AADMD: UOS Student Chapter
AMERICAN ACADEMY OF DEVELOPMENTAL MEDICINE AND DENTISTRY

# THALASSEMIA

VOLUME 1 ISSUE 3

AADMD

NEWSLETTER

Editor in chief

DR. MOHAMMED B.ESTAITIA

Editor

DIMA SALEH

# CONTENTS

# THE BOARD

SARAH JUMAAH

## AN INTERVIEW WITH: SIMA HABRAWI


**Q: What part of being the board's scientific officer do you enjoy the most?**

A: "What I enjoy most about my position is being able to truly reach out to the community and connect to people. Whether I'm contacting healthcare providers, parents, centers or influencers I truly enjoy this connection and the ability to raise awareness!"

**Q: What are your goals in life?**

A: "My career goal is to become a Pedodontist with a sub specialty in Craniofacial sciences and become part of the Cleft Lip and Palate management team. I would also want to be involved in Public Health and awareness, and be in an executive position in a global entity like the FDI or WHO to help vulnerable communities around the globe."

**Q: What is your vision for the association?**

A: "The AADMD-UOS is the only branch in the MENA region which puts a great deal of responsibility to represent a big part of the globe internationally. We have put a "master plan" to reach out to more countries in the MENA region and establish deeper roots in the UAE. We were the pioneers this year in doing 2 virtual awareness campaigns reaching over 1000 people and getting continuous engagement in 3 different languages for a period of 6 weeks.
Reaching out to more countries will also allow us to establish bigger events on a more international level with bigger exposure."

**Q: Where do you see the association in two years?**

A: "Definitely having a bigger and wider international exposure with more collaborative events with governmental organizations and NGOs. I would like to see us establishing a stable opportunity for students to volunteer not just in events but also in Public Health projects globally."

**Q: Did holding the position of scientific officer instill new qualities in your personality?**

A: "Yes, definitely, any leadership position allows you to grow and become a better person and leader. It definitely improved my time management skills, communications skills and public speaking ability! It also made me more aware of some challenges people of determination face on a daily basis! Which motivated me to target such challenges and improve them."

## AN INTERVIEW WITH: MARYAM FADHUL


**Q: Who is your role-model in life?**

A: "It is my mother. No words can describe her but she is ambitious, kind and she is the one that helped me become the strong woman I am today."

**Q: How did being part of AADMD improve your career as a health-professional?**

A: "It helped me with communicating with patients, especially ones with intellectual disabilities."

**Q: What part of being an editor do you find to be the most difficult?**

A: "I have been an editor for two years now and there are no difficulties in my job mainly because of the supportive environment I am surrounded by. Our board's president is very supportive of my team and I which gives us more confidence to excel at each task."

**Q: How far would you say AADMD's outreach has expanded throughout the region?**

A: "Honestly, we did not have a large audience at the start but many people began reaching out to us more ever since the last board was in-charge mainly because of the different events we organized and the many social media platforms we started. This expanded our outreach within our university and the entire country on a larger scale."

**Q: What message would you like to send to new members or people considering joining the association?**

A: "Many people should join us because we should give back to our community. Joining AADMD was the best decision I have ever made because it not only helps others but it helps you in many different ways as well."


# AN OVERVIEW

SARAH JUMAAH

Although it is amongst the most commonly inherited blood disorders in the UAE, with at least 8.5% of UAE's population either being affected or carriers according to the UAE Ministry of Health and Prevention, many are unaware of thalassemia and cannot distinguish between its different types. So, what is thalassemia? Simply put, it is an inherited blood disorder which results in the formation of abnormal hemoglobin (the protein molecule found in red blood cells responsible for carrying Oxygen) leading to the destruction of red blood cells, and eventually, developing anemia. Some thalassemia patients present symptoms such as fatigue, pale/ yellowish skin, slow growth, dark urine, facial bone deformities and abdominal swelling. Complications can include iron overload and infections if the condition is moderate to severe.

The type of thalassemia depends on the defective gene present in the patient's DNA and the hemoglobin chain it codes for, giving us types alpha and beta. In other words, Alpha-thalassemia is due to faulty alpha hemoglobin chain synthesis, while Beta-thalassemia is due to faulty beta hemoglobin chain synthesis. The severity of thalassemia, classified as either major and minor, depends on the amount of defective thalassemia-related genes one inherits from their parents, manifesting as different intensities of clinical symptoms patients develop.

Image showing the normal structure of a hemoglobin protein with a pair of alpha and beta chains each. According to the type of gene defect the patient has, the corresponding hemoglobin chain will be affected, hence determining the type of thalassemia, while severity depends on the amount of hemoglobin-coding gene defects one has.

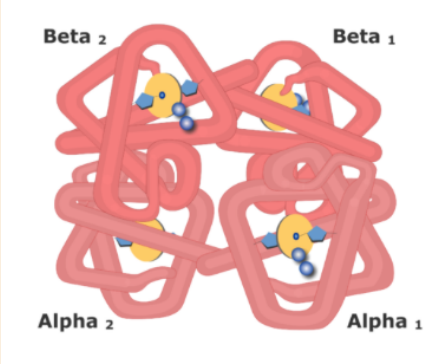



Image Courtesy of ©GetBodySmart 2021.

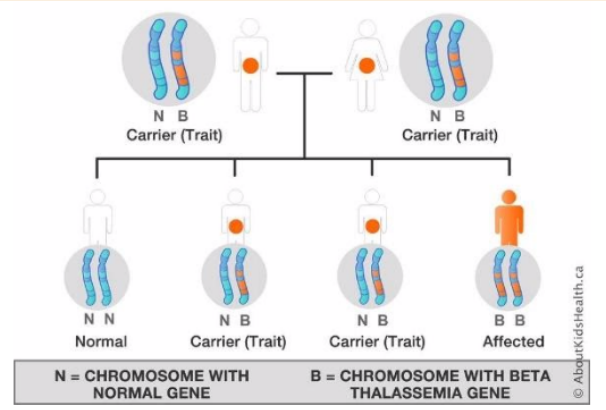



Image courtesy of ©2021 The Hospital for Sick Children.

Image showing hereditary patterns of thalassemia; it follows the recessive hereditary pattern in which both parents have to have the gene in order for their offspring to have thalassemia.

Prevention usually entails premarital screening to check for the presence of the defective genes, but in other cases, prevention is not possible. Treatments are inclusive of medication, procedures, self-care and nutrition depending on the severity of thalassemia; If severe, iron chelators are administered to help eliminate excess iron from the patients blood and bone marrow transplantation is performed, introducing healthy stem cells to the patients circulatory system which aids the formation of new, healthy red blood cells. Blood transfusions are performed routinely along with lifestyle modifications including exercise, consuming dairy products, teas and coffees, and avoiding foods rich in iron and proteins.

# COMMUNITY'S VOICE:


**LIVING WITH THALASSEMIA MAY POSE HURDLES IN THEIR PATIENT'S DAILY LIVES.**
**IN THIS SEGMENT, WE WILL TOUCH ON THALASSEMIA PATIENT'S CONCERNS, DIFFICULTIES AND ASPECTS OF THEIR LIVES THAT WE AS A COMMUNITY NEED TO BE AWARE OF.**

## 1. WHAT KIND OF DIFFICULTIES DO PEOPLE WITH THALASSEMIA OFTEN FACE?

Treatment-wise, patients must have blood transfusion every three weeks to maintain their levels of healthy blood cells, this can start as early as infancy, however, the quality of Thalassemia care heavily depends on which part of the world you live in, many countries do not have access to blood testing kits to ensure the blood's safety from viruses, additionally they could lack chelation medicine which prevents iron overload, that could lead to liver complications and heart failure.

Life-style wise, transportation seems to be one of the major concern as heavy physical activity can be quite strenuous to some.

## 2. WHAT ARE THE CONSIDERATIONS YOU KEEP IN MIND WHEN DEALING WITH THALASSEMIA PATIENTS?

The whole journey will seem confusing and difficult to the patients and their families, so we must ensure sufficient knowledge about their situation, additionally, it's also vital for them to find insurance that covers their necessary expenses and hospitals need to ease the whole procedure for them as it can get understandably too burdening for the patient and their families

## 3. WHAT TYPES OF TREATMENT OPTIONS ARE CURRENTLY AVAILABLE FOR THALASSEMIA PATIENTS?

Depends on the severity. In short words, each patient is their own individual case. It is encouraged for Thalassemia patients to visit a Thalassemia specialist annually through contacting their local Thalassemia center for more info.
Thalassemia minor patients often require little to no extra intervention to their medical care to Thalassemia major patients that would need long visits to receive blood transfusion every few weeks. Major patients also need annual MRI to detect iron overload, ECG, and visits to endocrinologists and cardiologists. They receive daily iron chelating agents to keep their iron levels controlled.
As for a definitive cure, bone marrow transplant is the most common approach with high success if the donors are the patient's relatives, and promising research using gene therapy is currently under clinical trials.

## 4. CAN YOU PREVENT THALASSEMIA?

Unfortunately, if you are born with Thalassemia then you will be affected by it either directly or by being a carrier to the gene. Genetic counselling can help detect the possibility of carrying Thalassemia in a new couple, so awareness is the most effective method of prevention.

## 5. ANY ADVICE TO THE COMMUNITY REGARDING THALASSEMIA PATIENTS?

Always offer an aiding hand when possible, like providing them small services such as car rides and accustoming to their own scheduling, and provide them with the support and comfort anytime they open up about the difficulties of their blood disorder. Be understanding when they miss certain events as their treatment has to be frequent.

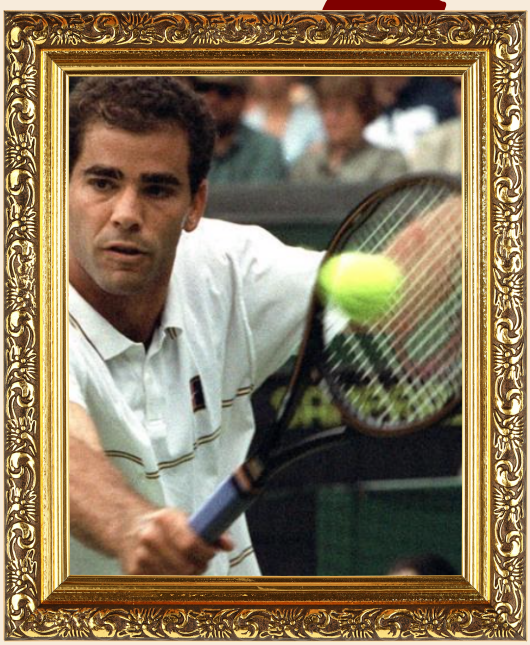
PUBLIC FIGURES:

# AMBASSADORS OF CHANGE

SARRA MKADMI

## PETROS SAMPRAS

Petros Sampras, or more famously known by his nickname Pete, is an American former professional tennis player. His career began from 1988 and ended at 2002 and throughout his profession; Sampras has won 14 Grand Slam singles, which was a record during his time. He also accomplished dominating 7 Wimbledon, and 2 Australian Opens, and overall, won 64 singles in total. It was a complete surprise when Sampras revealed to news reporters that he was in fact dealing with a medical condition: thalassemia minor. Although thalassemia minor is not a serious affliction, especially to individuals leading sedentary life styles, Sampras is a world champion athlete who exerts his body to extremes. While it was reported that Sampras collapsed from his exertions periodically, he did not let thalassemia stop him from earning a world ranking spot in the turf of the best athletes.



## ZINEDINE ZIDANE

Zinedine Yazid Zidane is an Algerian French former professional soccer player who played the position of the attacking midfielder for France. He was an outstanding player and won many individual honorary awards including FIFA World Player of the Year in 1998, 2000, and 2003! Nowadays, he coaches Real Madrid and is considered to be one of the most successful coaches in the world. Although Zidane's incredible technique and form of play is all common knowledge, especially to sport fanatics, not many know that he was a born with a genetic malady: thalassemia minor. Regardless, Zidane managed to overcome his medical condition and lead a healthy lifestyle.

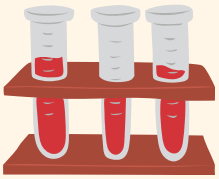
## AMITABH BACHCHAN

Amitabh Bachchan is an Indian Bollywood actor, film producer, TV host, singer and even a former politician. He is held as one of the most prominent and renowned celebrity in the Indian cinema-ology. Some of his famous work include hosting "Who Wants to be a Millionaire?", starring in Indian films such as "Zanjeer", and "Deewaar", and even guest starring in the American movie "The Great Gatsby".

As thalassemia is most commonly found in south East Asia, with every 1 out of 8 of thalassemia carriers being in India, it is of no surprise that Bachchan is a thalassemia minor person. Junior Bachchan, Amitabh's son, participated along with other Bollywood stars in spreading awareness on World Thalassemia Day. Celebrities would post a picture depicting half of their faces to social media. This urges their audience to test for thalassemia to prevent their children from "half-life". Bachchan leads a regular life style regardless of his genetic disorder.

SCIENCE SPEAKS

MEDICAL RESEARCH



MOHAMAD MONIF ASSKER

# A SEA OF BLOOD

Is what the Greek words "Thalassa" and "Haema" stand for. The reasoning behind such nomenclature is the description of the very first cases of such a condition, in populations living near the Mediterranean Sea. Despite the prevalence of this condition in other locations as well, [Africa, Asia, etc..] this name stuck on.

Another sea to mention, is the sea of different clinical manifestations, ranging from a barely noticeable abnormality that might show on an occasional blood test, to a severe & debilitating (possibly fatal) anemia. This variation in severity is due to the genetic nature of Thalassemia. Normal human Red Blood Cells shuttle oxygen through our bodies using Hemoglobin, a tiny molecule made of 4 protein chains, 2 of the type alpha, and 2 beta. In some families, genes responsible for making healthy functional hemoglobin chain types undergo mutations, and the severity of the disease increases the higher the number of mutated genes are. Since we have 2 types of chains, 2 types of thalassemia subsequently arise:

- Alpha thalassemia: where the alpha chains are deficient, and the beta chains are overproduced in order to compensate, however these chains bind oxygen poorly and result in depravation of body tissue of oxygen, termed hypoxemia.
  - Mostly because of a deletion of HBA1 and/or HBA2 genes.

- Beta thalassemia: where the beta chains are deficient, and the alpha chains are overproduced to compensate, which accumulate and damage the red cell causing it to die, and causing the spleen to work extra hard to remove those dead cells causing it to enlarge.
  - Mostly because of promoter region mutation of HBB gene, causing a reduction instead of deletion of the chain and therefore a generally milder form of disease.
  - Over 200 types of mutations in this type lead to high variation in presentation.

Each subtype can be either major, with complete absence of chains, or minor, with only a reduction in chain production.

## WHAT'S AT HAND

Currently, severe thalassemia is managed by frequent blood transfusions. Constant screening of Hepatitis B virus and blood typing are needed to ensure the recipients' safety. While milder forms are treated by targeting the symptoms and correcting anemias rather than replacing blood altogether. Both types of management however come with a risk of having iron overload (associated with severe risks of its own) that may be opposed by giving iron chelating agents [mainly desferrioxamine B [DFO]).

Bone marrow transplant is a procedure that saves the patient most of the hassle of transfusion-chelation treatment modality. It is a potential cure to some as the deficient factory of blood cells is replaced altogether. However, it comes with marked risks on its own. As many bodies tend to refuse transplants, only the luckiest of recipients are to benefit from a successful match with their donors without sustaining a rejection of the new marrow.

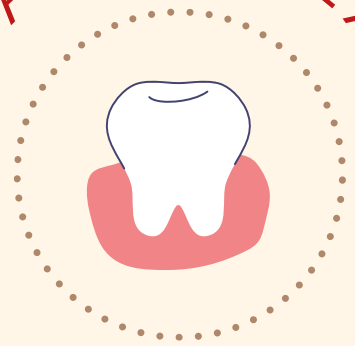
## THE UP & COMING

Two additional -and possible alternative- iron chelating agents have emerged recently, Deferiprone & Deferasirox. Both of which appear to have markedly less side effects on the heart & liver compared to DFO. Some strategies suggest combining either with DFO for even more pronounced results, but that's to be determined with further testing.

Signs of iron overload are also measured in a much safer way now, using a magnetic resonance imaging technique termed Magnetic biosusceptometry (SQUID). This is a markedly less invasive method of determining liver iron instead of the old-fashioned way of taking a liver biopsy to the microscope.

Induction of fetal hemoglobin (HbF) is also suggested to replace some of the less efficient thalassemic Hb, and therefor a target in reducing it's severity. Which works by inducing the genes responsible for manufacturing HbF instead of the thalassemic Hb, thereby compensating for what is lost.

# SCIENCE SPEAKS
## DENTAL RESEARCH



HIBATALLAH SALEM

Thalassemia refers to a group of inherited blood related defects in the production of either α or β polypeptide chains of the globin portion of the hemoglobin molecule, which makes for the 2 main types of thalassemia. It is characterized by hypochromic (pale), hemolytic (blood cell destruction) anemia of varying degrees. Based on their clinical and genetic orders, thalassemia is classified mainly into major and minor types. β - thalassemia major, also known as Cooley's Anemia, exhibits the most severe changes in cranio-facial and dental anatomy, while other types vary from mild symptoms to being generally asymptomatic.

Due to the chronic anemia, bone marrow spaces are enlarged in the facial skeleton to compensate for the blood loss. The maxilla grows exponentially larger and more protruded than the mandible, maxillary sinuses are obliterated, the upper lip is retracted, along with depressions being present in the cranial vault with frontal bossing; giving the face a chipmunk-like appearance.

Dentally; anterior open bite, increased overbite and crossbite are common occlusal discrepancies. Furthermore, studies have shown that people with β-thalassemia major have higher risk of developing rampant caries and gingival inflammation. Other studies attribute the increase in caries incidence to the skeletal malformations majorly, and partly to the impaired immune response present in those individuals.

Dental management for these individuals starts from the earliest years through frequent blood transfusions and iron chelation therapy. Studies have shown a strong correlation between the display of symptoms and age when starting treatment. Additionally, regular dental check-ups and maintenance are needed to reduce the risk of dental caries and gingival disease development. Orthodontic treatment with a specialist may be required, depending on the patient's conditions.

Finally, a change in the current statistics may be seen in the near future, due to increased awareness and support for oral health programs and increased diligence in parents and guardians compared to earlier years.

# MY ROLE AS A MEMBER OF THE COMMUNITY

ALAA AL HAJJI

**THALASSEMIA IS A MAJOR PUBLIC HEALTH ISSUE IN THE UAE. LATEST STUDIES FOUND OUT THAT ONE IN 12 PERSONS SAID TO BE THALASSEMIA CARRIER IN THE UAE. THEREFORE, AS MEMBERS OF THIS COMMUNITY, WE CAN HELP THESE PEOPLE IN DIFFERENT WAYS. HERE ARE SOME TIPS THAT YOU CAN FOLLOW TO OFFER A HAND FOR THALASSEMIA PATIENTS:**

## DONATE BLOOD REGULARLY.

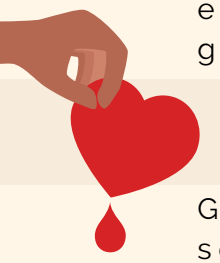
UAE is rich with blood donation centers, so you can easily access them. If possible, try to visit the nearest blood donation center every now and then.

## RAISE AWARENESS ABOUT THALASSEMIA ON SOCIAL MEDIA.

Nowadays, your voice can spread far through platforms such as Instagram and Facebook…. So, if you come across any article or video that talks about thalassemia you can easily share it with your friends. However, make sure that the information is accurate and correct!

## SUPPORT AND MOTIVATE THEM.

You can support thalassemia patients in different ways. For instance, you an offer them a ride to the transfusion center, encourage them to follow their chelation schedule and attend group therapy when relevant.



## CARE FOR THEM

Give them space so they can follow their blood transfusion schedule without making them feel disabled or embarrassed. For example, allow them to have sick leaves without a struggle.

# MYTH VS FACT

AYAH DIB

**WE RECEIVE ENORMOUS INFORMATION EVERYDAY, SOME OF WHICH ARE MYTHS. WHEN IT COMES TO DISEASES, WE SHOULD KNOW WHAT'S TRUE AND WHAT'S NOT. THIS IS YOUR OPPORTUNITY TO LOOK AT SOME MYTHS VS FACTS!**

Myth: partners who are both thalassemia carriers will always conceive children with thalassemia major.

Fact: if both partners are carriers there is only 25% chance of having a thalassemia major child.

Myth: Thalassemia is an inherited disorder that can't be avoided or prevented.

Fact: actually Thalassemia can be 100% prevented! And this is easily done by pre-marital screening.

Myth: There is no available, effective treatment for Thalassemia.

Fact: Thalassemia can be successfully and effectively managed with proper medical treatment that includes adequate blood transfusions and iron chelation therapy.

Myth: Thalassemia is incurable.

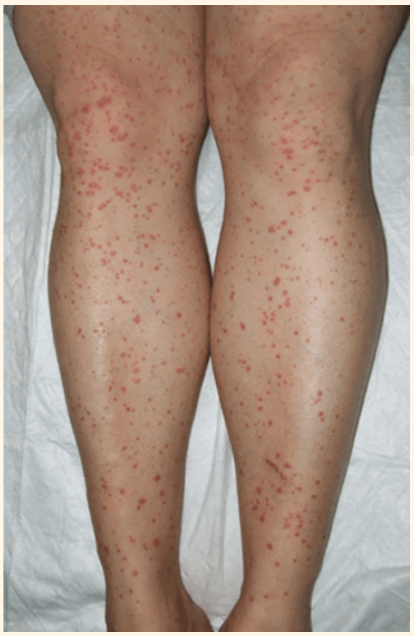
Fact: bone marrow transplantation is considered as an effective cure for Thalassemia. however, it carries some complications and health risks, and it requires a genetically matched doner.

# RARE DISORDERS

KHADIJAH ZAIDAN

## What are the platelets?

Platelets are an important component of the blood; it helps in clotting and controlling the bleeding. Let's say you got a papercut on your finger, soon, it stopped bleeding. Why and how did that happen? The process starts with your blood vessels sending signals to the platelets. The platelets will then hurry and come to the injury site and stick to it; forming a clot. Platelets will also send chemicals; attracting more of its kind. However, these platelets have an important structure on their surfaces that plays a major role in the regulation of its adhesion and aggregation, this glycoprotein is called (GPIIb/IIIa).



purpura on the lower extremities.

## What is immune thrombocytopenia?

It is a blood disorder with a low platelet count (less than 150,000 mcl). ITP is an autoimmune disorder where your immune system forms antibodies that attack the GPIIb/IIIa. Thus, leading to the degradation of the platelets. In some rare cases, the T-cells will attack the platelets. Furthermore, it could present as primary ITP which is idiopathic or "of unknown cause". Or it could be secondary ITP, which is associated with other diseases such as, systemic lupus erythematous, chronic leukemia, or some viruses such as (HIV, HCV).

**Types:**

**1.Acute**

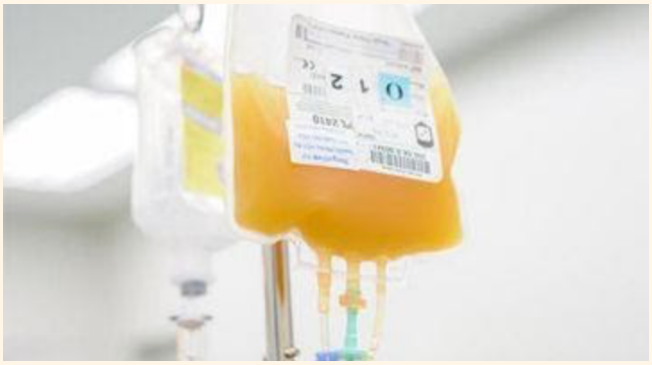
Affects children (2-6 years old)

Often follows a viral infection (other than HIV or HCV)

**2. Chronic**

Affects adults (20-50 years old)

Relapsing course



PLATELET TRANSFUSION

## Clinical presentation

The chief complaint is usually a petechial rash in the lower extremities (non-palpable purpura). It could also be associated with nasal bleeding, oral bleeding, stomach and intestinal bleeding, as well as heavy menstrual bleeding in females. If left untreated in elderly patients, hemorrhages within the brain and spinal cord might occur. Surprisingly, some patients live with the disease for too many years without having any symptoms.

## Management

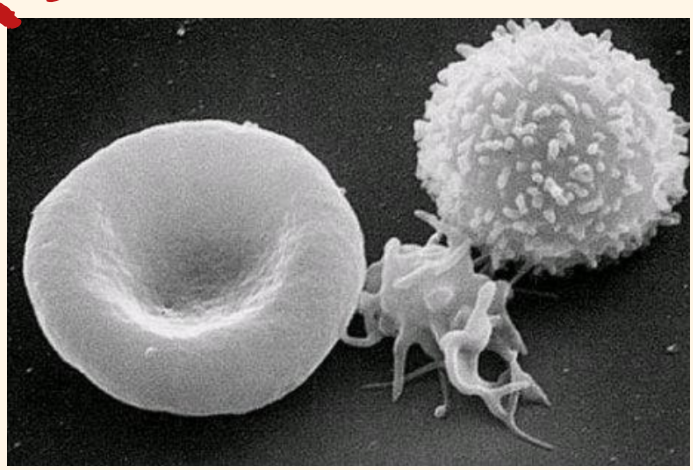
If there is no symptoms, no treatment is required, but the doctors will watchfully wait for any sign of progression.

If symptoms occur, steroid or intravenous immunoglobulin is required.

If the platelet count is very low, the solution is platelet transfusion.

Rituximab is a chimeric monoclonal antibody targeted against CD20. CD20 is a surface antigen presented on B-cells that produces the autoantibodies. Thus, Rituximab is going to act against CD-20, hence, leading to less formation of these harmful autoantibodies.

In severe uncontrollable bleeding, the removal of the spleen (Splenectomy) could be required.

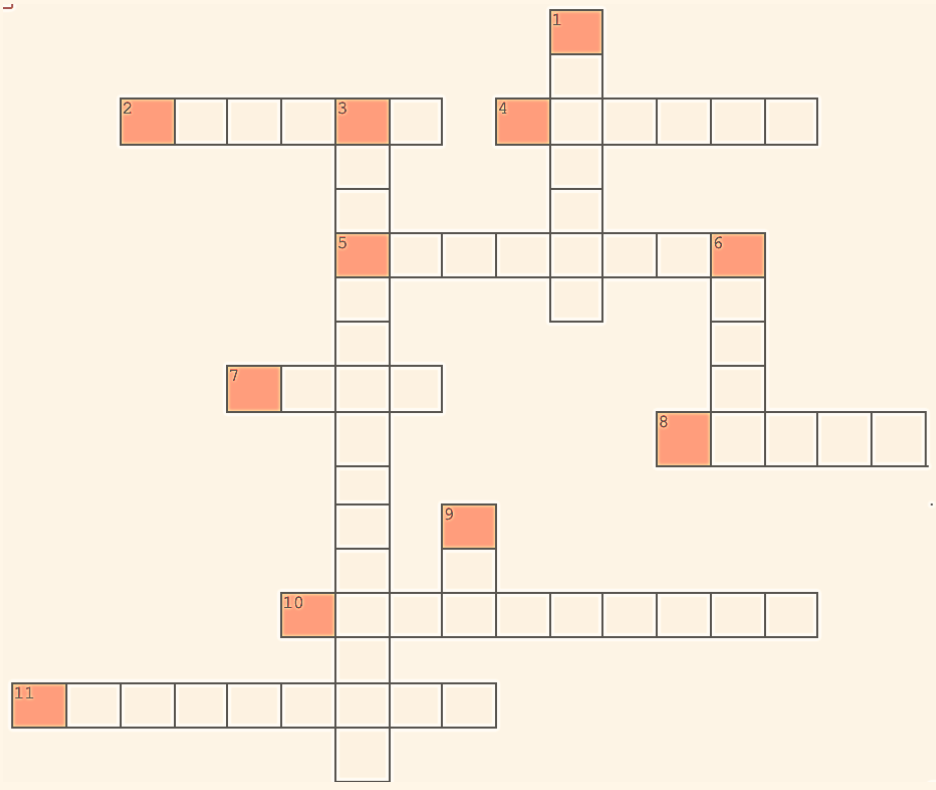


ELECTRON MICROSCOPY, FROM LEFT TO RIGHT:
RED BLOOD CELL, PLATELET, WHITE BLOOD CELL

# THE ARCADE

AYAH DIB

**WANT TO TEST YOUR CROSSWORD SKILLS? BELOW ARE STATEMENTS REGARDING DOWN SYNDROME, COMPLETE THE BLANKS THEN WRITE THE LETTERS IN THE PUZZLE HORIZONTALLY OR VERTICALLY ACCORDING TO THE NUMBERS SHOWN.**




## ACROSS

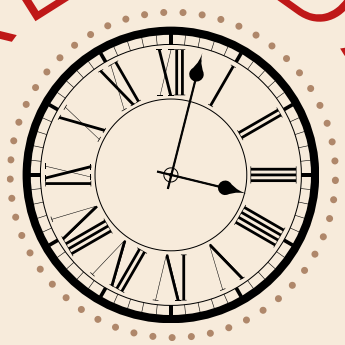
2. Beta thalassemia is also called anemia
4. Thalassemia leads to excessive destruction of red blood cells which leads to.....
5. People with thalassemia trait in one gene are called.....
7 overload is a complication of thalassemia treatment.
8. one type of thalassemia is hemoglobin H disease that is caused by inactivation of three chain genes.
10. Thalassemia is caused by abnormal ..... production.
11. Thalassemia is inherited in an autosomal manner.

## DOWN

1. Thalassemia is a blood disorder
3. Thalassemia can be diagnosed using hemoglobin .....
6. Red blood cells of thalassemia patients are .....
9. Children with moderate to severe thalassemia are usually diagnosed by age.....

Answer key in The Appendix.

# THE AADMD CALENDAR



ALAA AL HAJJI

- GROUP B STREP THROAT AWARENESS MONTH
- JUVENILE ARTHRITIS AWARENESS MONTH


LEIOMYOSARCOMA AWARENESS DAY


WORLD HEPATITIS DAY

# THE APPENDIX

SOLUTION OF THE ARCADE:

1. GENETIC
2. COOLEY
3. ELECTROPHORESIS
4. ANEMIA
5. CARRIERS
6. SMALL
7. IRON
8. ALPHA
9. TWO
10. HEMOGLOBIN
11. RECESSIVE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