UNIQUE

UNDERSTANDING GENES & CHROMOSOMES

ANKRD11 and KBG Syndrome in adults

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This guide is designed to help individuals with KBG syndrome, their families, and healthcare and other professionals involved in their care. It contains information about the cause, the ways in which it can affect people into adulthood and suggestions about the healthcare and support that may benefit people with this condition. Some of the information has been taken from Unique's ANKRD11 and KBG syndrome guide, which has a focus on experiences in childhood. The guide also draws on findings from a survey in 2024 (Life beyond childhood: insight into the lived experience of 91 adults with KBG syndrome through an online patient/caregiver reported co-produced questionnaire), along with other recent research, full details of which can be found in the references section at the end of this guide.

What is KBG syndrome?

KBG syndrome was first described in 1975, and its name is derived from the initials of the first three patients reported with the condition. Most people with KBG syndrome have a characteristic (and sometimes subtle) facial appearance, very large permanent teeth, and variable degrees of intellectual (learning) disabilities and behavioural differences. Because the features can be subtle and are not always present, the diagnosis may not be made until adulthood. Some adults only receive a diagnosis after having genetic testing when their child, with more complex concerns associated with KBG syndrome, is diagnosed.

Other features seen in some affected individuals include hearing loss, undescended testes in boys, seizures, skeletal anomalies, and short stature. Most affected people are the first person in their family to carry the gene change, but a small proportion have inherited it from a parent, who is likely to have mild features of KBG syndrome. The condition affects boys and girls, and there are both mildly and more significantly affected individuals of both sexes.

Most people with KBG syndrome have:

- Mild learning (intellectual) disability; however, this can range from having no difficulties with learning to a severe learning disability, and some behavioural differences
- Large permanent upper middle teeth (macrodontia of upper central incisors)
- Characteristic facial appearance: a triangular-shaped face; wide-spaced eyes and thick eyebrows, which sometimes join in the centre (synophrys)
- Short fingers (brachydactyly) with a curved 5th finger (clinodactyly)

What causes KBG syndrome?

KBG syndrome is caused by changes in a gene called *ANKRD11,* located in chromosome 16 (band q24.3.). This may be due to a change (pathogenic variant) within the gene that disrupts its function, or to the loss (deletion) of the whole gene or part of it. We have two copies of chromosome 16 in our cells, so we also have two copies of the *ANKRD11* gene. The other copy is unaffected.

How common is KBG syndrome?

KBG syndrome is rare. To date (2025), it is known to affect more than 500 families worldwide. It is likely that there are many people with KBG syndrome who are never diagnosed because many of the features can be mild in those with a change (variant) in *ANKRD11*, including the degree of learning (intellectual) disability.

Why did this happen?

When children are conceived, their parents' genetic material (DNA) is copied in the egg and sperm that makes a new child. The biological copying method is not perfect and occasionally random, rare changes occur in the genetic code of children that are not seen in the DNA of their parents.

KBG syndrome occurs when one of these random, rare changes affects the *ANKRD11* gene in

chromosome 16. This happens naturally and is not due to the biological parents' diet, environment or lifestyle. In most people with KBG syndrome, the genetic change was a random (or "*de novo*") change, meaning the change occurred for the first time in that family in the affected individual. Occasionally, one parent may have the same change (variant) and pass it on to their child. No one should be blamed for variants in their DNA and no parent is at fault when a new DNA change occurs in their child.

Can it happen again?

There are many adults with KBG syndrome who have gone on to have children. If a parent has KBG syndrome, the chances of passing the condition on to a child are 50% (1 in 2) as the parent could either pass on their altered copy of the gene or the unaffected copy. This inheritance pattern is called autosomal dominant (because the change is on an autosomal chromosome and an outcome can be seen if only one copy of the gene is altered).

It is not possible to reliably predict the effect KBG syndrome would have on a future child's development, health and behaviour. There are many different options available if you have KBG syndrome and want to have children. Please do speak with a clinical geneticist or genetic counsellor who will help you explore your options. Each family situation is different, and a clinical geneticist or genetic counsellor can give specific advice to you and your family.

Unique publishes separate guides to Single gene disorders – autosomal dominant inheritance, Planning your next child, Prenatal genetic testing and diagnosis, A clinical genetics appointment and Supporting siblings of children with a rare genetic condition.

What is everyday life like for adults with KBG Syndrome?

KBG syndrome can affect individuals in different ways. You may be an adult recently diagnosed with KBG syndrome wondering how this might affect you, or a parent wondering how your child with KBG will get on as an adult.

In 2024, there was an online survey for adults with KBG syndrome on how this condition affects their daily life. The responses of 91 individuals were analysed by researchers. The main findings from this survey are described here, and the full details can be found in the References section (see page 15). It is important to remember that everyone with KBG syndrome is an individual and many of these findings will not apply to you. However, they may be able to help guide you when planning for the future. It is also hoped this information will help guide professionals that provide your healthcare and support, so that it is better tailored to your needs.

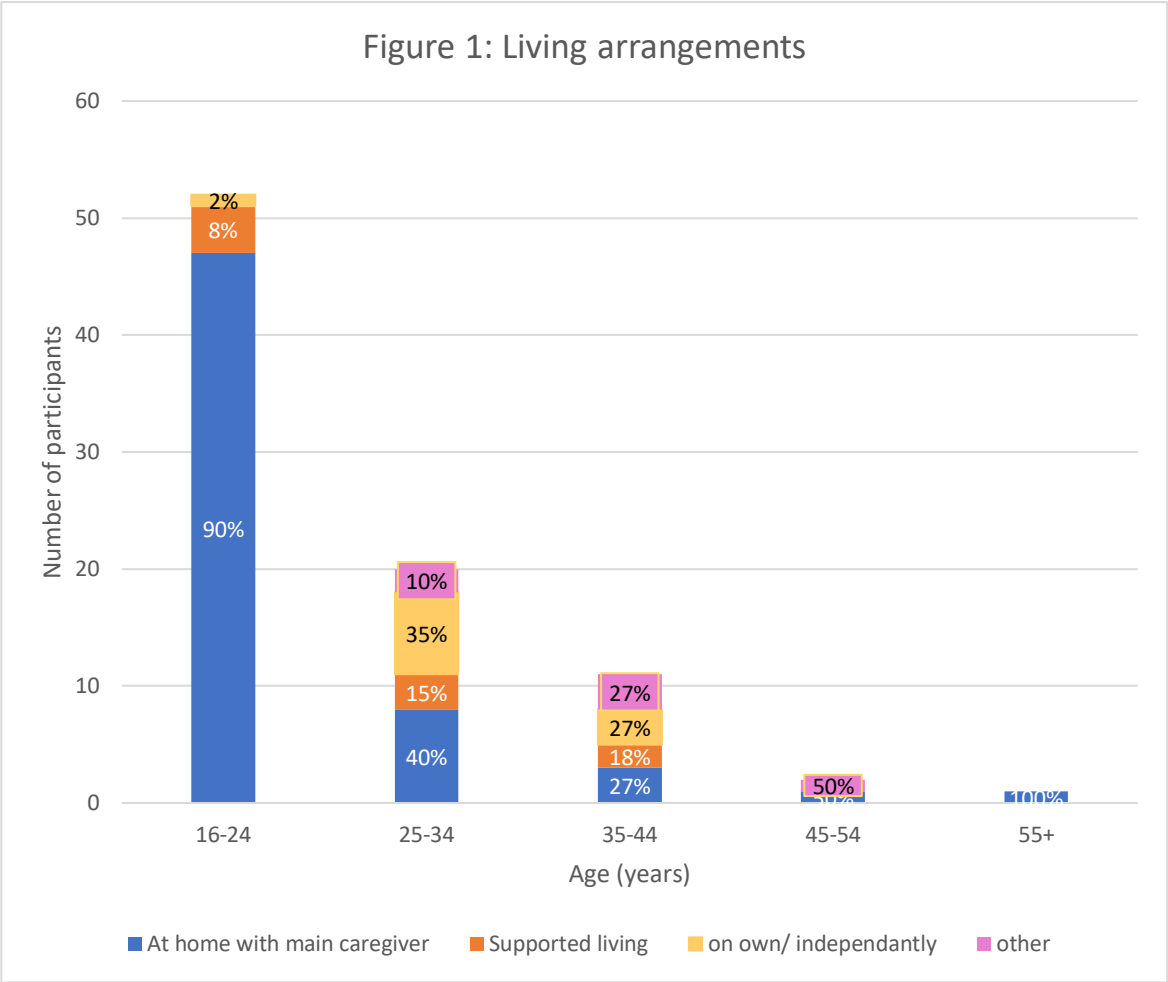
Living arrangements

Adults with KBG syndrome are able to learn different degrees of independent living skills. The level of an individual's learning (intellectual) disability from childhood will continue to affect them into adulthood. A person's level of learning disability and whether they experience seizures, along with the amount of support that is available to them, are likely to affect their ability to live independently. Some adults with KBG syndrome continue to live with their parents, while some move into residential settings where there are caregivers who can provide support. Others have gone onto have families of their own and manage their own household and work.

The 2024 survey found 66% of adults with KBG syndrome lived with their main caregivers, 12% lived independently, 10% were in supportive living, and 7% reported 'other' arrangements. A higher proportion of 16-24 year olds lived with their main caregiver (90%), compared to 25-34 year olds (40%) and 35-44 year olds (27%). The data around living arrangements is shown in the graph on the next page (Figure 1).

We know that, given the right opportunities to learn, many adults with intellectual disability will continue to develop independent living skills. It is possible for some adults to move out of their

parent's home and start living independently or in supported accommodation. However, due to the way that this survey was designed (a cross-sectional study where data is collected at a single point in time) it was not possible to give an average age at which independent living skills were achieved for these participants.



"Our 19-year-old daughter lives at home with support from myself [her mother], dad and sister."

"Our 22-year-old son is supervised at home. He has as a life plan [through a service and advocacy organization for people with all types of disabilities]."

"Our 20-year-old daughter lives with us."

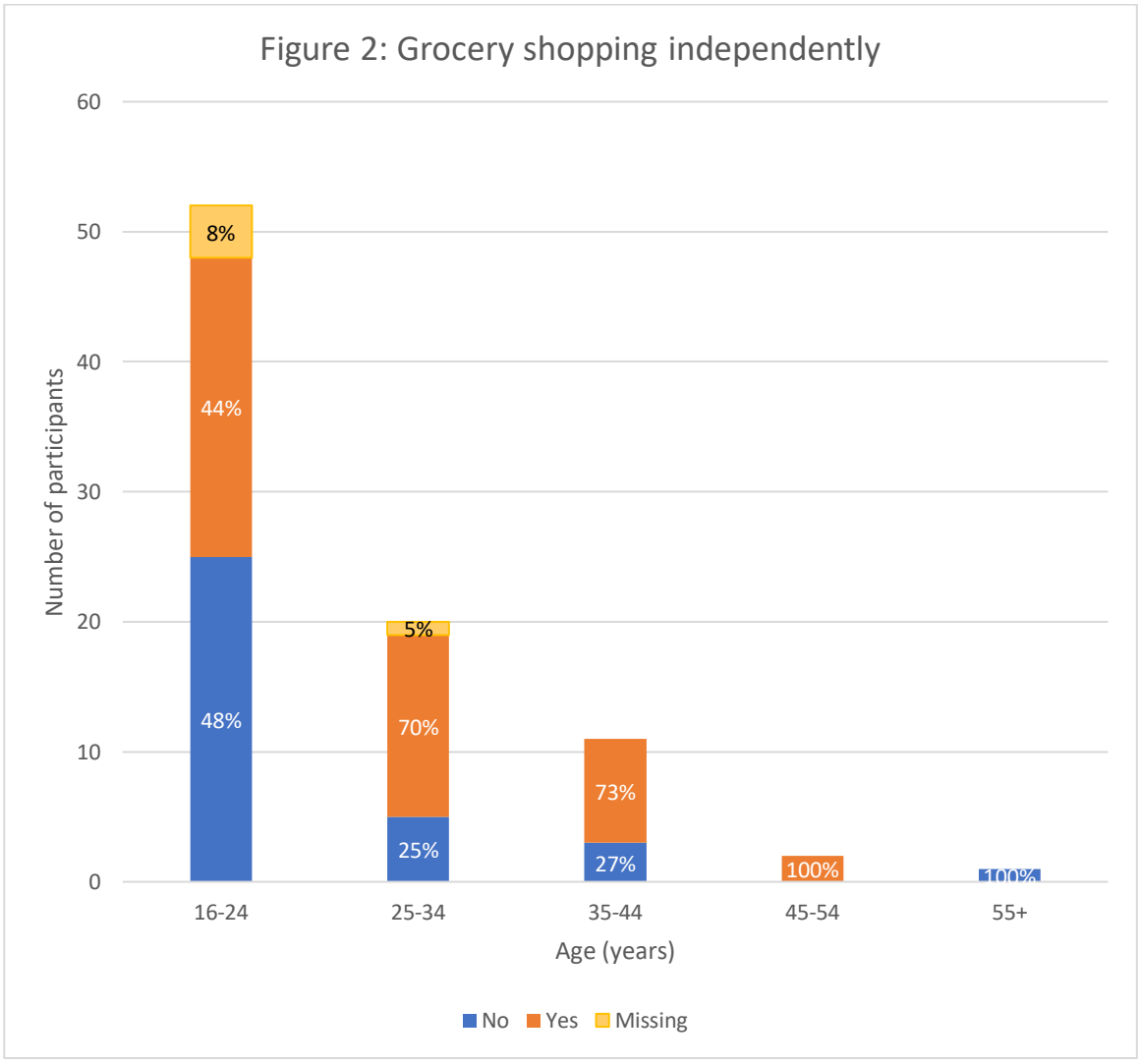
"Our 20-year-old daughter is in a specialist post-16 residential college, which is for students who have an autism diagnosis but are high functioning. She lives at home during school holidays with us and her two younger brothers. Her dyspraxia causes difficulties with simple tasks like washing and brushing her hair, getting medication out of blister packs, food preparation and opening jars etc. She struggles with shoe-laces and has poor spatial awareness so struggles to do anything at a height i.e. climbing a ladder, jumping into a pool."

Grocery shopping

Many adults with KBG syndrome are able to do their grocery shopping independently. Adults with mild to moderate learning (intellectual) disability are likely to need extra guidance and support to develop the skills needed to do grocery shopping. It is important that, if needed, additional learning opportunities tailored to an individual's specific needs are provided in adulthood, as many adults with KBG syndrome can continue to develop these skills.

The 2024 survey found that 52% of participants were able to do their grocery shopping independently and 37% reported they were not. The data on grocery shopping is shown in the graph below (Figure 2). You can see that the proportion of individuals able to do their grocery shopping independently increased with age, from 44% in the 16-24 year olds, to 70% in the 25-34 year olds, 73% in the 35-44 year olds, and 100% in the 45-54 year olds.

Only one individual over the age of 55 years responded to the survey and they were not able to shop independently. It was recognised by the researchers that far fewer older adults responded to the survey (only 2 individuals in the 45-54 years age group, and this single individual in the 55+ year group), and so those percentages may not be representative of what young adults diagnosed today will experience in the future.



"She does not do grocery shopping as she lives at home."

"She can buy simple grocery items by herself but would need support to pack and carry heavy bags if it were a large shop. Her anxiety about asking assistants for help or worry about being approached by strangers means she prefers to have support from her support workers or parents to do her shopping."

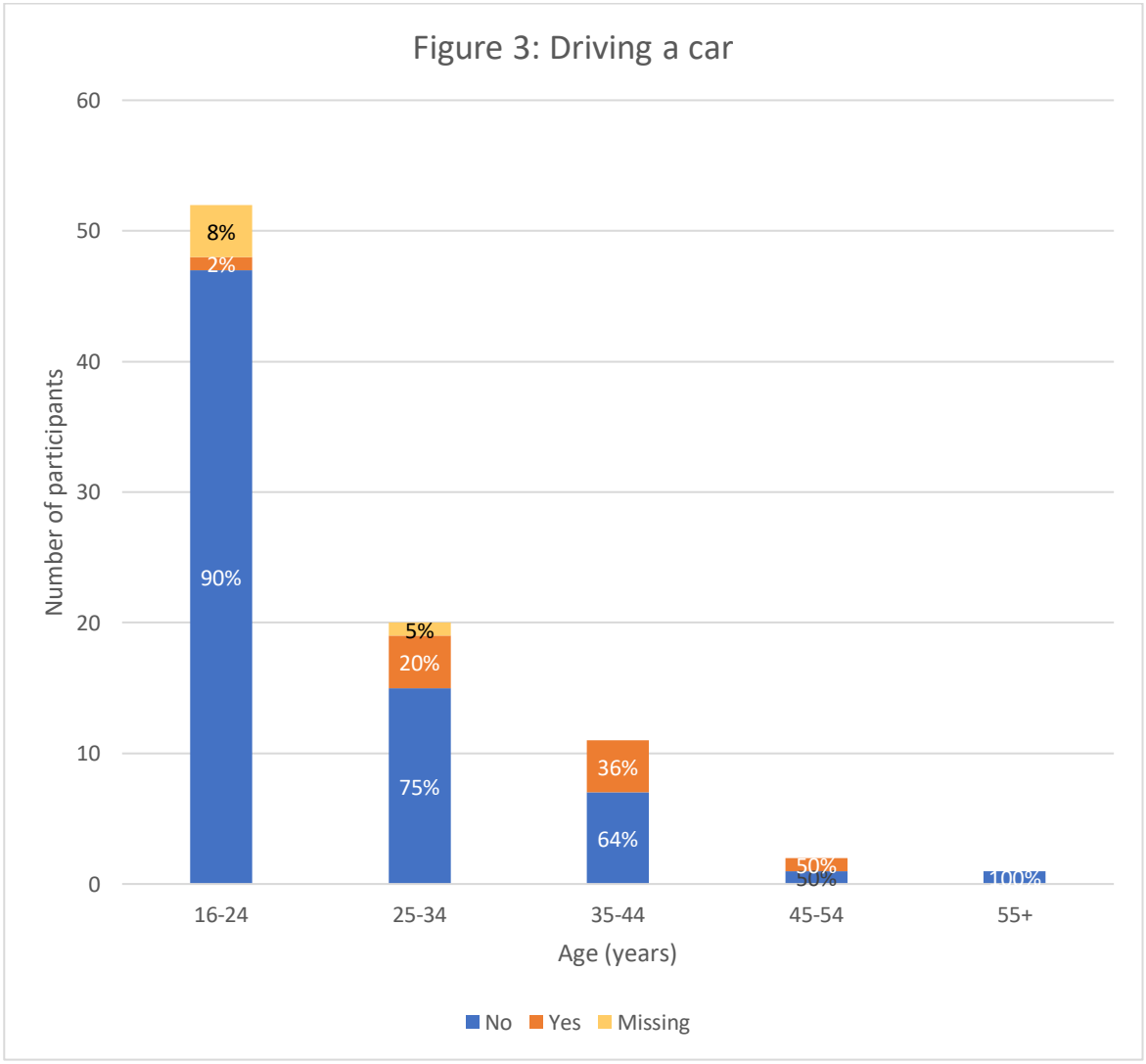
"Assisted [with grocery shopping]."

"She can buy small amounts of groceries but struggles to carry anything heavier than this."

Driving

Some adults with KBG syndrome can drive a car. The chances of being able to drive a car are likely to be influenced by an individual's level of learning (intellectual) disability and whether they experience seizures, amongst other things.

The 2024 survey found that 78% of participants could not drive a car, 11% could, and data were missing for 11%. The proportion able to drive a car increased from 2% (1 individual) in the 16-24 year olds, to 20% (4 individuals) in the 25-34 year olds, 36% (4 individuals) in the 35-44 years olds, and 50% (1 individual) in the 25-54 year olds. Again, because there were far fewer participants in the older age categories, it is important to recognise they are not likely to be representative of the experience of most young adults with KBG as they grow older.



"Not driving yet at 19 years."

"Unable to drive."

"Doesn't drive."

"She has epilepsy and suffers from occasional seizures therefore driving is contraindicated."

Education

Adults with KBG syndrome can be successful in a wide range of educational settings. Some will thrive in a special educational setting or have some modifications to the curriculum according to their learning needs. Others have undertaken vocational training and even gone on to complete higher education university degrees.

The 2024 survey found that while 12% of participants were currently enrolled in school, 25% had completed high school or college; 18% had undertaken vocational training; 1% had obtained an undergraduate degree; 1% had earned a master's degree; and 2% had not attended any educational setting. Of respondents who were attending, or had attended, an educational setting, 26% described requiring a special educational setting or having some modifications to the curriculum. These results clearly show the wide range of educational attainment that can be possible with KBG syndrome, and that the diagnosis alone should not affect someone's future aspirations.

"At 19 years, our daughter is studying at a special needs college doing catering. She gets support and is getting on well with a small group. She has selective mutism so can only communicate with family and close friends and struggles with communication with college staff and other adults."

"Our son had special educational needs and graduated with a general [high school] diploma."

"Mainstream until age 17. Will start at specialist college soon [after an education tribunal]."

"Our daughter is in a post-16 college placement. She has achieved 3 GCSEs at level 4 and 3 GCSEs at level 3. She achieved a merit in a BTEC in Performing Arts at level 2 at a mainstream college and has recently completed functional maths which is equivalent to a level 4. She is studying English functional skills and hopes to pursue a course in music next year, which she will be supported to attend by support staff at her college.

She has struggled throughout her school life to get the help she needed, and support came late as she masked many of her difficulties and as parents, our concerns were ignored. We have had to go to education tribunal twice to achieve a suitable education placement that can meet her needs. We hope that now we have the right support (or at least much better), things will improve, and the college staff will help get her to a place where she can enter the workplace in some form."

Employment

Adults with KBG syndrome are employed in a wide range of jobs. Many will require support, and/or roles adapted to their abilities and interests, to reach their full potential at work. Individuals with intellectual disability, attention-deficit/hyperactivity disorder (ADHD), autism spectrum condition (ASC) or dyslexia are entitled to Reasonable Adjustments at work under the Equality Act 2010 in the UK. These are changes an employer makes to remove or reduce disadvantage related to someone's disability. They should be specific to an individual person and tailored to their needs, for example they could take into consideration someone's sensory differences.

The 2024 survey found that one third (1 in 3) of participants were employed. They had a wide range of careers from the following categories: Hospitality and Food (6 individuals), Environment and Land (4), Retail and Sales (4), Home Services (3), Social Care (3), Teaching and Education (2), Administration (2), Animal Care (1), Creative and Media (1), Delivery and Storage (1), Healthcare (1) and Sports and Leisure (1).

The amount of support adults with KBG require to do their best at work ranges considerably. Some will not need any support at all, others may need adapted conditions, and some may need 1:1 support.

"Our son has a job coach and works 4 hours a week with the coach in a grocery store."

"Our daughter is not employed."

"Our daughter is still in education. She does work experience at a farm and in a community garden with college staff."

"Our daughter spent one year as Young Persons Champion for our local Council promoting the Local Offer page. Volunteers at local Sue Ryder shop, a local charity, and as a leader at Rainbows (Girl Guides aged 4-6 years)."

Leisure

Adults with KBG syndrome can participate in a wide range of social activities, according to their interests. The survey found that 60% (44 individuals) maintained social activities. Of these, 9 individuals attended organised social activities and outings, and 9 participated in sports - with 1 to an elite level. Six were involved in the creative arts: 4 in music, 1 in drama and 1 in dance. Nine went out with friends, 2 of them enjoyed going to the cinema and 1 had sleepovers. Two respondents enjoyed playing computer games, 3 maintained social connections through living and working with others, and 2 through religious groups. Some participants described challenges accessing social activities, such as struggling with large groups and not liking a change in routine or unpredictable events.

"Our son undertakes supervised leisure activities. He loves counting coins, music, camping and shopping."

"Our daughter doesn't have hobbies but spends time with us, going on walks, bike rides, and days out. She only socialises with her boyfriend once a week, who she trusts."

"Our daughter suffers from overwhelming anxiety which impacts on every aspect of daily living. She needs considerable support from her support workers / us to prepare her for anything new or different but with support she is able to access a range of community-based and leisure activities.

She plays the piano and sings in her local rock choir which gives her much enjoyment and sense of community. Music has always been her go to in terms of therapy and she is most happy when singing or listening to music. Feeling part of something where she is able to contribute and not feel different has been invaluable. She attends a tap-dancing class, enjoys classes at her local gym and likes to attend social activities at her college such as quiz nights (she has a great memory for facts, particularly music facts and is a great asset in any pub quiz). She is a member of her church and enjoys spending time outdoors with her dogs and at the farm where she volunteers."

"Likes to walk with the dog and help in the garden. She attends an inclusive karate class, watches Netflix and Asian dramas and likes to be taken out to eat."

Relationships

Many adults with KBG syndrome have long-term relationships and have gone on to have children. Others have not been in romantic relationships, some of whom are not interested in having one. The desire to have a relationship and a family is of course a personal choice and different people will have different feelings about this. It is important that assumptions are not made about someone's thoughts and feelings just because they have a learning (intellectual) disability, and that support navigating relationships and sex education is provided that is tailored to an individual's level of understanding and needs. For people looking to have children, a clinical geneticist or genetics counsellor can help explore which options are right for them.

In the 2024 survey, 11% participants had children, 13% were sexually active, and 12% currently

had a long-term partner or were married. The majority of participants (81%) had never been in a romantic relationship. Of these, 34% expressed a desire for a relationship, while 34% stated they were not interested in romantic relationships.

"Our daughter has anxiety, likes a routine, and struggles with communication. She only takes to trusted adults and her close friends. She currently has a boyfriend who she has known for 7 years, from school and college, and she sees him once a week. She has one or two friends at college."

"Our daughter needs support with social and communication skills and struggles to form and maintain relationships. She has regular input from her speech and language therapist at college to support with this. She is beginning to develop some friendships at college with support from the college staff. She is very attached to her parents whom she seeks constant reassurance from as she struggles to communicate her needs to support staff."

"Our daughter struggles to read social situations and make and keep friends. She has not been formerly diagnosed as autistic, but an Education Psychologist assessment concluded that she probably is."

What health conditions affect adults with KBG syndrome?

There are a number of mental and physical health conditions associated with KBG syndrome that can affect individuals into adulthood. Some children with KBG syndrome will have had complex needs and health issues, which will continue to affect them in adulthood to varying degrees. Others will not have had any associated health problems in childhood, and this may also continue into adulthood. It is important that there is an awareness of the health issues for adults with KBG syndrome so that early identification and appropriate treatment can be started.

The following is a description of some of the main health concerns reported by adults with KBG syndrome, with some data taken from the 2024 survey. However, it is important to remember that many of these have only affected a small number of individuals and will not apply to everyone.

Mental health and behaviour

Some adults with KBG syndrome have no mental health or behavioural concerns at all. There are however high rates of anxiety, ASC and ADHD reported. Some, but not all adults, have a learning (intellectual) disability. This can range from those who are mildly affected to others with severe impairment. Sensory-seeking and sensory-avoiding behaviours are common and are known to be associated with neurodevelopmental conditions such as ASC and ADHD.

In the 2024 survey, 19% of participants had also experienced depression and 12% had recurring unwanted thoughts, ideas or sensations (obsessions) that compelled them to act in certain ways. Aggression towards themselves or others was reported by 35% of participants. Binge eating and reduced eating were also reported in 15% and 11% respectively.

It is important to explore any contributing factors for someone who is experiencing mental health issues, such as anxiety or behavioural difficulties, in order to support them better. This should be a holistic assessment looking at someone's biological, psychological and social wellbeing. Support could be provided by multidisciplinary teams involving doctors, nurses, psychologists, occupational therapists and speech and language therapists, amongst others. This is in line with National Institute for Health and Care Excellence (NICE) guidelines in the UK which supports a positive behavioural support approach to behaviours that challenge for people with learning (intellectual) disability.

"Our daughter has anxiety and is stressed easily so things need to be organised, controlled and in a routine. She has anxiety attacks if plans change or she is too tired."

"Our son has anxiety, depression and attention deficit disorder."

"Our daughter struggles with anxiety which impacts on her life every day and if her anxiety mounts, she can display challenging behaviour and show autistic traits. She will have meltdowns, can hit, pull hair, swear, throw things and shout as signs of frustration when perceived needs are not being met or she feels out of control and unable to communicate effectively. She asks questions repetitively to try and make sense of and control the world around her and often seems to be in a high state of arousal, requiring much reassurance and support. She struggles with transitions, anything that is different or unexpected or when out of routine and needs careful preparation for new activities / events in advance. She is very sensory and sensitive to smells, tastes, loud noises and busy environments. She has a diagnosis of autism, ADHD, dyspraxia and anxiety. She can suffer from low mood due to feelings of being different, low self-esteem, loneliness, and lack of confidence. She takes medications for anxiety and ADHD. Her epilepsy causes further anxiety, and she worries about sleeping at night for fear of having seizures.

Mental health support has been particularly poor, especially transition into adult services. She previously had some support by Child and Adolescent Mental Health Services (CAMHS) but since transferring to adult services (involving an 18 month wait) she has had very little input at a time when she has struggled most. Neurology services are also very stretched, and we have had far less frequent follow up by adult services than when she was in the paediatric team. Appointments regularly get cancelled and set back."

"Our daughter is usually very upbeat when everything is going her way but can spiral into depression if plans are changed or something goes wrong, and she takes a while to calm down."

Neurological conditions

It is known that KBG syndrome is associated with seizures in children and adults. The 2024 survey found that almost a third of participants had experienced seizures at some point. For those who had experienced seizures, around half were currently controlled in adulthood by medication or a ketogenic diet, just over quarter had treatment resistant seizures, and just under a quarter no longer required medication. It was noted that of the 21 participants currently experiencing seizures, none of these individuals were living independently. Tics, where repetitive muscle movements result in sudden and difficult-to-control body movements or sounds, were reported by 31% of participants.

"No neurological conditions."

"Our daughter has suffered from epilepsy since the age of 8 and suffers seizures approximately four times per year. She has symptomatic epilepsy with generalised tonic seizures and complex partial seizures arising from the temporal lobe. Her MRI shows hippocampi inversion incomplete; EEG shows right temporal spike with hyperventilation. She is treated with medication."

"Suffers from short absence seizures (10-20 seconds) but does not take medication."

Sleep

It is probably less well known how KBG syndrome affects an individual's sleep, and this is an area which may be explored in future studies. The 2024 survey found that around a quarter of participants (26%) had sleep problems which negatively impacted their quality of life in adulthood. On the other hand, 24% did not experience any sleep-related issues. Difficulty falling asleep affected 28%, with almost half of these requiring medication like melatonin to help initiate sleep. Similarly, 26% had trouble staying asleep, while 21% experienced an increased need for sleep. Suddenly falling asleep during the day was reported by 9% participants. Additionally, 5% experienced sleep apnoea, and 4% had restless leg syndrome. Identifying and treating sleep problems should be part of holistic treatment plans, especially since this is likely to impact some of the mental health and behavioural difficulties that are also associated with KBG syndrome.

"She gets very tired due to having KBG syndrome. She is worn out quickly. She talks in her sleep and calls out. This affects her sleep."

"She struggles to settle to sleep due to anxiety. She also worries about seizure activity overnight. She takes Melatonin with good effect."

"Sleeps well."

Digestive (gastrointestinal) system

There is an increasing amount of research to suggest that digestive (gastrointestinal) problems affect adults with KBG syndrome. The most commonly reported conditions are constipation and reflux. It is important that individuals with KBG syndrome and their families are informed about how to manage these conditions and where to seek support if needed. In the 2024 survey, 29% of participants reported constipation and 21% reflux. It was also identified in this survey that there were higher than expected rates of abdominal migraine (8%) and cyclical vomiting (7%). This has also been found in previous research studies. There is a need for further research into these associations.

"She only eats small amounts or she feels sick, so she can only manage small amounts of food and is very underdeveloped and small for her age."

"He has constipation and reflux and is of small stature."

"She suffers from chronic constipation and has a difficult relationship with food. She can also be very sensory in relation to food and has a strong dislike for anything that has a strong taste or smell such as oranges, garlic etc. She is overweight and struggles to maintain a healthy weight, despite her best efforts with healthy eating and exercise. She can eat to self soothe when anxious and sometimes hides food. She struggles to order food options on a menu, conflicted between wanting food that she likes and food that is healthy."

"No known problems but does have days where she gets 'stuck on the loo' for a long time."

Heart (cardiovascular system) conditions

Heart conditions have been found in a small number of people so far with KBG syndrome. These can be present at birth (congenital) or develop later in life. There have been some reports of widening (dilatation) of the aorta (a blood vessel from the heart) and heart valve problems. Most of these will have been identified in childhood; however, it is considered an area where further research is needed and tests like an electrocardiogram (ECG recording of the electrical activity of the heart) and echocardiogram (ultrasound scan of the heart) should be considered in adulthood.

"Her heart has been checked previously. All is OK at the moment."

"No concerns."

"No known problems."

Bones (musculoskeletal conditions)

KBG syndrome is known to affect the spine. Some individuals may be born with or develop a spinal curvature, either a sideways curve of the spine (scoliosis), a rounding of the upper back (kyphosis), or an inward curvature of the lower spine (lordosis). The 2024 survey found that adults also experienced hip problems (15%), arthritis (8%) and osteoporosis (5%), along with foot deformities (8%), which sometimes required surgery. The researchers highlighted the importance of optimising bone health, maintaining healthy physical activity, and appropriate investigation and treatment of painful joints.

"Her bones are weak. She drops things and cannot grasp things."

"One foot two sizes smaller than the other. Both feet have high inside arches."

“She has dyspraxia and suffers from poor co-ordination and is prone to falls. She has small hands and feet and wears orthotics in her shoes. She is also of small stature. We are unaware of any musculoskeletal conditions, but this has not been explored clinically.”

Hearing and vision (sight) concerns

It is well known that KBG syndrome is associated with hearing loss (impairment). This can be a result of differences in the structure of the ear and recurrent ear infections and can require hearing aids. In the 2024 survey, 35% of participants reported that hearing concerns were affecting their quality of life in adulthood. The survey also found 36% of participants reported visual difficulties, with a variety of different causes.

Hearing and visual loss (impairment) are likely to affect a person’s ability to engage in activities, and can contribute to low mood and anxiety, along with sensory seeking / avoiding behaviours, especially in people who have learning (intellectual) disability. This makes it particularly important that regular hearing and visual checks are carried out for adults with KBG syndrome.

“She has grommets to help her hearing as it was only 20 per cent. She has problems with her eyes. She wears glasses – has done from 4 years until now.”

“Has reading glasses.”

“She has mild-moderate hearing impairment and wears hearing aids (we understand that this is due to a congenital issue in her ossicles). She also wears glasses.”

“Congenital hearing loss - severely deaf in both ears. Registered partially sighted – nystagmus”

Teeth (dental) concerns

Dental concerns are very common in people with KBG syndrome. A number of issues have been described including misshapen teeth, issues with dental enamel, issues with the palate, dental decay, an excess of teeth, crumbling teeth, and the absence of teeth, as well as milk teeth that did not fall out. A high standard of dental care is important to minimise damage and regular dental check-ups are really important. Adults with complex needs may also benefit from specialist hospital dental services and may require treatment under general anaesthetic.

“She has had root canal dental treatment twice. She has over-crowded teeth, and large front incisors. Her roots are very short so she cannot wear a brace.”

“Large teeth as described in diagnosis.”

“Macrodontia [large teeth], lower and upper teeth very muddled. Currently looked after by the local Dental Access Centre.”

“No concerns.”

Unique publishes separate guides to Teeth: common concerns.

What care and support might an adult with KBG benefit from?

Adults with KBG syndrome are, of course, all unique individuals. The health care and support they will benefit from should be tailored to their own personal needs and preferences. Some individuals will need none at all and others will have complex needs requiring care from multiple professionals and support in their day to day lives.

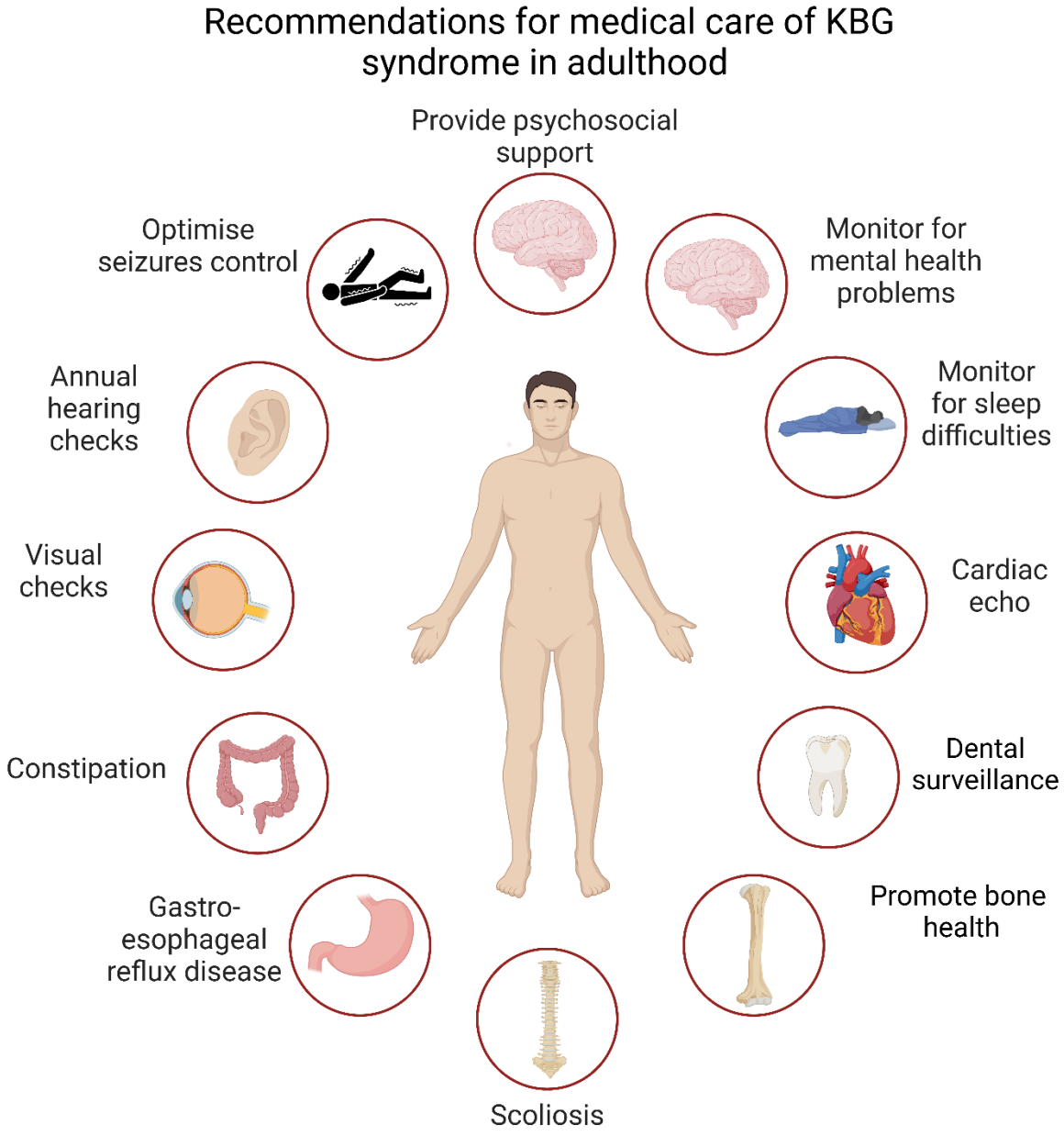
We do know that some difficulties are more common for adults with KBG syndrome, and hopefully early identification and support for these will help to improve quality of life and allow individuals to achieve their full potential. The researchers who carried out the 2024 survey created the image on the next page to illustrate their recommendations for healthcare provision for adults with KBG syndrome. These can be used to help individuals and professionals plan for the future.

"She needs help and support with all tasks. College work needs prompting, and she needs help to check her clothes – she cannot tell if her clothes are dirty. She needs prompting to shower. Help with travelling – cannot read timetables."

"He is isolated and would love to socialise to build friendships. He attends day program two days per week and works one day per week. After three years he just got accepted into a respite home (28 nights per year)."

"Regular heart, hearing and eye checks. Specialist dental care. Support with independence and mobility. Well-being support. Everyday life can be isolating. She has few friends and is reliant on parents to take her to places. She spends a lot of time on her own. She talks out loud to herself when she feels safe – mainly at home. Very routine orientated, gets up and goes to bed at the same time, eats and makes drinks at the same time each day."

"We have little family support and no respite or support options available to us during school holidays so the impact on our family is significant. We have had to push for a social care assessment and applications for benefits such as Personal Independence Payment, a Blue Badge and Universal Credit, but we now get these for our daughter. She finds them helpful, and they allow her to feel more independent having money of her own."



Families say ...

"We have struggled throughout our daughter's life to get the help and support she needs as each individual diagnosis has been looked at in isolation, rather than looking at the complex combination of difficulties and how they interact and impact her. Her KBG diagnosis was only made when she was in her late teen. Had we had the diagnosis earlier, the other diagnoses would have been easier to realise, and support could have been provided much earlier."

"I find as a parent that overall, the services available and awareness of anyone with special needs has improved. However, most websites are about gathering more data. I wish there were more supportive in-person options. Our son still feels isolated and would love to be on the show "Love on the Spectrum" on Netflix."

Sources

The information in this guide is drawn from the other Unique KBG syndrome guide, a survey of adults with KBG syndrome published in 2024 (preprint available online), published medical literature, and Unique members. In 2025, Unique had 93 member families with KBG syndrome, over 10% (1 in 10) of whom are known to be adults. The first-named author and publication date for articles in the medical literature are given to allow you to look for the abstracts or original articles on the internet in PubMed (www.pubmed.ncbi.nlm.nih.gov/). You can obtain most articles from Unique.

References

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Note: an asterisk indicates articles which are “open access” and available to everyone at www.pubmed.ncbi.nlm.nih.gov

Thanks to Dr Allan Bayat (Department of Pediatrics, Danish Epilepsy Center, Dianalund, Denmark & Department of Regional Health Research, University of Southern Denmark, Odense, Denmark) for his contribution to the original study and for formatting the figure “Recommendations for medical care of KBG syndrome in adults” in this leaflet.

Websites, Facebook groups and Support Groups

KBG UK private FB group: FB group for UK-based patients, their families/carers, physicians and researchers: www.facebook.com/groups/1580076562129125/

KBG Foundation - US-based nonprofit devoted exclusively to KBG syndrome:
www.kbgfoundation.org/
www.facebook.com/groups/kbgsyndromefamilygroup/
www.facebook.com/KBGFdn/

KBG Syndrome Association (KBGSA) - US-based patient advocacy organization dedicated to improving the lives of individuals diagnosed with KBG Syndrome:
www.kbgsyndrome.org/
www.facebook.com/KBGSyndrome/

National deaf children's society (NDCS) - Dedicated to creating a world without barriers for deaf children and young people: www.ndcs.org.uk/

National Autistic society - Supports the autistic people in the UK and their families:
www.autism.org.uk/

Dyspraxia foundation - Supports individuals and families affected by developmental dyspraxia:
www.dyspraxiauk.com/dyspraxia-foundation

Kids SENDIASS - Free and impartial information, advice, and support for the families of children and young people with disabilities or special educational needs: www.kids.org.uk/sendiass-home/

Epilepsy Action - A UK-based charity committed to supporting a better life for everyone affected by epilepsy: www.epilepsy.org.uk

Young Minds - A UK-based charity fighting for a world where no young person feels alone with their mental health: www.youngminds.org.uk/

CALM - Campaign Against Living Miserably: www.thecalmzone.net/

Inform Network Support

Rare Chromosome Disorder Support Group
The Stables, Station Road West, Oxted, Surrey, RH8 9EE, UK.
Tel: +44(0)1883 723356
info@rarechromo.org I www.rarechromo.org

UNDERSTANDING GENES & CHROMOSOMES

Join Unique for family links, information and support:

www.rarechromo.org/join-us/

Unique is a charity without government funding, existing entirely on donations and grants. If you can, please make a donation via our website at:
www.rarechromo.org/donate/
Please help us to help you!

This information guide is not a substitute for personal medical advice. Families should consult a medically qualified clinician in all matters relating to genetic diagnosis, management and health. Information on genetic changes is a very fast-moving field and while the information in this guide is believed to be the best available at the time of publication, some facts may later change.

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Version 1 (CA)

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Charity Number 1110661
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