International 22q11.2 Foundation

Newsletter March 2024

In this newsletter…

Greetings from the International 22q11.2 Foundation! We are eager to welcome spring and meet everyone at 22q at the Zoo events on May 19th (p. 1)!

Congratulations to Dr. Anne Bassett, who was named to the Order of Canada (p. 2). Also, the paper on the clinical recommendations for children with 22q11.2DS is an Editor's Choice in the GIM journal (p. 2)!

All 13 info sheets in the Gastrointestinal (GI) Series have now been published (p.3). We also bring you information on measles and how to prevent it (p. 4).

Have a look at the two research summaries. They center on remote vs in-person assessments (p.5) and language abilities and 22q differences (p. 6). In addition, meet Jayden (p. 7) and read the inspirational story of him and his mother.

We couldn't do our work without your support! Please consider making a donation using one of the many options (p. 8).

Happy reading!

22q at the Zoo Worldwide Awareness Day

Sunday May 19th, 2024

22q at the Zoo is an annual event that happens at zoos and other fun places all around the world, giving families, friends and professionals a chance to socialize, network and raise public awareness of 22q11.2 syndromes.

In this event developed by the International 22q11.2 Foundation, volunteers spread awareness by handing out information about chromosome 22q differences. Participants are recognizable by wearing official "**22q at the Zoo**" red T-shirts and "**Ask Me About 22q**" buttons.

Visit our Foundation's **22q at the Zoo** webpage to check for an event in your area. The webpage will be updated on an ongoing basis as more volunteers step up to host the event.

If there are no 22q at the Zoo events in your area, you can host one! Please visit the **Host a 22q at the Zoo Event** section on our website for more information. We even have an event planner letter ready for you! No zoo? No problem. Consider gathering at a park, a farm, or even an aquarium.

If you have any questions, please email us at info@22q.org. Thank you.

See you at the Zoo on May 19th!

Dr. Anne Bassett Named to the Order of Canada

Congratulations to Dr. Anne Bassett, Medical Director of the International 22q11.2 Foundation, who was appointed as a Member of the Order of Canada on December 28th, 2023. She was honored "for her research and clinical contributions to advancing our understanding of the genetics of schizophrenia, particularly in the identification and characterization of genetic subtypes of the disorder".

Dr. Bassett is the Dalglish Chair in 22q11.2 Deletion Syndrome (22q11.2DS) and has been caring for affected patients and their families for over 30 years. An adult with 22q11.2DS is 20 times more likely than someone in the general population to develop schizophrenia, a serious but treatable psychiatric illness. At the Dalglish Family 22q Clinic in Toronto, Ontario, Dr. Bassett and her team provide ongoing care for adults with 22q11.2DS and have a world-renowned clinical research program, with the aim to help adults with 22q achieve the best health and quality of life possible.

According to the website of the Governor General of Canada, the Order of Canada was created in 1967 to honor people who make extraordinary contributions to the country. People who are invested into the order have all enriched the lives of others and made a difference to Canada. They exemplify the Order's motto: DESIDERANTES MELIOREM PATRIAM ("They desire a better country"). Our Foundation is very proud that Dr. Bassett's contributions are being recognized by this special honor!

Anne S. Bassett, CM, MD, FRCPC
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Professor, Department of Psychiatry,
University of Toronto
Director, Dalglish Family 22q Clinic,
Toronto General Hospital
Director, Clinical Genetics Research Program,
Centre for Addiction & Mental Health

Clinical recommendations paper on Editor's Choice in Genetics in Medicine!

We are very happy to announce that the 2023 clinical recommendations for children with 22q11.2 DS is an Editor's Choice in the journal Genetics in Medicine (GIM)! GIM is the official journal of the American College of Genetics and Genomics (ACMG) and has a current impact factor of 8.822, ranking it 15th out of 175 journals in the category of Genetics and Heredity.

The International 22q11.2 Foundation would like to congratulate to all contributors for this recognition of their enormous efforts, and likewise compliments to all those who contributed to the adult and prenatal papers, which were also extremely well received.

A panel of international 22q experts, including some of our Medical Advisory Board Members, have worked on the clinical recommendations documents in the past few years. Multiple patient advocacy groups, including our Foundation, also provided input. The compilation of these clinical recommendations documents shows how research and clinical care work together in knowledge translation – experts learn from the patient they see, and the insights gained benefit people with 22q and their families around the world.

Please click the links below to access the 3 clinical recommendations papers:

- Updated clinical practice recommendations for managing children with 22q11.2 deletion syndrome
- Updated clinical practice recommendations for managing adults with 22q11.2 deletion syndrome
- Prenatal screening and diagnostic considerations for 22q11.2 microdeletions

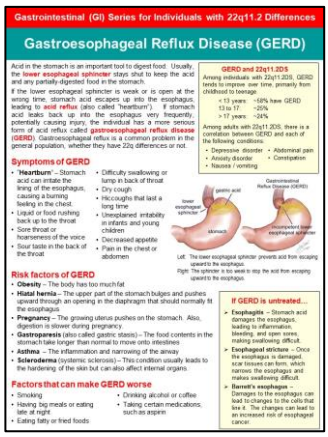
Gastrointestinal (GI) Series

More information sheets now available on our website

https://22q.org/symptoms-care/health-conditions-explained/

In our December newsletter, we mentioned the publication of the first four information sheets in the Gastrointestinal (GI) Series. We are happy to tell you that all 13 topics of the series are now available in the "**Health Conditions Explained**" section of our website.

Special thanks to Dr. Maria Mascarenhas, Pediatric Gastroenterologist and Nutrition Pediatrician; Joanne Loo, Programmatic and Educational Tool Developer; and Dr. David W. Low, Plastic Surgeon and Medical Illustrator.


Gastrointestinal (GI) Series for Individuals with 22q11.2 Differences

Gastroesophageal Reflux Disease (GERD)

Acid in the stomach is an important tool to digest food. Usually, the lower esophageal sphincter stays shut to keep the acid and any partially-digested food in the stomach.

If the lower esophageal sphincter is weak or is open at the wrong time, stomach acid escapes up into the esophagus, leading to acid reflux (also called "heartburn"). If stomach acid leaks back up into the esophagus very frequently, potentially causing injury, the individual has a more serious form of acid reflux called gastroesophageal reflux disease (GERD). Gastroesophageal reflux is a common problem in the general population, whether they have 22q differences or not.

GERD and 22q11.2DS

Among individuals with 22q11.2DS, GERD tends to improve over time, primarily from childhood to teenage:

< 13 years	~68% have GERD
13 to 17	~25%
> 17 years	~24%

Among adults with 22q11.2DS, there is a correlation between GERD and each of the following conditions:

- Depressive disorder
- Abdominal pain
- Anxiety disorder
- Constipation
- Nausea / vomiting

Symptoms of GERD

- "Heartburn" – Stomach acid can irritate the lining of the esophagus, causing a burning feeling in the chest.
- Liquid or food rushing back up to the throat
- Sore throat or hoarseness of the voice
- Sour taste in the back of the throat
- Difficulty swallowing or lump in back of throat
- Dry cough
- Hiccoughs that last a long time
- Unexplained irritability in infants and young children
- Decreased appetite
- Pain in the chest or abdomen

Gastroesophageal Reflux Disease (GERD)

Left: The lower esophageal sphincter prevents acid from escaping upward to the esophagus.

Right: The sphincter is too weak to stop the acid from escaping upward to the esophagus.

Risk factors of GERD

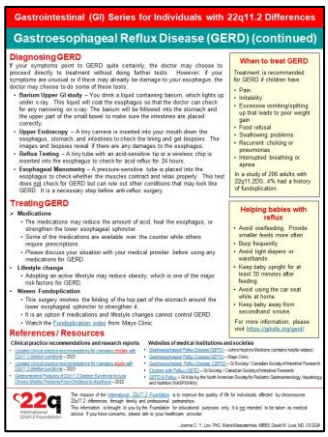
- Obesity – The body has too much fat
- Hiatal hernia – The upper part of the stomach bulges and pushes upward through an opening in the diaphragm that should normally fit the esophagus
- Pregnancy – The growing uterus pushes on the stomach. Also, digestion is slower during pregnancy.
- Gastroparesis (also called gastric stasis) – The food contents in the stomach take longer than normal to move onto intestines
- Asthma – The inflammation and narrowing of the airway
- Scleroderma (systemic sclerosis) – This condition usually leads to the hardening of the skin but can also affect internal organs.

Factors that can make GERD worse

- Smoking
- Having big meals or eating late at night
- Eating fatty or fried foods
- Drinking alcohol or coffee
- Taking certain medications, such as aspirin

If GERD is untreated...

- Esophagitis – Stomach acid damages the esophagus, leading to inflammation, bleeding, and open sores, making swallowing difficult.
- Esophageal stricture – Once the esophagus is damaged, scar tissues can form, which narrows the esophagus and makes swallowing difficult.
- Barrett's esophagus – Damages to the esophagus can lead to changes to the cells that line it. The changes can lead to an increased risk of esophageal cancer.


Gastrointestinal (GI) Series for Individuals with 22q11.2 Differences

Gastroesophageal Reflux Disease (GERD) (continued)

Diagnosing GERD

If your symptoms point to GERD quite certainly, the doctor may choose to proceed directly to treatment without doing further tests. However, if your symptoms are unusual or if there may already be damage to your esophagus, the doctor may choose to do some of these tests:

- Barium Upper GI study – You drink a liquid containing barium, which lights up under x-ray. This liquid will coat the esophagus so that the doctor can check for any narrowing on x-ray. The barium will be followed into the stomach and the upper part of the small bowel to make sure the intestines are placed correctly.
- Upper Endoscopy – A tiny camera is inserted into your mouth down the esophagus, stomach, and intestines to check the lining and get biopsies. The images and biopsies reveal if there are any damages to the esophagus.
- Reflux Testing – A tiny tube with an acid-sensitive tip or a wireless chip is inserted into the esophagus to check for acid reflux for 24 hours.
- Esophageal Manometry – A pressure-sensitive tube is placed into the esophagus to check whether the muscles contract and relax properly. This test does not check for GERD but can rule out other conditions that may look like GERD. It is a necessary step before anti-reflux surgery.

When to treat GERD

Treatment is recommended for GERD if children have:

- Pain
- Irritability
- Excessive vomiting/spitting up that leads to poor weight gain
- Food refusal
- Swallowing problems
- Recurrent choking or pneumonias
- Interrupted breathing or apnea

In a study of 206 adults with 22q11.2DS, 4% had a history of fundoplication.

Treating GERD

- Medications
  - The medications may reduce the amount of acid, heal the esophagus, or strengthen the lower esophageal sphincter.
  - Some of the medications are available over the counter while others require prescriptions.
  - Please discuss your situation with your medical provider before using any medications for GERD
- Lifestyle change
  - Adopting an active lifestyle may reduce obesity, which is one of the major risk factors for GERD.
- Nissen Fundoplication
  - This surgery involves the folding of the top part of the stomach around the lower esophageal sphincter to strengthen it.
  - It is an option if medications and lifestyle changes cannot control GERD
  - Watch the Fundoplication video from Mayo Clinic

Helping babies with reflux

- Avoid overfeeding. Provide smaller feeds more often
- Burp frequently
- Avoid tight diapers or waistbands
- Keep baby upright for at least 30 minutes after feeding
- Avoid using the car seat while at home
- Keep baby away from secondhand smoke

For more information, please visit https://gikids.org/gerd/

References/ Resources

Clinical practice recommendations and research reports

- [redacted]

Websites of medical institutions and societies

- [redacted]

22q

The mission of the International 22q11.2 Foundation is to improve the quality of life for individuals affected by chromosome 22q11.2 differences through family and professional partnerships.

This information is brought to you by the Foundation for educational purposes only. It is not intended to be taken as medical advice. If you have concerns, please talk to your healthcare provider.

Joanne C. Y. Loo, PhC; Maria Mascarenhas, MBBS; David W. Low, MD, 03/2024

GI SERIES

- **GI Issues in 22q11.2 Deletion Syndrome**
- **The Digestive System**
- **Feeding Difficulties**
- **Swallowing and Dysphagia**
- **Constipation**
- **Gastroesophageal Reflux Disease (GERD)**
- **Esophageal Dysmotility**
- **Gastroparesis**
- **Cholelithiasis (Gallstones)**
- **Nausea and Vomiting**
- **Autoimmune Issues in the GI System**
- **Inguinal Hernias**
- **Nonalcohol Fatty Liver Disease (NAFLD)**

Health Conditions Explained

HEART SERIES

The Heart and Normal Blood Flow
Ventricular Septal Defect (VSD)
Tetralogy of Fallot (ToF)
Truncus Arteriosus (TA)
Interrupted Aortic Arch (IAA)

DENTAL SERIES

Let's Talk 22q Teeth – Info for Families
Dental Health in Children with 22q – Info for Dentists

PALATE SERIES

The Velopharynx
Cleft Palate & Submucous Cleft Palate
Velopharyngeal Dysfunction: Introduction and Causes
Velopharyngeal Dysfunction: Diagnosis
Velopharyngeal Dysfunction: Surgery
Velopharyngeal Dysfunction: Speech Issues

MENTAL HEALTH SERIES

Mental Health and 22q11.2 Deletion Syndrome
Mental Health and 22q11.2 Duplication Syndrome
What Parents Can Do for Their Children
Attention Deficit Hyperactive Disorder
Autism Spectrum Disorder
Anxiety Disorders
Psychotic Disorders [Including schizophrenia]
Mood Disorders
Mental Health Q&A (Spring 2023)

22q GLOSSARY

Second Edition (217 entries)

About Measles

- **Measles is a serious respiratory infection caused by a virus.**
- Symptoms include:
  - High fever
  - Aches and pains
  - Runny nose
  - Coughing
  - Red eyes
  - Rashes
- Complications include:
  - Ear infection
  - Deafness
  - Pneumonia
  - Immune system having to re-learn how to deal with bacteria and viruses. The patient might get sick often in the next few years
  - 1 in 1000 patients have swelling of the brain, often resulting in permanent brain damage
  - 3 in 1000 patients have complications in the respiratory and nervous systems
  - Degenerative disease of the nervous system about 7 to 10 years down the road
  - Death
- Measles **spreads very easily** through coughing and sneezing.
  - 9 out of 10 people who come into contact with measles will be infected
  - The measles virus can stay in the air for 2 hours
- **Measles is preventable through vaccination (MMR or MMRV)**
  - Two doses of MMR vaccine are about 97% effective at preventing measles
  - One dose of MMR vaccine is about 93% effective
  - **MMR and MMRV vaccines are safe and effective**
  - Vaccination schedule
    [Travelling internationally | Not Travelling]
- For individuals who are **immune compromised**:
  - **There have been four studies of the safety of the MMR vaccine in people with 22q11.2DS which showed that most people can get the vaccine safely**
  - Consult with an immunologist if you are not vaccinated yet

Information about Measles

- Centers for Disease Control and Prevention (CDC) – Measles (Rubeola)
- Canadian Paediatric Society (CPS) – Measles

Research reports about MMR vaccines and 22q11.2 deletion syndrome

Vacinas Vivas em Crianças com Síndrome de DiGeorge/ Deleção 22q11.2 (Portuguese)
[Live Vaccine in Children with DiGeorge/22q11.2 Deletion Syndrome].

Miranda M, Martins AT, Carvalho S, Serra-Caetano A, Esteves I, Marques JG.
Acta Med Port. 2019 Aug 1;32(7-8):514-519.
doi: 10.20344/amp.9802.

Live vaccine use and safety in DiGeorge syndrome.

Hofstetter AM, Jakob K, Klein NP, Dekker CL, Edwards KM, Halsey NA, Baxter R, Williams SE, Graham PL 3rd, LaRussa P.
Pediatrics. 2014 Apr;133(4):e946-54.
doi: 10.1542/peds.2013-0831.

Live viral vaccines in patients with partial DiGeorge syndrome: clinical experience and cellular immunity.

Moylett EH, Wasan AN, Noroski LM, Shearer WT.
Clin Immunol. 2004;112(1):106-112. http://doi.org/10.1016/j.clim.2004.02.008

Safety of live viral vaccines in patients with chromosome 22q11.2 deletion syndrome (DiGeorge syndrome/velocardiofacial syndrome).

Perez EE, Bokszczanin A, McDonald-McGinn D, Zackai EH, Sullivan KE.
Pediatrics. 2003 Oct;112(4):e325.
doi: 10.1542/peds.112.4.e325.

Comparing remote vs in-person assessments

Remote assessment of the Penn computerised neurocognitive battery in individuals with 22q11.2 deletion syndrome

White LK, Hillman N, Ruparel K, Moore TM, Gallagher RS, McClellan EJ, Roalf DR, Scott JC, Calkins ME, McGinn DE, Giunta V, Tran O, Crowley TB, Zackai EH, Emanuel BS, McDonald-McGinn DM, Gur RE, Gur RC. J Intellect Disabil Res. 2024 Jan 16. https://doi.org/10.1111/jir.13115 . Epub ahead of print. PMID: 38229473.
This article requires payment or institution subscription.

Online interactions and services have become very common. Even some healthcare appointments and tests can be done online. Have you ever asked whether remote tests are as good as the ones done in person?

A group of researchers in the Children's Hospital of Philadelphia (CHOP) and University of Pennsylvania compared remote and in-person assessments of the Penn computerised neurocognitive battery (CNB). CNB is a well-established and widely-used set of 12 tests and takes 1 hour to complete. The tests look like computer games but check a person's executive function, episodic memory, complex cognition, and social cognition. Basically, how well the person thinks, reasons, plans, switches between tasks, responds to situations, self-inhibits, and remembers things.

The study involved 384 individuals with 22q11.2 deletion syndrome (22q11.2DS) who were between 7 and 35 years old, in stable health, and had an IQ of at least 70. 222 CNB tests were done in person, while 162 were done remotely. In remote tests, participants were asked to share their screen with the test coordinator. The coordinator gave all the instructions online in real time.

When the researchers looked at the accuracy of the results between the in-person group and the online group, there was no significant difference. In 3 of the 11 tasks, the in-person participants' reaction time was slower than in remote settings. The researchers conclude that for the Penn CNB, remote and in-person assessments are equivalent.

Knowing that a remote Penn CNB assessment is as good as an in-person one is great news for families affected by 22q11.2DS. Taking a test online means that the affected person does not have to travel and can instead do it in the comfort of their own home. When it is easier and more convenient to take the tests, more people may be willing to take them. Researchers can then recruit more people from more diverse backgrounds in their studies, making the studies more applicable for everyone and enabling scientific discoveries.

Neurocognition and 22q11.2DS

Neurocognition refers to the brain processes that allow people to gain information, think, and reason. These processes include the ability to stay focused, remember facts, understand and use information, etc.

Children with 22q11.2DS often have deficits in neurocognition. (For more information, see the Updated clinical practice recommendations for managing **children** with 22q11.2 deletion syndrome). Many children also experience a decline in their cognitive abilities. Early cognitive decline is strongly related to an increased risk of developing a psychotic illness, a type of treatable mental health condition in which the patient has difficulty telling apart what is real and what is not. In general, changes in emotions, thinking, behavior, and functioning may be early signs of mental health issues. It is important to identify these often subtle changes in order to intervene as early as possible and yield the best results.

The Penn CNB has been used for cognitive assessments in different populations, including in individuals with 22q11.2DS. The results can provide an indication of the person's cognitive abilities, which can help healthcare providers figure out the needs of the affected individuals and their families.

Caution: While a remote Penn CNB is as good as an in-person one, we do not know if the same is true for other tests.

Language abilities and 22q differences

Language Profiles of School-Aged Children with 22q11.2 Copy Number Variants

Verbesselt J, Solot CB, Van Den Heuvel E, Crowley TB, Giunta V, Breckpot J, McDonald-McGinn DM, Zink I, Swillen A. Genes. 2023; 14(3):679. https://doi.org/10.3390/genes14030679. This article can be accessed for free.

Language deficits are known to be a major feature of 22q11.2 deletion syndrome (22q11.2DS). This study is the first to use standard language assessment test to evaluate the language abilities of children with 22q11.2 duplication syndrome (22q11.2DupS).

About the Study

Research locations (combination of 2 sites):

- CME-Leuven (Center for Human Genetics), Belgium (Dutch)
- Children's Hospital of Philadelphia (CHOP), PA, USA (English)

Study population:

- 29 children with 22q11.2DS and 29 with 22q11.2DupS
- Age range: 6 to 16 years

Language assessment test:

- Clinical Evaluation of Language Fundamentals (CELF)

What does the language test CELF measure?

- Sentence comprehension
- Linguistic concepts
- Word structure
- Word classes
- Following directions
- Formulated sentences
- Recalling sentences
- Understanding spoken paragraphs
- Word definition
- Sentence assembly
- Semantic relations
- Reading comprehension
- Structured writing
- Pragmatics profile

Source: CELF-5 Test Objectives and Descriptions

Results

- Many children with 22q difference (whether a deletion or duplication) have mild to moderate language impairments.
- The distribution of scores for children with 22q11.2DupS is in between those of 22q11.2DS and the norm.
- Children with the duplication who had delayed milestones as infants did worse than those without delayed milestones.
- Multiple aspects of language use were impaired in both children with the deletion and those with the duplication.
- A previous study using a parent survey found that 47% of children with 22q11.2DupS had language deficits. The current study, which uses the CELF test, found the rate to be 62% This higher rate found using the more objective method indicates that the previous finding may have been an underestimation of language deficit in 22q11.2DS.

Communication Problems	Kids with 22q11.2DS	Kids with 22q11.2DupS
Parent-reported in Children's Communication Checklist (2022 study in Belgium)	79%	47%
CELF language assessment (current study)	83%	62%

Take Home Message

- Many children with 22q11.2DupS have language deficits.
- Regular follow up of language development using standardized testing is recommended.
- Early identification of deficits allow access to support and reduce long term difficulties.

Meet Jayden

Jayden was born in Burlington, Iowa September 7th, 2009, and is now 13 years old. His mother says it has been hard on him with his syndrome, but he's learning every single day. He has a caring heart and when it comes to his family and friends, he will stick up for you.

Nicole, Jayden's mother is personally familiar with 22q since she has 22q deletion. For this reason, she was tested while pregnant, she just wanted to know what she was dealing with. When the results came in it was confirmed Jayden was 22q as well. She says, "It was hard at first to know he had the syndrome, but we are doing great!" Currently, Jayden attends public school in special education. If you ask him he says he doesn't like school but his mother says once he gets there he enjoys it! He graduated from speech therapy during 5th grade which makes him and his mom happy, and as she says, "his speech is better than mine." School has been difficult for him, he used to get in trouble but as he has matured this has gotten so much better.

Medically he is doing well, he doesn't follow a special diet but still has some behavior problems but this improves with maturity. How are Jayden and his mom dealing with these difficulties? She says, "I try to get him to talk to me. Sometimes it's hard for him to open up. We get through things together and stay strong for each other."

Jayden loves being with family and friends, video games, football, and basketball. His favorite sport is basketball, and he loves playing the game so much that he plans to go pro! I think when he gets older, he wants to go pro.

What advice does Nicole share with other parents of 22qties? As a parent with 22q herself, she says, "Don't let this syndrome define who you are. We are who we are for a reason."

She also shares, "It's okay to cry and struggle with things just keep moving forward because we are doing the best we can. It's hard but we are awesome and strong!"

To read more inspirational stories, please visit: **https://22q.org/inspirational-stories/**

The International 22q11.2 Foundation would love to hear personal stories about friends and family persevering and thriving with 22q! Please download this questionnaire and email your info to us at info@22q.org, along with two, high quality photos, and we will contact you for more info! We look forward to hearing from you!

Donate Now

Our mission: to improve the quality of life for individuals affected by chromosome 22q11.2 differences through family and professional partnerships.

What would we do with the funds:

- Support Research
- Support family conferences
- Support awareness
- Support Newborn Screening
- Support and raise awareness for 22q!

Your support makes a difference!

There are so many options for **Team 22q Fundraising**! Take part in one of the Foundation's event or create your own.

Please visit the **Donate** page on our website for information on donating online, by mail, or via other methods.

Please also **Shop** on our webstore.

For more information, please visit our website at **www.22q.org** or email us at **info@22q.org**.

Team 22q Fundraising

- **Tell 22 Friends** about 22q
- Become a **monthly donor**
- Be a 22q Citizen Journalist and share your event on social media!
- Plan a **22q at the Zoo** event
- Plan a **22K for 22q** event
- (It can also be 2.2K!)
- Create Your Own Event or Fundraiser Contact us at **info@22q.org**

Thank you!

Connect With Us

The mission of the **International 22q11.2 Foundation** is to improve the quality of life for individuals affected by chromosome 22q11.2 differences through family and professional partnerships.
This information is brought to you by the Foundation for educational purposes only. It is not intended to be taken as medical advice. If you have concerns, please talk to your healthcare provider.

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