

Understanding Bone Health in KAT6 Syndromes

What Families Should Know About Bone Density

Why Bone Health Matters

Bones are constantly changing throughout life. Specialized cells build new bone while others remove old bone. The KAT6A and KAT6B genes help regulate many developmental processes, including pathways involved in bone growth and remodeling.

Children with KAT6 syndromes may have several factors that can increase their risk for reduced bone density. While researchers are still learning exactly how KAT6 variants affect bone health, understanding these risks allows families and healthcare providers to monitor bone health proactively.

Why Bone Density May Be Reduced

Low Muscle Tone (Hypotonia)

Muscles place healthy stress on bones every time we move. This mechanical stress helps bones become stronger.

Because hypotonia is common in KAT6 syndromes, bones may receive less stimulation needed for normal strengthening.

Delayed Weight-Bearing

Standing, walking, jumping, and other weight-bearing activities help build bone throughout childhood. Children who experience delays in sitting, standing, or walking may develop lower bone mineral density over time.

Feeding and Nutrition Challenges

Many individuals with KAT6 syndrome experience feeding difficulties, gastrointestinal problems, or require tube feeding.

These challenges can sometimes make it harder to maintain adequate intake of nutrients important for bone health, including:

- Calcium
- Vitamin D
- Protein
- Phosphorus

Possible Effects of the KAT6 Genes

Emerging laboratory research suggests that KAT6 proteins may play a role in regulating the balance between:

- Osteoblasts (cells that build bone)
- Osteoclasts (cells that break down bone)

Researchers are continuing to investigate whether changes in these pathways contribute directly to reduced bone density in people with KAT6 syndromes.

Supporting Healthy Bones

Talk with your child's healthcare team about whether the following may be appropriate:

✓ Bone Density Testing

A DEXA (DXA) scan measures bone mineral density and may be recommended if there are concerns about bone health or fracture risk.

✓ Blood Tests

Your physician may recommend monitoring:

- Vitamin D
- Calcium
- Phosphorus
- Alkaline phosphatase

These tests help identify nutritional deficiencies or metabolic concerns that can affect bone health.

✓ Weight-Bearing Therapy

Physical therapists can recommend activities and equipment that safely promote bone strength, including:

- Standing frames
- Gait trainers
- Adapted walkers
- Supported walking programs

✓ Specialist Referral

If low bone density (osteopenia or osteoporosis) is identified, your physician may refer your child to a pediatric endocrinologist or metabolic bone specialist to discuss additional evaluation and treatment options.

✓ Nutrition

Ensure adequate intake of:

- Calcium
- Vitamin D
- Protein

A dietitian can help if feeding difficulties are present.

Signs Families Should Watch For

Contact your healthcare provider if you notice:

- ✓ Pain without an obvious injury
- ✓ Crying during transfers or position changes
- ✓ Refusal to stand or bear weight
- ✓ Swelling or tenderness of an arm or leg
- ✓ Fractures after minor falls or injuries

Looking Ahead

Researchers continue to study how KAT6A and KAT6B influence bone development. As new discoveries emerge, recommendations for monitoring and treatment may continue to evolve. Routine medical follow-up, good nutrition, physical activity, and appropriate screening can all help support lifelong bone health.

Medical Disclaimer

This resource is intended for educational purposes only and should not replace medical advice from your healthcare provider. Families should discuss any concerns regarding bone health, testing, or treatment with their child's medical team.