like brothers.

But, there were many things that made ‘Teddy Blue’ very special too; he was different from the other bears. He never growled or got angry with Jake and Raffi. He was always kind, gentle and loving. They all went on lots of adventures together like fishing in the river and running in the meadow, although most of the time Jake would give ‘Teddy Blue’ piggybacks, but he didn’t mind.



Teddy Blue' was not as big and strong like the other bears and he needed a little more help with things and would get tired easily. Jake and Raffi always took care of him together. 'Teddy Blue' was sooooo grateful.

When it was cold 'Teddy Blues' favourite place was curled up at Jake's feet in the tepee with Raffi sitting on his nose quietly singing a soothing song.

Raffi could be quiet sometimes, HONESTLY! But usually, he would be buzzing around, singing loudly and making sure everyone knew he was there. Raffi was the one to make everyone laugh, quite the comedian in fact!



Every year Raffi, the Bumble Bee, would take the seeds from the flowers and scatter them all over the hills and they would grow into very pretty flowers.

Jake would often pick them for his Mom and 'Teddy Blue' would always steal a few for himself when he thought the others weren't looking, he loved to eat the flowers! Jake and Raffi thought it was very funny.



Everyone in the village thought that because ‘Teddy Blue’ was born on the night of a shooting star it made strange things happen to him, things that didn’t happen to other bears.

For example, when something went wrong in the forest, for whatever reason, ‘Teddy Blues’ eyes would move from side to side, sometimes really fast and sometimes they would just get stuck in one position, usually upwards as if he was trying to look high high up into the sky. His whole body would shake and even go a little stiff. When this would happen poor ‘Teddy Blue’ couldn’t do anything to make it stop.



Luckily Jake and Raffi had seen this a hundred times before and they knew exactly what to do.

Jake would help 'Teddy Blue' get comfortable and Raffi would sit on his nose and sing his favourite soothing song, just as if they were back in the tepee on a cold night. If they could just help 'Teddy Blue' get to sleep he would feel soooo much better when he woke up and then they could play again.



When ‘Teddy Blue’ was afraid or when he was sick he would be very weak and sweated and sweated until his sparkly blue fur was wet and crumpled.

Jake always carried ‘Teddy Blues’ favourite blanket and a drink in his red bag in case he needed it, that’s what makes Jake super special, he’s always thinking of others, he even got his Mom to knit ‘Teddy Blue’ a jumper to keep him warm, it was soooo funny when he put it on, it was yellow and black stripes and he looked just like a bumble bee too, just like Raffi!



One day Raffi took care of ‘Teddy Blue’ while Jake went fishing on his own... ‘Teddy Blue’ was wondering when Jake would be back, it was getting dark and Raffi had worn him out with all his funny jokes and croaky singing, which was completely out of tune.

‘Teddy Blue’ wasn’t going to have to wait much longer because Jake was on his way back to the tepee after catching 7 fish. As he passed the hill, just outside the village, he spotted some amazing and mystical yellow flowers glowing in the light of the stars, it was very strange, they almost looked magical, they weren’t Raffi’s flowers he was sure of that, he’d picked enough and these glowed too brightly... perhaps they really were magic?



Without even thinking Jake started picking the flowers and putting them in his red bag.

He wanted ‘Teddy Blue’ to eat them, he knew he loved flowers, but this was somehow different, he had a funny feeling they would make him feel better and grow stronger and not sweat and not move his eyes from side to side, he was getting soooo excited that he forgot to pick up the fish he’d caught that day!

He ran all the way back to his friends. He called ‘Teddy Blue’ “look, look what I found you, eat them” Jake frantically told him, “see what happens”. Meanwhile Raffi was hysterically buzzing around the room trying to make sense of what Jake was saying, he was so confused, they didn’t look like his flowers.



Jake sat down and explained to them both how he came to find them and how he thought they were special magical yellow flowers that would make ‘Teddy Blue’ feel better.

It was truly amazing... the special yellow flowers really were magic ‘Teddy Blue’ was feeling much better and brighter each day. He began to grow stronger, he started to run, although a little wobbly, across the meadow, catch fish in the river and even reach up to the butterflies. He was soooo happy eating the special yellow flowers, because they were delicious too.



Everything was going well. Jake, ‘Teddy Blue’ and Raffi grew up together as brothers.

Jake's Grandpa even led them to find herds of bison and together they watched as the sunset behind the hills. But one day ‘Teddy Blue’ noticed that Jake was sad, very sad. His Grandpa was ill. He had been gradually losing strength, he was moving slowly, he spoke softly, and sometimes he would have trembling hands. Jake's Grandpa was very distressed because he could no longer go with them. And Jake did not know how to help.

‘Teddy Blue’ began to think and think and think.



On a bright, clear beautiful night ‘Teddy Blue’ looked up to the stars and asked his Mom

- "Mom, help me, what I can do to help Jake and Grandpa?"

He was sure he heard her reply because suddenly he thought of a solution. That night ‘Teddy Blue’ did not return to the tepee, instead he walked all night looking for the special magic yellow flowers that Jake and Raffi had always brought to him. He was getting tired but he knew he had to help his friends.



In the morning Jake and Raffi woke up early to find ‘Teddy Blue’ sitting in front of them, looking tired and holding lots and lots of magical yellow flowers...

-“These are for your Grandpa” ‘Teddy Blue’ blurted out and then he promptly went to sleep.

Little by little, day after day, Grandpa ate the magical yellow flowers and began to feel better. Each day getting stronger, moving faster and singing louder... His hands even stopped shaking!

All the villagers were soooo happy, they sang to the stars in the bright sky and danced and danced round the fire.



Raffi, the Bumble Bee was wildly buzzing around promising to carry these special seeds and drop them all over the hills so they would have all the special magical yellow flowers they would ever need.

‘Teddy Blue’ was quietly looking up into the sky mouthing the words “thank you” to his Mom, who he knew helped him that night.

Jake was dancing, patting his mouth making a “wa wa wa wa wa” noise, he couldn’t be happier... he didn’t want this moment to ever change!



So take note, if one of these evenings you see a mountain of bright glowing mystical looking yellow flowers and the sky is filled with stars, perhaps there is something magical waiting for you too!



Characters Introduction

Teddy Blue

I am Teddy Blue, Jake and Raffi are my friends. Since I was a cub I am not as big and strong as the other bears. Sometimes strange things happen to me, things that I cannot control. By luck, Jake and Raffi always take care of me. I really enjoy eating flowers!

Jake

I am an Indian boy, I live in a tepee and I like to go fishing in the river, run in the meadow, pick beautiful flowers for my mother, and laugh with Teddy Blue at Raffi's jokes. I try to take care of all the people in the village and, especially, of my friends, Teddy Blue and Raffi.

Raffi, the bumble bee

I am Raffi, the bumble bee. I love to make everyone laugh, particularly Teddy Blue. He really enjoys my songs. I take the seeds from the flowers and scatter them all over the hills, filling them with coloured flowers.

Jake's Grandpa

I am Jake's Grandpa. I like going with the children to see the herds of bison. Bison are normally quiet but they can be a hazard too. For some time now I cannot go with them because I feel weak and my hands tremble.

The Flowers

We are the flowers. We grow yellow and bright with the stars' light. We have a magical ingredient and, moreover, we are delicious!



Congenital (or paediatric) neurotransmitter diseases.

These are a group of genetic diseases involving the metabolism of neurotransmitters. Neurotransmitters are chemical messengers released by neurons to communicate with other neurons.

The disease causes defects at birth which mainly affect the central nervous system (fundamentally the brain) and can lead to serious neurological problems, especially if adequate treatment is not provided.

Symptoms associated with this group of diseases vary in severity. Milder cases present themselves with clumsiness in young children, psychomotor retardation, abnormal movements ... The more severe cases may present dystonia (uncontrollable movements), tremor, hypotonia (low muscular tone) or stiffness (excessive muscular tone), ataxia (clumsiness or uncoordinated gait), psychomotor retardation, hypomimia (face less expressive), oculogyric crises (abnormal eye movements in the form of rotations and fixed upwards or sideward gaze), excessive sweating, temperature instability and general autonomic dysfunction, sleep disturbances, and fluctuation of symptoms throughout the day.

Accurate diagnosis of these diseases is performed by analysing neurotransmitters' metabolites in the cerebrospinal fluid (CSF). CSF is obtained via a lumbar puncture. Although, in order to reach a definitive diagnosis, investigations of molecular studies are also required.

There are different diseases altering dopamine and serotonin metabolism, two critical neurotransmitters required for everyday living. As well as dopamine and serotonin it is possible that many other neurotransmitters are also compromised.

The story of “**Jake, Teddy Blue, Raffi, the bumble bee, and the mystical yellow flowers**” tries to explain, in an adapted and easy way, the symptoms associated with Aromatic Amino Acid Decarboxylase deficiency (AADC). AADC is essential in the metabolism of both dopamine and serotonin.



The pharmacological treatments used for patients with AADC deficiency mostly try to improve neurotransmission related to dopamine and serotonin, which cannot be normally formed in this disease. The treatment is based on new generation dopaminergic agonists which try to improve dopaminergic function in the neurons; namely Pramipexol and Ropinirol and drugs which inhibit the degradation of dopamine and serotonin, known as MAO inhibitors; namely Selegiline, Phenzelzine and Tranylcypramine. Patients suffering with AADC deficiency usually take multiple medications, carefully balanced, to improve or help stabilise their condition.

In Parkinson Disease (PD), a very prevalent condition in the general population, there is a degeneration of dopaminergic neurons that rises with age. Sometimes, patients suffering PD can share some symptoms with children affected by paediatric neurotransmitter disorders. This is why, in this story and in real life, there are common medications for both types of patients.

Teddy Blue is a little bear affected by Aromatic Amino Acids Decarboxylase (AADC). Due to this, his development is not like the development of other bears. He is not as strong as them, he is smaller, and he suffers episodes of dystonia, oculogyric crisis and excessive sweating.

Jake's Grandpa is an old man suffering Parkinson Disease, a deficiency in the dopaminergic function at the brain. This disease causes movement difficulties such as dystonia and tremor.

The mystical yellow flowers are magic flowers; they have in their composition dopaminergic substances that improve dopamine's function. They have therapeutic affects for both of AADC Deficiency and Parkinson Disease. For this reason, Teddy Blue and Jake's Grandpa can share them.

Congenital neurotransmitter diseases are extremely uncommon diseases and were first described some decades ago. However, in recent years there is an increasing awareness and more patients are being diagnosed.

There is a need for diffusion and divulgence of information relating to these diseases. This will allow for earlier diagnosis and treatment which may improve the quality of life of children suffering them.

With this story, although we haven't included all the symptoms, we can understand which are the main ones that AADC deficient patients suffer, moreover, the importance of helping them as much as possible to be comfortable in their daily life.

On the other hand, the story of **“Jake, Teddy Blue, Raffi, the bumble bee, and the mystical yellow flowers”** reinforces that, sometimes, research into rare diseases can be helpful for other patients with more common conditions, like Parkinson Disease.

In this story, whilst looking up at a bright starry sky, we can believe like **“Jake, Teddy Blue, Raffi, the bumble bee, and the mystical yellow flowers”** that love, friendship and understanding can truly make magical things happen!