KBG Syndrome

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

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.

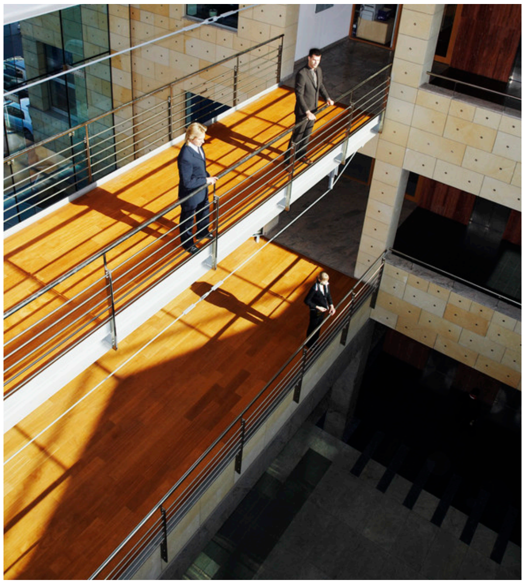
In each instance, the reaction to the diagnosis is different even though the diagnosis IS the same. One will breathe a sigh of relief that they finally have an answer and then, they will wonder, ***what is next?***

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?***

quick facts

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

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



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

Start with you!
Allow yourself 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.

In many ways what you are experiencing now is very close to the grieving process. You need to know that it is a natural response and it is okay to grieve. The stages 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.

You are not alone
At some point, after the initial diagnosis has been made and the shock wears off, you may feel overwhelmed. 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.

Let them help!
None of us need 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 a private Facebook Group that can be found by visiting the KBG Foundation Page and requesting approval to join.

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.

What is next?

Diagnosed

believe in yourself

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

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.** Most likely, they are hearing KBG Syndrome for the first time as well and will be researching along side you. They will not automatically understand what KBG is, or how to treat it, nor will they grasp immediately what you need for your treatment of the symptoms you are experiencing, or even if any treatment is needed. It can be a very frustrating time.

KBG syndrome does not come with a set diagram of issues, or a clear-cut treatment routine, it’s a treat-as-needed syndrome, which is as great as is it infuriating. Great because each patient will be treated individually with an excellent prognosis overall. Frustrating, because there is not any one pill, or any one combination of treatments that can be ordered to treat the condition itself. 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?

Diagnosed

find others

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

We have compiled a list of specialists you may need to visit.

Not all 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 also very 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 also 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.

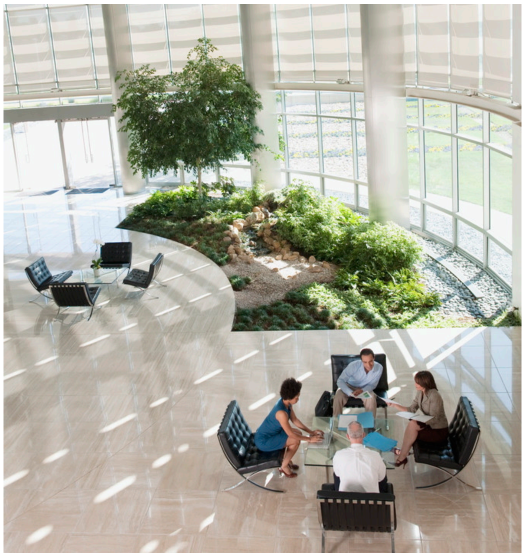
Orthopedist - Bone anomalies are also common in KBG - check for hip, spine and feet issues as well as Kyphosis (a hunched-back). May refer you for physical therapy.

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.



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