

By: Risha Desai
Neel Manne

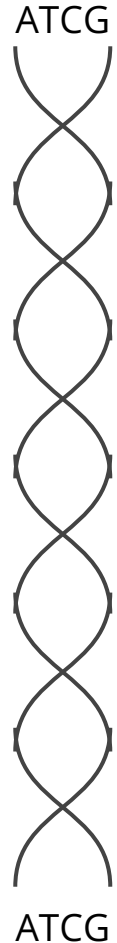


My Doctor Appointment Book

Cardiofaciocutaneous CFC

The Basics of Cardiofaciocutaneous Syndrome

Everyone inherits their genes from their parents. Half of the genes come from their mother and half from their father. That is why we look like our parents. There are 4 base pairs in a DNA strand; adenine, thymine, cytosine, and guanine (A,T,C,G). Genes are pieces of DNA that code for proteins. Proteins are building blocks that make us who we are. Sometimes DNA codes are altered (mutations or variations) and result in genetic disorders. The genes associated with CFC syndrome (BRAF, MEK1, MEK2, and KRAS).



About me

My name is _____.

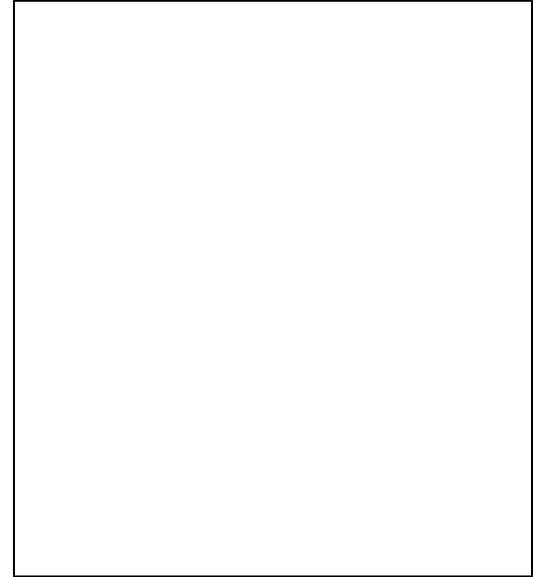
I am ___ old.

My most favorite thing to do is to _____.

I love to _____

I have CFC and special needs. Many parts of our bodies need extra care. Therefore we need to visit many doctors. Everytime I take a trip to the doctor's office, I color in a star for the specific body part or appointment.

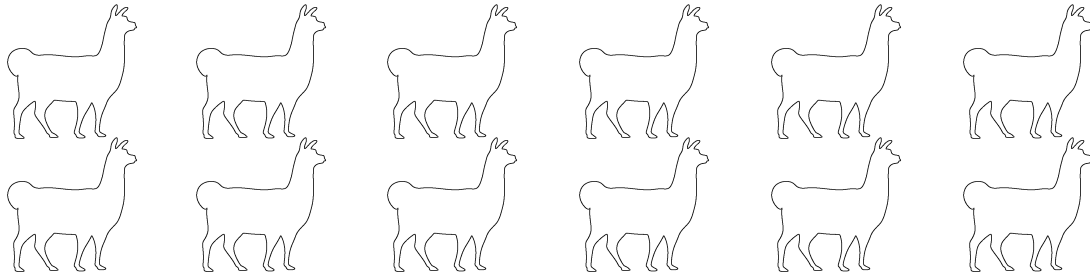
A photo of me:



Eyes

Everyone needs to get their eyes checked, but with my syndrome, I may need more frequent visits. I have eyes that are wider apart than most kids and my eyelids droop sometimes. My optic nerve in my eye also has some problems. I might need surgery to straighten my eyes. After surgery I still have to go to appointments for eye checkups.

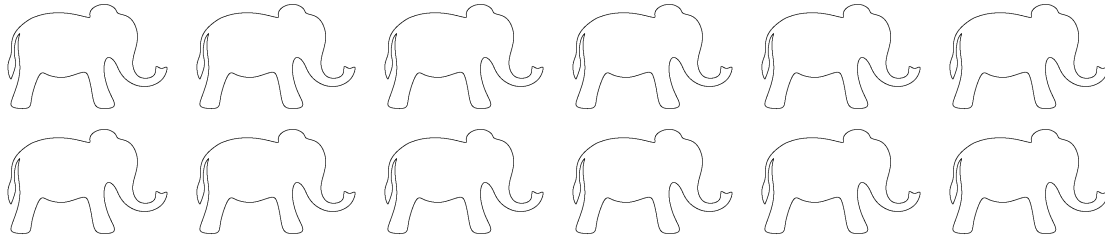
Color in a llama after each eye appointment



Brain

I have to see a neurologist because there are parts of my brain that are developed differently. Some warnings of neurological problems are not being able to talk as quick as others. It is important for me to get an imaging done of my brain to understand the present problems.

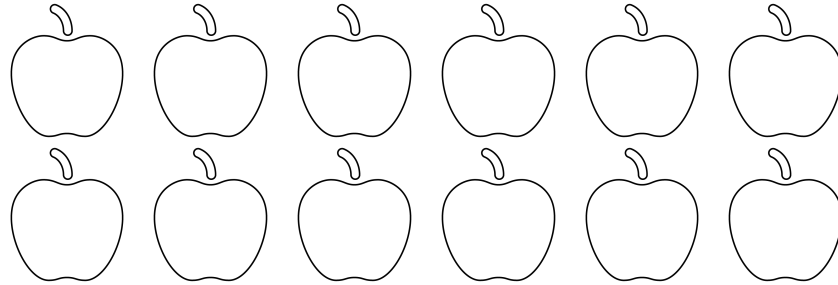
Color in a elephant after each neurology appointment



Food and Nutrition

Since I may have problems swallowing food or have reflux, I might not gain weight as easily, I may have to use a gastrostomy tube. This tube carries food to my stomach. The tube is a replacement of swallowing food. As I grow older, I will outgrow the tube and will be able to eat food through my mouth.

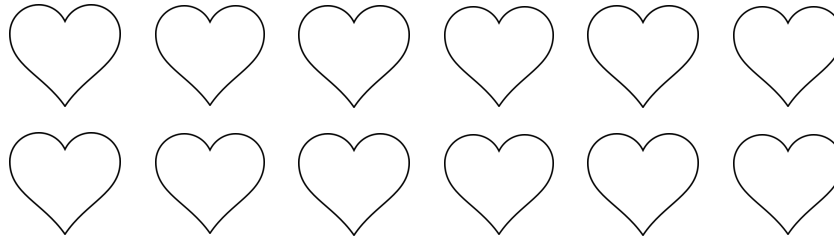
Color in a apple after each gastrointestinal appointment



Heart

Sometimes I have to see the heart doctor because I have problems with the rhythm of my heartbeat or my heart muscles become too thick. To help, the doctor will perform a special ultrasound to check the rhythm of my heart and then analyze it.

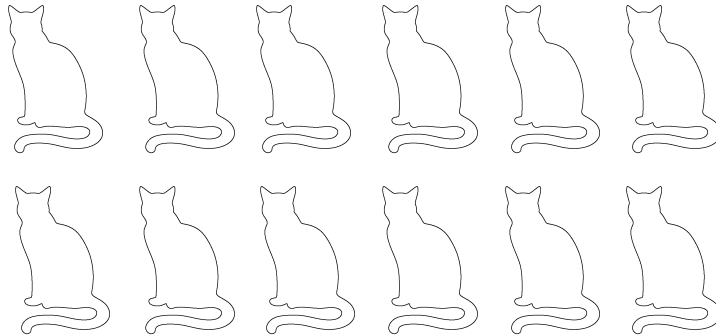
Color in a heart after each cardiac appointment



Lungs

I might have problems breathing. So, I will need to see a pulmonologist, because of problems with my lungs.

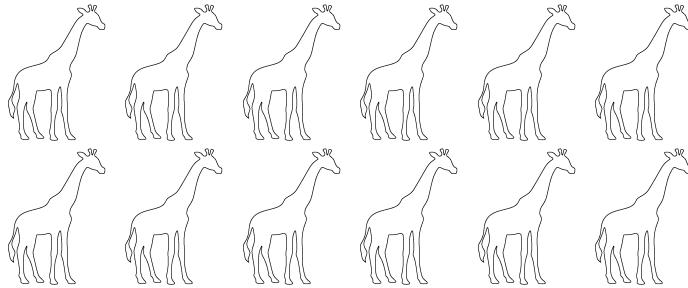
Color in a cat after each lung appointment



Bones, Joints, & Muscles

I have to visit my pediatric orthopedist and my physical therapist. I may have joint hyperextension, where my bones might move in the wrong direction. This can result in pain or my bones not in correct placement, and then I'll have to get surgery for that.

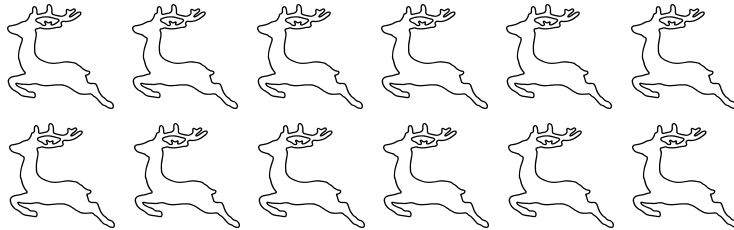
Color in a giraffe after each orthopedist or physical therapist appointment



Hormones

I might see my endocrinologist for growth hormones as I may have short stature. I may also see my endocrinologist because of premature puberty.

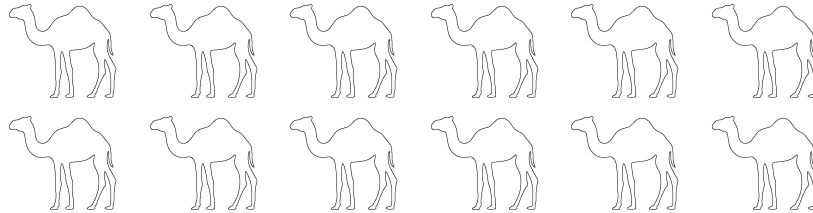
Color in a deer after each endocrinology appointment



Kidneys & Bladders

I can have problems with my kidneys that I may not be aware of. I may need ultrasounds and monitoring with nephrologists.

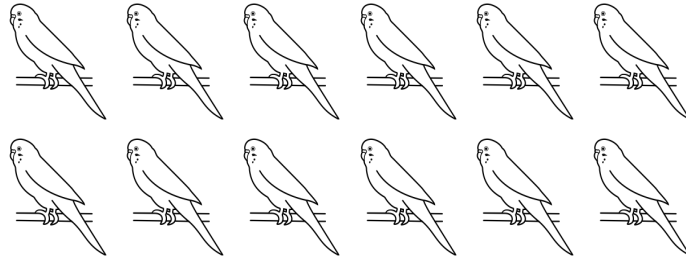
Color in a camel after each nephrologist appointment



Dermatology

I may have to see my dermatologist because I can develop dry, rough skin, thickening of my skin, or dark-enlarged moles. I must protect myself by using sunscreens and by visiting the dermatologist.

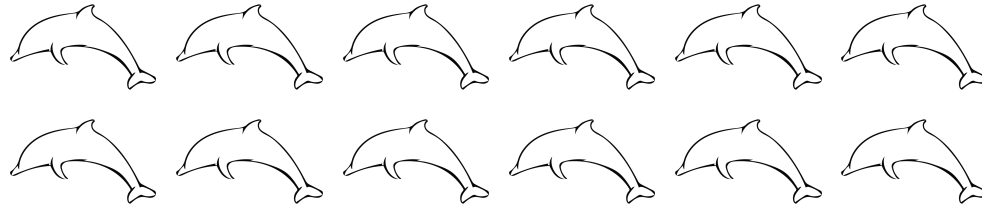
Color in a budgie after each dermatologist appointment



Therapies

I may take longer to meet my milestones than others and may need therapy services to help meet them faster. The different therapies will help me find ways to achieve my milestones and make improvements.

Color in a dolphin after each therapy appointment



Mentored by: Suma P. Shankar MD, PhD, University of California Davis

References:

- <https://www.cfcsyndrome.org>
- Rauen KA. Cardiofaciocutaneous Syndrome. 2007 Jan 18 [Updated 2016 Mar 3]. In: Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2022. Available from: <https://www.ncbi.nlm.nih.gov/books/NBK1186/>

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