Immune System Series for Individuals with 22q11.2 Differences

Allergies and Asthma

Allergies happen when the immune system over-reacts to a foreign substance that gets into the body, and reactions can range from very minor to life-threatening. Asthma is a long-term lung disease that when flared up, makes breathing difficult. Asthma attacks can be triggered by many things, including allergies. Allergies and asthma do not cause fevers and are not contagious. Researchers do not know the cause of either condition, but both are serious and require long-term, careful management.

Allergens and the Immune Reaction

A substance that triggers an allergic reaction is called an allergen.

 When an allergen enters the body of a person who is allergic to it, the immune system makes special antibodies (IgE) that will bind to mast cells. The mast cells release histamine, which causes the allergy symptoms.



Allergies in people with 22q differences

Frequency of allergies in children with 22q11.2DS (vs. those without 22q11.2DS)

- Asthma 41% (24%)
- Rhinitis 32% (40%)
- Eczema 15% (22%)
- Food allergy 11% (5%)
- Medication allergy 19% (1%)

Source: Chromosome 22q11.2 deletion syndrome and DiGeorge syndrome

Asthma and eczema have been seen in children with 22q11.2DupS, but the prevalence is not known.

Common allergies

Type of Allergen	Route	Main Symptoms
Airborne – pollen, animal dander, dust mites, mold	Breathing	Hay fever (allergic rhinitis) – sneezing, itchy skin, runny/stuffy nose, wheezing, tiredness, watery or swollen eyes
Food – peanuts, tree nuts, milk, eggs, wheat, soy, shellfish, etc.	Eating	Tingling in the mouth; swelling of the lip, face, tongue, or throat; hives; itchiness, stomach cramps, vomiting, diarrhea, anaphylaxis
Insect venom and stings – bees, wasps, etc.	Stinging	Swelling, hives, itchiness, skin warmth and color change, shortness of breath, anaphylaxis
Medications – (various)	(various)	Swelling, hives, itchiness, shortness of breath
Latex – gloves, balls, balloons, bandages etc.	Touching	Skin irritation (contact dermatitis), hives, runny nose, difficulty breathing

Anaphylaxis

- Anaphylaxis is a severe, life-threatening allergic reaction.
- Symptoms include (but are not limited to) swelling, difficulty breathing and/or swallowing, and drop in blood pressure.
- If anaphylaxis happens, inject epinephrine (e.g. EpiPen®) into the thigh immediately. A newer version that is given into the nose is available.
 This medication decreases the severity of the symptoms and gives the patient some time to seek medical help.
- Call 911 (or your <u>local emergency number</u>) symptoms can come back.

Managing Allergies

- Some medications (e.g. anti-histamine and nasal sprays) may decrease the symptoms.
- Avoid the specific allergen(s) that trigger the person's allergic reactions.
- Always carry two epinephrine injectors (e.g. EpiPen®) if you have ever experienced anaphylaxis to something you might be exposed to again.

Diagnosing Allergies

An allergist can do a skin prick test to check what a person is allergic to. He/she will use a panel of thin needles (each with a different allergen) to prick the skin of the patient and check which one(s) the skin reacts to.



Eczema

Eczema (atopic dermatitis) is a condition in which patches of skin get dry, bumpy, and itchy. Contact with an allergen can trigger the immune system to react, causing inflammation and eczema on the skin. For more information, see the <u>Eczema</u> webpage from the Cleveland Clinic.

Immune System Series for Individuals with 22q11.2 Differences

Allergies and Asthma (Continued)

Asthma Attacks

- People with asthma breathe normally when they are not having attacks.
- The frequency of asthma attacks is different from person to person.
- During an asthma attack, a person finds it hard to breathe because:
 - The muscles in the airway **tightens**. The airway becomes **narrower**, making it difficult for air to flow freely.
 - The lining of the airway **swells** up, further narrowing the airway.
 - The body produces more mucus, **clogging** up the airway.
- In addition to finding it hard to breathe, the person will likely be coughing and wheezing (making a whistling sound).

Asthma Triggers

- Asthma triggers are different from person to person. Examples:
 - Airborne allergens: pollen, dust mites, pet dander etc.
 - Air pollutants: smoke, chemicals from factories or cars
- Physical exercise
- Mold and pests
- Cold air
- Respiratory infections

Managing Asthma

- Follow the asthma plan specific for the person. The types and frequencies of medications are different from person to person.
- Follow vaccination recommendations for influenza ("the flu") and pneumonia
- Learn to recognize warning signs of an asthma attack (e.g. coughing, wheezing, shortness of breath)
- Avoid the specific triggers
- Asthma management usually relies on a controller inhaler for prevention of wheezing and a relief inhaler for an attack - but management is individualized depending on the severity and the frequency of attacks or flares
- The use of the relief inhaler too often means that the controller strategy needs to be optimized. Seeking specialist care is suggested.

Resources

- Allergies Mayo Clinic
- Allergies Cleveland Clinic
- Asthma Mayo Clinic
- Asthma Cleveland Clinic
- Anaphylaxis Cleveland Clinic
- Epinephrine Auto-injector The American College of Allergy, Asthma, & Immunology
- What is Epinephrine Allergy & Asthma Network
- Eczema Cleveland Clinic

Diagnosing Asthma

The doctor will try to rule out other possible conditions by doing a physical exam and asking about the symptoms.

He/she may also use spirometry or a peak flow meter to see how well the lungs work. This may be done before and after the use of a medication called a bronchodilator, which opens the airway. A person who has asthma will have better lung function test result after using a bronchodilator.

See other possible tests on these webpages: Asthma (Mayo Clinic) and Asthma (Cleveland Clinic).

Using the Correct Inhaler



People with asthma often have more than one inhaler. Some may hold medications for long-term asthma control, while others are quick-relief (rescue) medications that open swollen airways. It is very important to use the right inhaler at the right dose at the right time.

- Updated clinical practice recommendations for managing [children adults with 22q11.2 deletion syndrome - 2023
- Chromosome 22q11.2 deletion syndrome and DiGeorge syndrome -2019

Graphics

- Mast cell releasing histamine, adult using inhaler <u>Biorender.com</u>
- Allergy cartoon Freepik



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.