The Focus Foundation

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The Focus Foundation presents...

A PARENT'S GUIDE TO A 49,XXXXY DIAGNOSIS

You've just found out your son has extra X chromosomes. **What's next?**











Critical info and hope for today...

and every day forward, based on 20 years of experience!

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What is 49,XXXXY?

If you've just heard the term "49,XXXXY," it's completely understandable to feel unsure or overwhelmed. Because this diagnosis is rare, all parents have questions or worries early on. 49,XXXXY is a variation of Klinefelter syndrome (KS) and means your child was born with three extra X chromosomes. It occurs in about 1 in 85,000-100,000 boys.

Children with 49,XXXXY learn and grow at their own pace. Most boys may require extra support with speech, movement and learning, as these skills may come a little more slowly. Most children may also have physical differences that require monitoring, such as growth differences or orthopedic challenges, but not every child will experience the same difficulties.

With early support, therapies, medical care, and patient guidance, boys with 49,XXXXY continue to make steady progress and move forward in meaningful ways. These supports help build communication, mobility, confidence, and independence. Most importantly, your child is loved, capable, and full of potential!

Treatment

Therapies like physical, occupational, speech, and neurodevelopmental therapy (NDT) are key tools that can help your son build important skills for everyday life and a strong foundation for the future. These therapies should target delays in motor, speech, coordination, learning, and daily tasks, offering a strong base for future growth and long-term development.

Another avenue to explore is **hormonal replacement therapy (HRT).** Boys with 49,XXXXY have decreased levels of testosterone in their bodies, which plays a role in learning and developmental challenges. Providing testosterone during certain key stages in life can help support healthy development!



"Early intervention truly matters. Getting common 49-related concerns checked early and paying attention can make a big difference. The diagnosis itself is a gift, because it gives you the information you need to support your child and help them reach their potential."
-Jamie, mom of 12 year old with 49,XXXXY

Treatment Continued

Infancy (EHT)

- The first period for intervention is during the mini-puberty of infancy, with treatment at **4, 5 and 6 months of age**. During this time, infants experience critical changes in hormonal levels that are crucial for their neurodevelopment. Early hormonal treatment (**EHT**) involves giving three testosterone injections over three months. Scientific research indicates that **EHT** enhances cognitive, social, and behavioral capabilities in boys with 49,XXXXY.

Childhood (HBT)

- The second intervention period occurs between the ages of **5 and 8 years of age** typically. During this time, a hormonal booster treatment (**HBT**) can be administered. This involves three testosterone injections over three months to help stimulate the onset of puberty.

Puberty (TRT)

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Common orthopedic issues & optimizing mobility

Musculoskeletal problems are common in boys with 49,XXXXY. In infancy, low muscle tonus in trunk, lower, and upper extremities are often present from birth.

Congenital hip dislocation occurs more often in boys with 49,XXXXY, so ultrasound of both hips is an important assessment tool in monitoring changes.

Congenital Muscular Torticollis is more frequent and is typically observed as early as two months of age.

Treatment should include an evaluation by a pediatric orthopedist and pediatric physical therapist in order to proactively address these issues and receive treatment when necessary.

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Communication, speech and feeding



Children with 49,XXXXY often experience delays and differences in communication, cognition, speech, and feeding, though abilities vary widely. However, they understand spoken language very well.

Speech development is typically delayed. Many children have difficulty with **speech motor planning**, meaning they may understand more than they are able to say verbally. Spoken language may develop slowly and many young children benefit from augmentative and alternative communication (AAC). This may include gestures, sign language, picture systems, or speech-generating devices. These supports promote speech development, and often reduce frustration while making it easier for them to share their thoughts and feel understood! They also often encourage receptive language development.

Some children also have early feeding challenges. Low muscle tone, oral-motor coordination differences, sensory sensitivities, and a higher frequency of oral structural differences (such as tongue ties and lip ties) affect chewing, swallowing, and tolerance of different textures.

Speech-language pathologists support both feeding and communication skills, helping children build effective ways to express themselves and participate in daily life.

Developmental milestone ranges

Sitting	**12-18 months**
4-point Crawling	**12-30 months**
Walking	**30-36 months**
First Words (other than “mom/dad”)	**24-36 months**



These ranges reflect common patterns seen in children with 49,XXXXY. Development varies widely, and progress continues steadily with appropriate therapies and support!

Behavioral challenges and social development

Many children are socially interested and affectionate, but may have difficulty with emotional regulation, attention, or adapting to transitional changes in routine. Anxiety, impulsivity, or challenges with transitions and sensory processing are common. These behaviors are related to 49,XXXXY and the communication challenges, and never intentional behavior problems.

Social skills may develop on a variable timeline. Most children benefit from explicit teaching & support to build skills such as turn-taking and peer interaction. With guidance, boys with 49,XXXXY will form meaningful relationships and enjoy social connection!



Behavioral and social support strategies may include structured routines, visual supports, clear expectations, and positive reinforcement. When needs are understood and supports are in place, behavior improves and boys engage at home, school, and the community better.

49,XXXXY: Fact vs. Fiction

Many people haven't heard of 49,XXXXY, so it can be misunderstood and myths can spread—*especially* online. Here are a few common myths and what the research actually shows:

"Testosterone does not influence brain development in boys with 49,XXXXY."

Research shows early testosterone matters. Studies find benefits for learning and behavior when specific strategies are implemented at strategic times.

"Boys with 49,XXXXY are nonverbal and will never talk."

With early, specialized speech therapy, and tools like sign or AAC to bridge the gap, boys build real communication and many become verbal.

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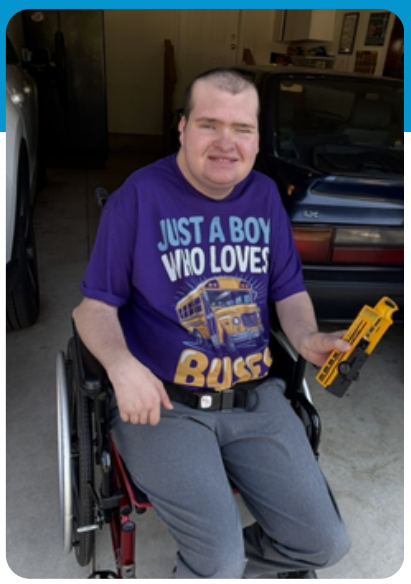
"All boys with 49,XXXXY have feeding problems."

Many boys feed without difficulty. For those who do struggle, it's important to rule out a tongue-tie early on and occupational therapy can make a real difference.

"Boys with 49,XXXXY are badly behaved."

Challenging behavior usually reflects difficulty with communication, emotional regulation, or transitions — not defiance. With understanding and the right supports, behavior improves.

Looking Ahead



When you first hear that your child has 49,XXXXY, it's normal to feel unsure of what to expect or to feel mixed emotions due to this surprise. Your son can absolutely live a full, happy, and meaningful life! With love, support, and understanding, boys with 49,XXXXY grow into delightful young men who achieve their goals, build friendships, and thrive in their own unique ways. They are delightful young men.

"My advice to other parents is simple: hold onto HOPE! Our 49er boys are so much more than the diagnosis or limits in any medical article. They reach milestones but in their own time. Surround yourself with a trusted expert team like The Focus Foundation and Dr. Sprouse have been to our family. Lean on parent support groups as well to connect with others that have walked this same path. And embrace the sunrises and sunsets with a full heart knowing your son will thrive in his own unique way."
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You're not alone in this journey. Dr. Samango-Sprouse and her staff are here to support you, care for your child and make sure he becomes the best he can be! We are here to help and we have physicians, therapists and interventionists throughout the world. Together, we can help your son shine—not despite his diagnosis, but because of the strength, empathy, and perspective that it helps him develop. Every boy's path is different, but with the right tools and belief in his abilities, he can flourish.

About Us

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VARIATIONS
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The Focus Foundation is a non-profit organization dedicated to helping children with X & Y chromosomal variations reach their full potential. For two decades, we have been at the forefront of research, early identification, and neurodevelopmental care for children with 47, XXY, 47,XYY, 47,XXX, 49,XXXXY and related conditions.

Our Expertise

The foundation was co-founded by **Dr. Carole Samango-Sprouse,** a world-recognized leader in X and Y chromosomal variation research.

- She is the author of the book *X & Y Chromosomal Variations: Hormones, Brain Development, and Neurodevelopmental Performance*.
- She has published over **120** peer-reviewed scientific papers.
- She is a Clinical Professor of Pediatrics at George Washington University, as well as an Adjunct Associate Professor at Florida International University.
- Her work has shaped how clinicians worldwide understand early development in children with 47, XXY.

What We Do

We bridge the gap between cutting-edge science & everyday life by offering:

-Syndrome-specific evaluations by experts who understand the unique developmental needs of our patients

-Guidance on early intervention + individualized support

-Access to ongoing research + the newest findings

-A family-centered approach that emphasizes strengths, progress, and hope

Glossary of Terms

Helpful terms for parents to understand their son's 49,XXXXY diagnosis.

- *Amniocentesis*- a prenatal test involving extracting amniotic fluid from the uterus to screen for certain health conditions.
- *Androgen deficiency*- a condition when the body does not produce enough male hormones, such as testosterone.
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- *Mini-puberty*- a period of increased hormonal activity during infancy, between 2-6 months, with a peak around 3-4 months. Mini-puberty has a significant impact on cognitive and physical development of all males.
- *Mosaicism*- a condition where an individual has two or more genetically different sets of cells in their body. Mosaicism in boys with KS can mean they have some cells that are 47,XXY and some that are 46,XY (normal male karyotype) and can cause milder symptoms. The typical level of mosaicism is between 15-20%
- *Muscular Torticollis*- a condition in which the infant's neck muscles are tight or shortened, resulting in the head being tilted or turned to one side. This can restrict motion, affect muscles on that side of the body, or cause plagiocephaly (flattening on one side of the head). It is addressed with early pediatric physical therapy.
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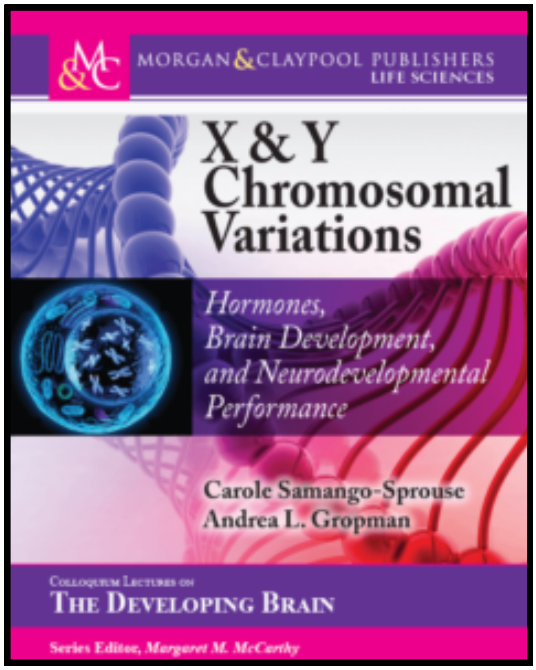
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Check out Dr. Samango-Sprouse's book! Available at Barnes & Noble or Amazon, link available on our website!

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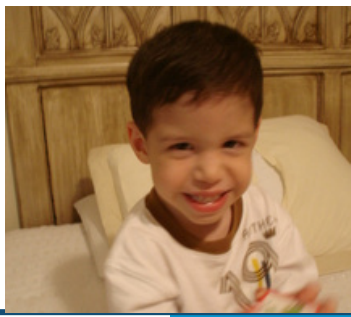
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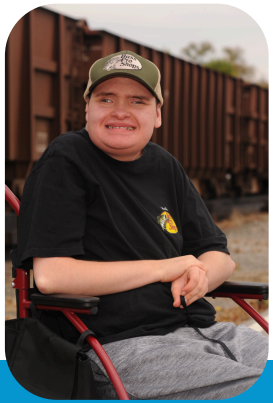
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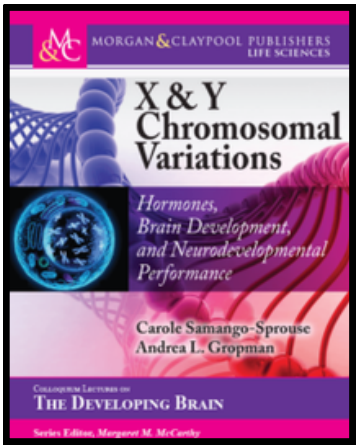
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Find us on
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Check out Dr. Samango-Sprouse's book! Available at Barnes & Noble or Amazon, link available on our website!

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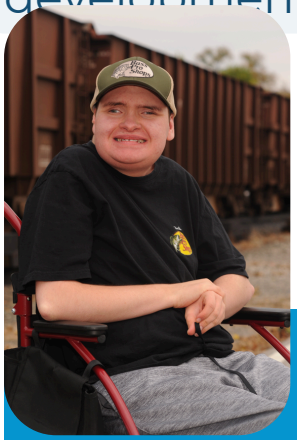
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Another avenue to explore is **hormonal replacement therapy (HRT).** Boys with 49,XXXXY have decreased levels of testosterone in their bodies, which plays a role in learning and developmental challenges. Providing testosterone during certain key stages in life can help support healthy development!



"Early intervention truly matters. Getting common 49-related concerns checked early and paying attention can make a big difference. The diagnosis itself is a gift, because it gives you the information you need to support your child and help them reach their potential."

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Communication, speech and feeding

Children with 49,XXXXY often experience delays and differences in communication, cognition, speech, and feeding, though abilities vary widely. However, they understand spoken language very well.

Speech development is typically delayed. Many children have difficulty with **speech motor planning**, meaning they may understand more than they are able to say verbally. Spoken language may develop slowly and many young children benefit from augmentative and alternative communication (AAC). This may include gestures, sign language, picture systems, or speech-generating devices. These supports promote speech development, and often reduce frustration while making it easier for them to share their thoughts and feel understood! They also often encourage receptive language development.

Some children also have early feeding challenges. Low muscle tone, oral-motor coordination differences, sensory sensitivities, and a higher frequency of oral structural differences (such as tongue ties and lip ties) affect chewing, swallowing, and tolerance of different textures.

Speech-language pathologists support both feeding and communication skills, helping children build effective ways to express themselves and participate in daily life.

Developmental milestone ranges

Sitting	**12-18 months**
4-point Crawling	**12-30 months**
Walking	**30-36 months**
First Words (other than “mom/dad”)	**24-36 months**



These ranges reflect common patterns seen in children with 49,XXXXY. Development varies widely, and progress continues steadily with appropriate therapies and support!

Behavioral challenges and social development

Many children are socially interested and affectionate, but may have difficulty with emotional regulation, attention, or adapting to transitional changes in routine. Anxiety, impulsivity, or challenges with transitions and sensory processing are common. These behaviors are related to 49,XXXXY and the communication challenges, and never intentional behavior problems.

Social skills may develop on a variable timeline. Most children benefit from explicit teaching & support to build skills such as turn-taking and peer interaction. With guidance, boys with 49,XXXXY will form meaningful relationships and enjoy social connection!



Behavioral and social support strategies may include structured routines, visual supports, clear expectations, and positive reinforcement. When needs are understood and supports are in place, behavior improves and boys engage at home, school, and the community better.

49,XXXXY: Fact vs. Fiction

Many people haven't heard of 49,XXXXY, so it can be misunderstood and myths can spread—*especially* online. Here are a few common myths and what the research actually shows:

"Testosterone does not influence brain development in boys with 49,XXXXY."

Research shows early testosterone matters. Studies find benefits for learning and behavior when specific strategies are implemented at strategic times.

"Boys with 49,XXXXY are nonverbal and will never talk."

With early, specialized speech therapy, and tools like sign or AAC to bridge the gap, boys build real communication and many become verbal.

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Every boy is different. 49,XXXXY presents across a wide spectrum in development, personality, and medical needs. Strengths and challenges vary from child to child, and no single story defines them all.

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Height varies widely. Some boys are shorter, especially early on, while others are average or quite tall. There's no single height that defines them, but they tend to be tall.

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Many boys feed without difficulty. For those who do struggle, it's important to rule out a tongue-tie early on and occupational therapy can make a real difference.

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Challenging behavior usually reflects difficulty with communication, emotional regulation, or transitions — not defiance. With understanding and the right supports, behavior improves.

Looking Ahead



When you first hear that your child has 49,XXXXY, it's normal to feel unsure of what to expect or to feel mixed emotions due to this surprise. Your son can absolutely live a full, happy, and meaningful life! With love, support, and understanding, boys with 49,XXXXY grow into delightful young men who achieve their goals, build friendships, and thrive in their own unique ways. They are delightful young men.

"My advice to other parents is simple: hold onto HOPE! Our 49er boys are so much more than the diagnosis or limits in any medical article. They reach milestones but in their own time. Surround yourself with a trusted expert team like The Focus Foundation and Dr. Sprouse have been to our family. Lean on parent support groups as well to connect with others that have walked this same path. And embrace the sunrises and sunsets with a full heart knowing your son will thrive in his own unique way."
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You're not alone in this journey. Dr. Samango-Sprouse and her staff are here to support you, care for your child and make sure he becomes the best he can be! We are here to help and we have physicians, therapists and interventionists throughout the world. Together, we can help your son shine—not despite his diagnosis, but because of the strength, empathy, and perspective that it helps him develop. Every boy's path is different, but with the right tools and belief in his abilities, he can flourish.

About Us

The Focus Foundation
research · awareness · treatment

The Focus Foundation is a non-profit organization dedicated to helping children with X & Y chromosomal variations reach their full potential. For two decades, we have been at the forefront of research, early identification, and neurodevelopmental care for children with 47, XXY, 47,XYY, 47,XXX, 49,XXXXY and related conditions.

Our Expertise

The foundation was co-founded by **Dr. Carole Samango-Sprouse,** a world-recognized leader in X and Y chromosomal variation research.

- She has published over **120 peer-reviewed scientific papers**.
- She is the author of the book *X & Y Chromosomal Variations: Hormones, Brain Development, and Neurodevelopmental Performance*.
- She is a Clinical Professor of Pediatrics at George Washington University, as well as an Adjunct Associate Professor at Florida International University.
- Her work has shaped how clinicians worldwide understand early development in children with 47, XXY.

What We Do

We bridge the gap between cutting-edge science & everyday life by offering:

-Syndrome-specific evaluations by experts who understand the unique developmental needs of our patients

-Guidance on early intervention + individualized support

-Access to ongoing research + the newest findings

-A family-centered approach that emphasizes strengths, progress, and hope

Glossary of Terms

Helpful terms for parents to understand their son's 49,XXXXY diagnosis.

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- *Klinefelter Syndrome (KS)*- a sex chromosome condition that occurs when a male has at least one extra X chromosome. 47,XXY is the most common chromosome constellation of this disorder.
- *Mini-puberty*- a period of increased hormonal activity during infancy, between 2-6 months, with a peak around 3-4 months. Mini-puberty has a significant impact on cognitive and physical development of all males.
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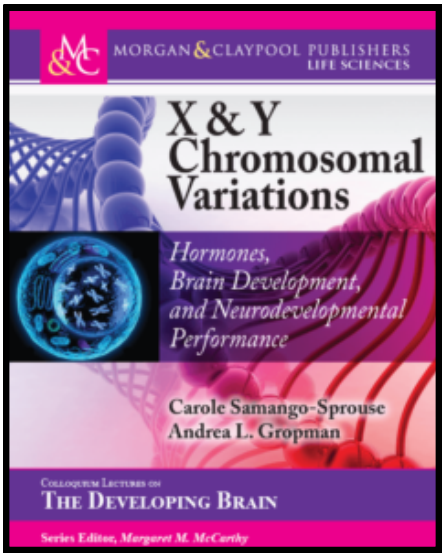
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Check out Dr. Samango-Sprouse's book! Available at Barnes & Noble or Amazon, link available on our website!

THIS BROCHURE WAS FUNDED BY THE FOCUS FOUNDATION

Created by: Suzanne Nibley, Margaret Olaya, and Dr. Carole Samango-Sprouse

The Focus Foundation presents...

A PARENT'S GUIDE TO A 49,XXXXY DIAGNOSIS







Critical info and hope for today...

and every day forward, based on 20 years of experience!

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What is 49,XXXXY?

If you've just heard the term "49,XXXXY," it's completely understandable to feel unsure or overwhelmed. Because this diagnosis is rare, all parents have questions or worries early on. 49,XXXXY is a variation of Klinefelter syndrome (KS) and means your child was born with three extra X chromosomes. It occurs in about 1 in 85,000-100,000 boys.

Children with 49,XXXXY learn and grow at their own pace. Most boys may require extra support with speech, movement and learning, as these skills may come a little more slowly. Most children may also have physical differences that require monitoring, such as growth differences or orthopedic challenges, but not every child will experience the same difficulties.

With early support, therapies, medical care, and patient guidance, boys with 49,XXXXY continue to make steady progress and move forward in meaningful ways. These supports help build communication, mobility, confidence, and independence. Most importantly, your child is loved, capable, and full of potential!

Treatment

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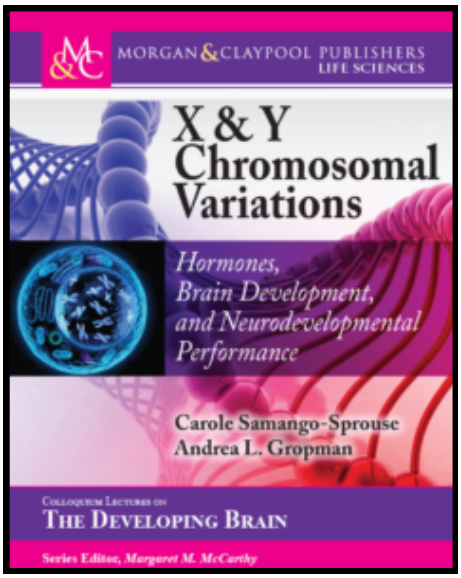
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Check out Dr. Samango-Sprouse's book! Available at Barnes & Noble or Amazon, link available on our website!

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