Autoimmune Disorders

Autoimmune disorders happen when the immune system mistakenly attacks our own healthy tissues. There are many types of autoimmune disorders. This info sheet will focus on the ones seen in people with 22q differences.

Autoimmune Cytopenias

Cytopenia means there is a lower levels of blood cells. Both immune thrombocytopenia (ITP) and autoimmune hemolytic anemia (AHA) are **autoimmune cytopenias**, where the immune system destroys the cells.

Immune Thrombocytopenia (ITP)

- Thrombocytopenia means having a lower level of platelets than normal
- ITP is also called idiopathic thrombocytopenic purpura
  - Idiopathic means unknown cause, and purpura means purple bruises
- **Platelets** are cells that help blood clot and stop bleeding (see right panel). In ITP, **the immune system attacks and destroys platelets**, resulting in a lower level of platelets in the blood and a difficulty with blood clotting.
- ITP is diagnosed by measuring platelet levels in the blood.
- Symptoms (may not be obvious) include:
  - Easy bruising
  - Purple spots on the skin
  - Heavy menstrual flow (period)
  - Bleeding in the mouth or nose
  - Blood in urine (pee) or stools (poop)
- People with ITP are advised to **avoid contact sports and certain medications that increase bleeding risk** (e.g. Aspirin, Advil, Motrin, etc.).

Autoimmune Hemolytic Anemia (AHA)

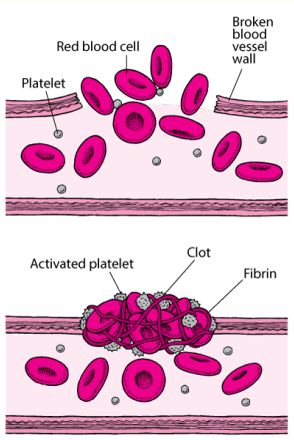
- Hemo- means blood, -lytic means burst, and anemia means having a lower level of red blood cells than normal.
- **Red blood cells (RBCs)** are cells that carry oxygen from the lungs to different parts of the body.
- In AHA, **the immune system attacks and destroys RBCs**, reducing their life from >100 days to just a few days. There are not enough RBCs in the blood to bring enough oxygen around the body.
- AHA is diagnosed by measuring RBC levels in the blood.
- Symptoms may not be obvious if the anemia is mild. They include:
  - Weakness and tiredness
  - Shortness of breath
  - Pale skin color
  - Light-headedness

Treatment Options for ITP and AHA

- Use of **medications** to slow down the destruction of the platelets or to increase their production.
- Injection of **antibodies** to target the autoimmune antibodies that are targeting the platelets or RBCs
- **Surgery** to remove the spleen (where platelets or RBCs are destroyed). However, not having a spleen increases the risk of infections.

Blood Clotting

When a blood vessel breaks, platelets change shape and stick with each other, red blood cells, and other proteins. This clump plugs the break and stops blood from flowing out of the vessel. i.e. The clot stops the bleeding.



Source of diagram:
How Blood Clots (Merckmanuals.com)

Autoimmune Cytopenias and 22q11.2DS

Autoimmune cytopenias, which include ITP and AHA, happen in ~8% of people with 22q11.2DS. The risks are higher in those with:

- Persistent **hypocalcemia**
- **Low levels of naïve T cells** (T cells that have never battled pathogens)
- **Low levels of class switched memory B cells** (B cells that secrete variants of antibodies)

See the original research publication and the commentary for more info.

Another study found that having at least 1 **severe infection** would strongly increase the risk of autoimmune disease.

Immune System Series for Individuals with 22q11.2 Differences

Autoimmune Disorders (Continued)

Juvenile idiopathic arthritis (JIA) / Juvenile rheumatoid arthritis (JRA)

- JIA happens when **the immune system attacks the tissues and fluids inside the joints** in the body.
- Symptoms may happen once in a while or continue for a long time.
  - Joints that are swollen, stiff, warm, painful, and hard to move
  - Swollen lymph nodes
  - Fever
  - Rash
  - Decreased appetite
  - Fatigue (tiredness)
- JIA may also lead to the **inflammation of the eyes**, but there may be **no symptoms early on.** It is highly recommended that children with JIA be **checked by an ophthalmologist as soon as possible to avoid vision loss**.
- It is difficult to diagnose JIA because many other medical problems can cause joint pains. Imaging and blood/urine tests will help provide with the diagnosis.
- There is no cure for JIA, but the symptoms can be managed using medications, exercises, as well as physical and occupational therapies.

Joints

A **joint** is the place where two bones meet. Each person has hundreds of joints. Learn about the structure of joints on Merkmanuals.com.

JIA & 22q11.2DS

JIA occurs in ~2% of children with 22q11.2DS. Rheumatoid arthritis (adult form of JIA) also happens in adults with 22q11.2DS.

Vitiligo

- Vitiligo happens when **the immune system destroys melanocytes**, which are cells that produce pigment for the skin.
- Small patches of the skin become white and they become larger over time.
- There is no cure for vitiligo, but symptoms can be managed using medications and light therapies.

Vitiligo & 22q11.2DS

Vitiligo ("vit-il-EYE-go") has been reported in a small proportion of children and adults with 22q11.2DS.

Graves' Disease

- Graves' disease (autoimmune thyroiditis) happens when **the immune system attacks the thyroid gland**, making it produce too much thyroid hormone. Since the thyroid hormone plays an important role in many parts of the body, Graves' disease leads to many possible symptoms. The common ones are:
  - Rapid heartbeat (palpitations)
  - Increased appetite
  - Weight loss
  - Feeling shaky and/or nervous
  - Diarrhea or more frequent bowel movements
- Graves' disease can be diagnosed using blood tests, thyroid scan (involves radioactive iodine), or an ultrasound to check the blood flow to the thyroid gland.
- There is no cure for Graves' disease, but the thyroid hormone level can be managed using medications, radioactive iodine, or a thyroid removal surgery.

Hyperthyroidism

In hyperthyroidism, the thyroid gland produces too much thyroid hormone, causing many body processes to speed up. Grave disease is one of the causes of hyperthyroidism.

Graves' Disease & 22q11.2DS

Graves disease has been reported in a small proportion of children and adults with 22q11.2DS.

Resources

- Immune thrombocytopenia (ITP) – Mayo Clinic
- Idiopathic thrombocytopenic purpura – Johns Hopkins Medicine
- Autoimmune hemolytic anemia – Boston Children's Hospital
- Immune hemolytic anemia – Mount Sinai Hospital
- Juvenile idiopathic arthritis – Mayo Clinic
- Juvenile idiopathic arthritis – Cleveland Clinic
- Vitiligo – Mayo Clinic
- Vitiligo – Cleveland Clinic
- Graves' disease – Mayo Clinic
- Graves' disease – Cleveland Clinic
- Updated clinical practice recommendations for managing [**children** | **adults**] with 22q11.2 deletion syndrome – 2023

International 22q11.2 Foundation

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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