KBG Syndrome

quick facts

1. In 1975 the first description of KBG Syndrome was established.

2. The name KBG Syndrome is derived from KBG identifier Dr. Optiz's tradition of using the surname initials of the families first diagnosed.

3. KBG Syndrome is an ULTRA Rare condition, with less than 200 people diagnosed worldwide as of 2017

Newly diagnosed with KBG Syndrome? There are a few things you should know.

"The Journey of a thousand miles begins with one step." - *Lao Tzu*

You or your child have taken that first step and been diagnosed with KBG Syndrome. ***Now what?***

For many, their KBG diagnosis is the culmination of years of wondering: *What is different about my child?*

For others, there was a nearly immediate understanding that 'something was amiss' and the search for answers began.

In each instance, the reaction to the diagnosis is different even though the diagnosis IS the same. One will breathe a sigh of relief that they finally have an answer and then, they will wonder, ***what is next?***

For the other, they will be shocked and saddened to suddenly be thrust into a world of medical terminology and specialists. Yet again, they will wonder, ***what is next?***

KBG syndrome does not come with a set diagram of issues, or a clear-cut treatment routine, it's a treat-as-needed syndrome. Where should you start?

"I cried for joy before I knew better, then I cried for help"

"The first step toward change is awareness. The second step is acceptance." –
Nathaniel Branden

Start with you!
You've learned something that will live with you the rest of your days. How do you deal with that?

Allow yourself time to adjust to your new reality and accept that your journey has just changed course. How much of a course change has a lot to do with whether you choose to be the passenger or the driver.

You can choose to sit still and let others decide the direction your health care will take, or you can put the time in to learn how to guide the course and get behind the wheel.

Grieve?
In many ways what you are experiencing now is very close to the grieving process. Allow it to play out and give yourself a chance to adjust. You need to know that it is a natural response and it is okay to grieve.

The stages of grief include:

Stage 1: Denial
Stage 2: Anger
Stage 3: Bargaining
Stage 4: Depression
Stage 5: Acceptance

Your grief will be centered in what was, what may have been, what can never be and what unknown lies ahead. What IS known: you are not the only person in world with a KBG Diagnosis.

Remember: You are not alone
At some point, after the initial diagnosis has been made and the shock wears off, you may feel overwhelmed. It's completely understandable; there is a lot to think about. You may feel lost but you need to know that others have been through this before.

What they have experienced is not exactly YOUR experience but they also have been diagnosed with KBG Syndrome and they know what that means. They also know how important it is to support others just starting on their own journey.

Diagnosed
give yourself time

What is next?

Diagnosed
believe in yourself

No one knows you or your child like you do. NO ONE.
You will probably begin to meet with many Doctors all with an intimidating amount of letters after their name, but keep in mind: **they still do not know you or your child.** Don't get scared or intimidated, most likely, they are hearing KBG Syndrome for the first time as well.

Don't be surprised if your Doctor does not automatically understand what KBG is, or how to treat it. It **IS** an ultra rare diagnosis. He or she may not grasp immediately what you need for your treatment of the symptoms you are experiencing, or even if any treatment is needed. Your Doctor is probably researching along side you; it can be a very frustrating time for everyone, not just the newly diagnosed patient.

KBG syndrome does not come with a set diagram of issues, or a clear-cut treatment routine, it's a treat-as-needed syndrome. What does that mean? It means if you start to have trouble hearing, you should go talk to a Doctor. You MAY now be showing the signs of hearing loss.

'Treat as symptoms arise' is as great as is it infuriating. It's frustrating, because there is not any one pill, or any one combination of treatments to treat the syndrome itself. It's great because each patient can be treated individually based on his or her symptoms.

So, how does one cope, knowing that one must always be on their toes, always looking for something to arise, or nothing to arise, just always looking?

Bolster your knowledge
Learn all you can about KBG Syndrome and what it means to have a rare disease.

Over the last 40 years, research into KBG Syndrome has been slow-going, and focused mainly on how to properly diagnose the Syndrome. We have gathered all published and pertinent research papers into one area of the Foundation website: **www.kbgfoundation.com/studies.html.**

The latest advancements in actual treatments has been defined by the KBG Families themselves utilizing the Private Facebook Group created by the Foundation.

You can have all the tools in the world but if you don't genuinely believe in yourself, it's useless.
- Ken Jeong

Diagnosed
find others

Growth is never by mere chance; it is the result of forces working together.
- James Cash Penney

Find others!
None of us should walk this path alone, not when there are others willing to lend emotional and physical support. The largest concentration of KBG families has gathered in the aforementioned private Facebook Group. The group is open to all patients, caregivers, doctors, researchers, educators and anyone with a shared interest of helping the KBG patient population. The group can be found by visiting the KBG Foundation Page and requesting approval to join.

Or type this link into your browser bar and send a request to join the group: **facebook.com/groups/KBGPatientGroup/**

Part of taking the time to get your feet under you is learning as much as possible about what this diagnosis may mean to your life. Your journey is your own, but the people in the group have walked the path and are willing to help guide your steps.

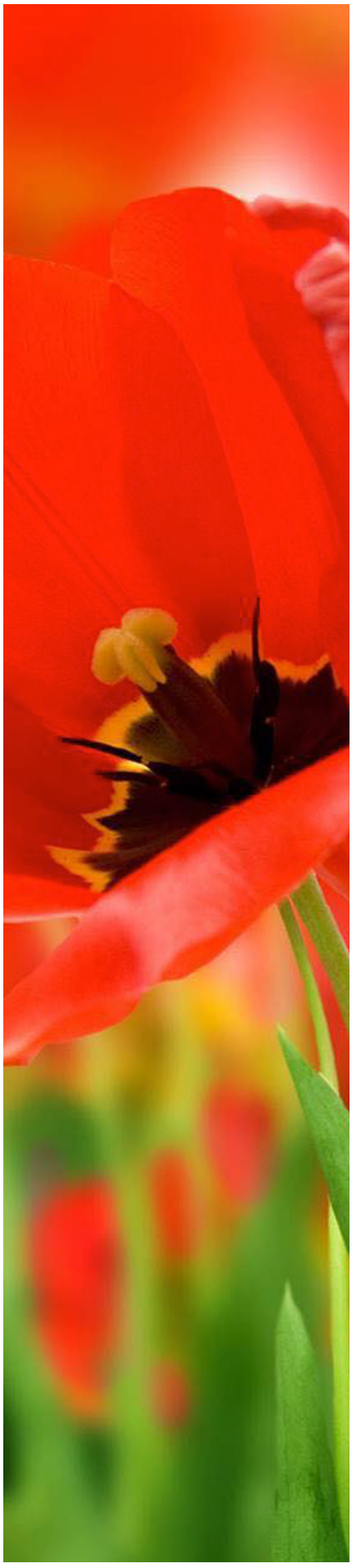
Working with this group doesn't just provide support but it also helps identify common traits and symptoms that can be used to develop treatments. Sharing information with others expands what we know and how we better serve the KBG population.

It takes time AND it takes a village.
We've gone from 60 known case to over 200 just by sharing! The more patients we have, the greater the chance of developing comprehensive treatments and maybe one day, a cure.

Until that time, detection and prevention are the mantra of the KBG Patient. Detection at the earliest possible time of the symptom and prevention of worsening.

To help aid the effort to stay healthy and intact, we have created a list of specialists that MAY need to be visited and a brief description of why and what follow up may be needed.

This list was compiled using data from the KBG Families and the NORD website.



What Specialists may you want to/need to visit?

Not all of the specialists on this list are needed for every KBG patient but when possible, a baseline should be set to determine if a symptom is getting worse or even if it is starting to develop.

Audiologist / ENT - Hearing issues are quite common in KBG and should always be examined. Look for inner ear malformations and persistent ear infections. Related to: Speech Therapy, Corrective Surgery

Craniofacial - Will help determine the need for intervention for the cranial manifestation of KBG Syndrome. Refers to: Orthodontist, Cosmetic Surgeon, Dentists, ENT, Neurosurgeon and others.

Cardiologist - KBG is associated with several types of heart conditions and should be examined. Oft times an early diagnosis of KBG is made because of newborn cardiac, pulmonology or gastrointestinal issues.

Dentist - Macrodontia (large teeth) are the tip of that iceberg - other dental issues have been reported and can be repaired. Refers to: Orthodontist or Oral Surgeon

Endocrinologist – Short stature is somewhat common in KBG patients and families have opted for growth hormone therapies. This specialist can help guide your decision. Also, precocious (early) puberty has been noted and may need to be addressed.

Epileptologist – This is a Neurologist who specializes in seizures/epilepsy. KBG is associated with abnormal brain activity but not always seizures. To know for sure, a baseline EEG should be set.

Gastroenterologist - Reflux is another common in KBG as well as anomalies of the esophagus, stomach and upper intestine.

Geneticist - Will provide genetic confirmation of KBG and look into additional genes but most likely will provide the diagnosis and may not schedule a follow up. Refers to: Other specialists as needed.

Hematology – A few KBG patients and their families have been diagnosed with bleeding disorders. No link has been identified but it may be beneficial to be tested.

Immunologist /Allergist / Rheumatologist - KBG families are reporting immune system function issues such as frequent or intense illnesses, common and frequent infections as well as severe seasonal allergies, this Doctor can help guide that investigation and may refer you to an Allergist or Rheumatologist.

Neurodevelopmental - This Doctor will examine the development of the central nervous system and the brain and how emotional responses correlate and cognition progresses. Refers to: Neurosurgeon and for therapies.

Neurologist - An EEG should be ordered, as well as an MRI to check for structural issues in the brain and to set a baseline for brainwave patterns. Related to: Neurosurgeon, Neurodevelopmental, Neuropsychology, Occupational, Physical and Speech Therapies.

Neuropsychology / Psychology /Psychiatry- These Doctors can help with behavioral and cognitive issues seen in a fair amount of KBG patients. Refers for therapies and can prescribe medications.

Orthopedist - Bone anomalies are also common in KBG - check for hip, spine and feet issues as well as Kyphosis (a hunched-back). May refer you for physical therapy.

Ophthalmologist - A few KBG patients have reported problems with eyesight, may not be more than the average population but strabismus is common and should be followed.

Pediatrician OR **General Practitioner** - Use your Doctor to refer you to specialists as needed. Refers to: Any other specialist as needed.

Pulmonologist - May be needed for aspiration issues and to check for other lung issues that may be present.

Urology / Nephrology – Several KBG patients experience bladder and kidney problems including malformations, and chronic infections.

Accommodated Education - If your child is NOT receiving accommodations in school, take the time to learn how to become successful in getting the education he or she needs in the manner your child learns best. KBG patients may have cognitive delays but they are JUST delays, the sooner they get the assistance they need, the better the outcome.

KBG SYNDROME FOUNDATION

#everylinkmatters

KBG Foundation

www.kbgfoundation.com
Facebook: KGBFdn
Twitter: @KBGfdn

Why is the list so long?
KBG Syndrome can affect the entire body; it is a protein creation disorder. In many instances, symptoms evaded detection until a parent or caregiver read about the cause of their similar or identical symptom in the KBG Private Group and asked their Doctor to investigate.

We have discovered more about KBG Syndrome as parents and affected individuals sharing experiences than we have within the medical community. Shared knowledge is the **ultimate** power.

The ultra-rare aspect of KBG Syndrome means not a lot of resources have been designated to understanding and treating it. This fact alone is the MAIN reason we encourage reaching out to others that not only understand what you are going through, but what you **may** go through.

The KBG Foundation was formed to provide support and facilitate communication between families, doctors and researchers in order to take some of the shock out of receiving the KBG Diagnosis. As parents, patients and medical professionals, we know that having a place to share your frustrations, concerns and triumphs can provide comfort and support throughout the complicated process of treating the KBG individual.

As many as have gone before us, a lot about KBG Syndrome is still unknown. We must remember that we are pioneers - paving the way for generations to follow.

The more we share, the more we learn, the more likely a viable treatment and, eventually, a cure can be found.

We REALLY are in this together!

Welcome to the family!