

Treatment

Many medical professionals in different fields are involved in the treatment for your children because the skills of many different areas are necessary to solve the problems caused by cleft palate. A Cleft team, which usually includes a plastic surgeon, a dental surgeon, an ear-nose-throat (ENT) surgeon, a pediatrician, a speech-language pathologist, and a nurse, will take care of your child. Treatments include mainly surgery, speech therapy, and dental therapy.

Surgery

Surgery for cleft palate repair is usually performed between 10 and 18 months after birth. The surgery, which is called “palatoplasty,” consists of reconstruction of the splitting palate, including not only the mucosa but also the underlying muscle, which is most important for the speech and swallowing. There are several methods of palatoplasty. One of the most common procedures, “push-back” palatoplasty, is shown in Figure 3. In this procedure, incisions are made on both sides of the palate. Then the palatal tissues, including mucosa and muscle, are moved from each side to the center back, and then sutured. With this procedure, the separated muscles are joined together and the palate can be reconstructed and elongated.

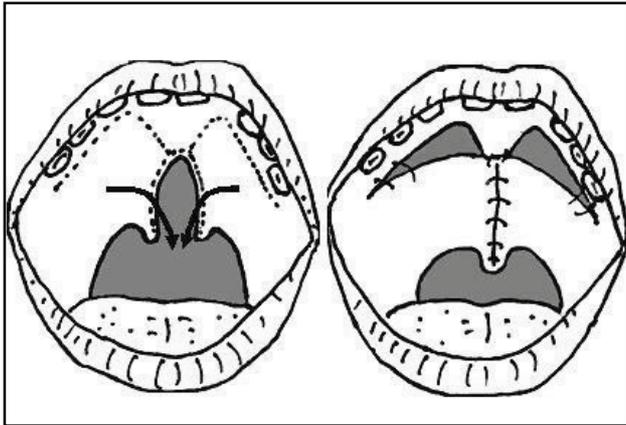


Figure 3

Speech therapy

After palatoplasty, children with cleft palate usually have speech therapy to learn how to use the reconstructed palate properly and acquire the correct pronunciation of sounds and words. The speech-language pathologist will evaluate your child’s speech production and language development. The goal of speech therapy is to help them acquire correct sound and good speech habits.

Dental Care and orthodontic treatment

Children with a cleft palate often need dental and orthodontic treatment. Since the growth of the upper jaw is slower and less than the lower jaw, a child’s upper teeth may not fit together properly with the lower teeth. In such cases, the orthodontist will help correct the alignment of the teeth and the relationship of the upper jaw to the lower jaw. If the tooth alignments cannot be made normal by orthodontics alone, they may need orthognathic surgery, which is called an osteotomy, to reposition the upper jaw both forward and down.

Ear treatment

Children with a cleft palate are susceptible to ear infections, so it is important to have a regular examination by an ENT doctor for your child’s ears. Since Children with severe ear infections are not able to hear language normally due to fluid collection in the middle ear, there is a risk for language delays and speech problems. To obtain proper drainage of the fluid in the middle ear, a small plastic tube is often inserted into the eardrum by an ENT surgeon.

Figures:

1 Appearance of cleft palate

2 Appearance of a submucous cleft palate (cited from *The Cleft Palate-Craniofacial Journal*, Allen Press Publishing Services. 2006, Iida T et. al. Cleft palate in Kabuki syndrome: a report of six cases)

3 Schematic illustration of “push-back” palatoplasty

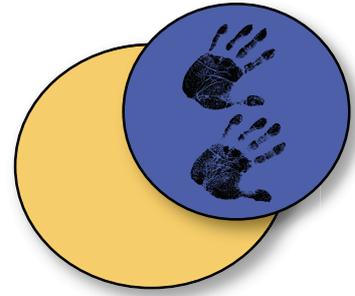
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Musculoskeletal Characteristics



Musculoskeletal characteristics typically include:

- short fingers
- short middle phalanx of fifth finger
- syndactyly - mild webbing between fingers
- cranial abnormalities
- vertebral abnormalities
- rib anomalies
- scoliosis
- Hypotonia
- joint laxity
- dislocations of hip, patella and shoulders



Vertebral anomalies can include butterfly vertebra, sagittal cleft, narrow intervertebral disc space, spina bifida occulta, and scoliosis.

Joint hypermobility is very common, in particular in the younger child. The hypermobility, exacerbated by hypotonia, can lead to dislocation of joints, in particular the hip, knees and shoulders. It is yet unclear whether joint laxity is neurogenic or due to a connective tissue disorder.

Short fingers, in particular the fifth finger, is common. Webbing between the fingers is less commonly seen.



Fetal pads and short fifth finger

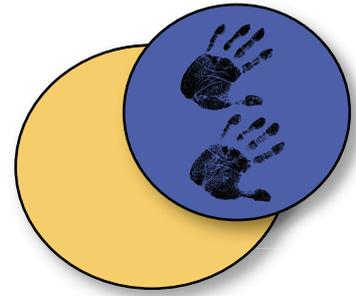


Dislocated Left Knee Cap



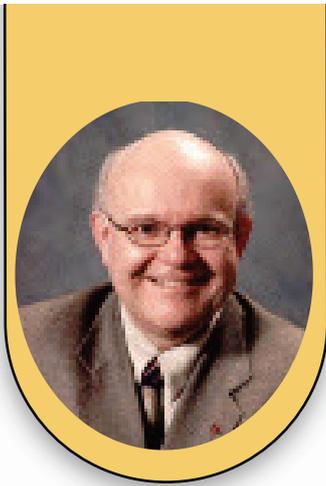
Severe pronation with active overuse of the foot everters in attempt to acquire stability

Dermatoglyphic Characteristics



With the recent genetic discovery for Kabuki, there will be less need to use dermatoglyphics for diagnosis. However, as one of the five cardinal characteristics, it may still be used as a contributing factor for diagnosis.





Dermatoglyphics and Kabuki Syndrome

By Dr. Albert Chudley

About Author:

Dr. Chudley is a medical geneticist and pediatrician. He is a professor at the University of Manitoba, at Winnipeg Children's Hospital.

Dermatoglyphics (writing on the skin in Greek) is the study of epidermal ridges. Epidermal ridges form early in fetal life, and are unique to each individual. They consist of patterns of ridges on the finger pads, palms and soles of all individuals. They form different patterns, and are unique to individuals. This means they can be used for personal identification in criminal investigations. In genetics and medicine, they are useful in diagnosis, since recurring abnormal patterns are often seen in a variety of genetic syndromes. In addition, creases are formed on palms and soles that are also altered in syndromes. Although creases are not part of epidermal ridges, which require a magnifying glass or an ink impression to examine thoroughly, creases are part of what a geneticist looks at during his or her dermatoglyphic analysis and examination.

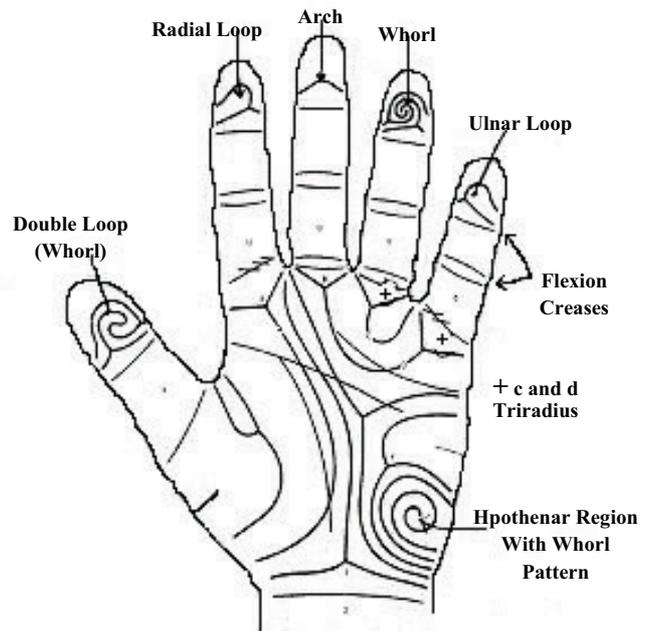
In Down syndrome, the creases are frequently abnormal on the palms with two of the three large creases forming what appears to be a single palmar crease (31% compared to 2% of controls). Also, individuals with Down syndrome have tibial arch patterns on the soles near the base of the great toes (60% compared to 0.5% of controls) and they tend to have 10 ulnar loops on their finger pads (30% compared to 7% of controls).

For Down syndrome, there is a diagnostic test, the chromosome analysis, that confirms the presence of 3 chromosome # 21's, instead of the usual 2. Therefore, dermatoglyphic analysis has become less important for the diagnosis of Down syndrome than for syndromes in which the genetic alteration has not been identified, such as for Kabuki syndrome (KS).

In many children with KS (over 75%), there are prominent fetal fingerpads. Usually these fingerpads become flat by the time of delivery, but in KS individuals, they remain prominent. This is not specific for KS, as they have been described in other syndromes, and can also be present in individuals without a genetic syndrome. Dr. Niikawa and co-authors brought our attention to the fact that in most people with KS, there are dermatoglyphic findings that separate affected individuals from unaffected. His findings showed that there was an increase in ulnar loops (63%);

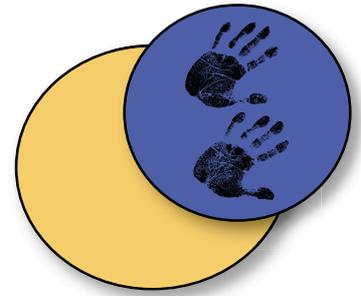
absence of digital triradius c (48%); absence of digital triradius d (30%); increase of hypothenar loops; and a single flexion crease of the 5th finger. Overall, in his study of dozens of KS individuals, about 93% showed some unusual dermatoglyphic findings. (See illustration).

Geneticists use dermatoglyphic analysis to help support the diagnosis of KS. However, as in Down syndrome, eventually the genetic cause of KS will be established, and the use of dermatoglyphic analysis will become less important



This illustration shows the various landmarks related to dermatoglyphics and some common patterns or formations. In Kabuki syndrome, there are at least five commonly seen changes: (1) increase in ulnar loops (2) absent of the digital c or d triradius –region highlighted with asterisk (3) increase in hypothenar patterns (4) single flexion crease in 5th digit (5) prominent fingerpads (not shown).

Intellectual, Sensory and Behavioral



Many individuals with Kabuki syndrome have sensory processing disorder. This inability to accurately organize sensory information can lead to behavior problems.



Some of the more commonly reported sensory issues include need for oral stimulation (chewing on non-food items), tactile defensiveness towards various sensations and stimuli, panic-like reactions to certain noises, and aversion to textures and/or smells of select foods. Anxiety, obsessive/compulsive traits and autistic-type behaviors are commonly observed. Individuals with Kabuki syndrome often have an obsessive need for routine. Mild depression has been reported in young adults.

Parents frequently report an excellent memory for face recognition, song lyrics, dates of events, etc.





Margot Schmiedge

Kabuki and Autistic Behaviors

By Margot Schmiedge and Jen Morton

The spectrum of characteristics associated with Kabuki syndrome is extremely varied. As with any newly described syndrome it is initially difficult to know if certain presenting characteristics are typical of the syndrome or simply typical for that individual. However, it has become increasingly evident that many individuals with Kabuki display autistic-type behaviors. Although few children have been officially diagnosed with autism, virtually all children have some degree of sensory processing disorder.

What is autism?

Autism is a spectrum disorder. This means the symptoms and characteristics can present themselves in a wide range of combinations and from mild to severe. In other words, two children with the same diagnosis can be very different from each other and have varying abilities/disabilities. Autism is a combination of several developmental challenges.

According to the Autism Society of America, the following areas are among those that may be affected:

Communication

- language develops slowly or not at all
- uses words without attaching the usual meaning to them
- communicates with gestures instead of words
- short attention span

Social Interaction

- spends time alone rather than with others
- shows little interest in making friends
- less responsive to social cues such as eye contact or smiles

Sensory Impairment

- may have sensitivities in the areas of sight, hearing, touch, smell, and taste to a greater or lesser degree

Play

- lack of spontaneous or imaginative play
- does not imitate others' actions
- does not initiate pretend games

Behaviors

- may be overactive or very passive
- throws tantrums for no apparent reason
- perseverates (shows an obsessive interest in a single item, idea, activity or person)
- apparent lack of common sense
- may show aggression to others or self
- often has difficulty with changes in routine

Behaviors often associated with children with Kabuki

Communication

- almost all families report language delays

Social Interaction

- some families report their child as being very social, others report their child as having little interest in friendships, preferring to play alone, often able to speak more freely with adults than peers
- poor eye contact (50% according to survey done by KSN)
- poor at understanding the unspoken "rules" of socialization
- poor at understand the give-and-take of a conversation or how to end one
- very literal thinkers, have difficulty thinking abstractly
- unable to 'read between the lines'

Sensory Impairment

- hypersensitive to touch (such as play dough, walking barefoot, etc)
- aversion to loud noises
- aversion to particular smells (cooking smells, etc)
- hypersensitive to visual stimuli
- aversion to particular food tastes and textures (often causing gagging)
- self-stimulatory behaviors such as hand flapping, head shaking, rocking, repeating phrases over and over (over 50% according to survey done by KSN)
- self injurious behaviors such as biting self and head banging
- very oral, many chew on non-food items (over 60% according to survey done by KSN)

Play

- some do not seek out friendships, preferring to play alone or with adults
- others seek friendships but prefer younger children
- many like to play the same thing or watch the same videos over and over

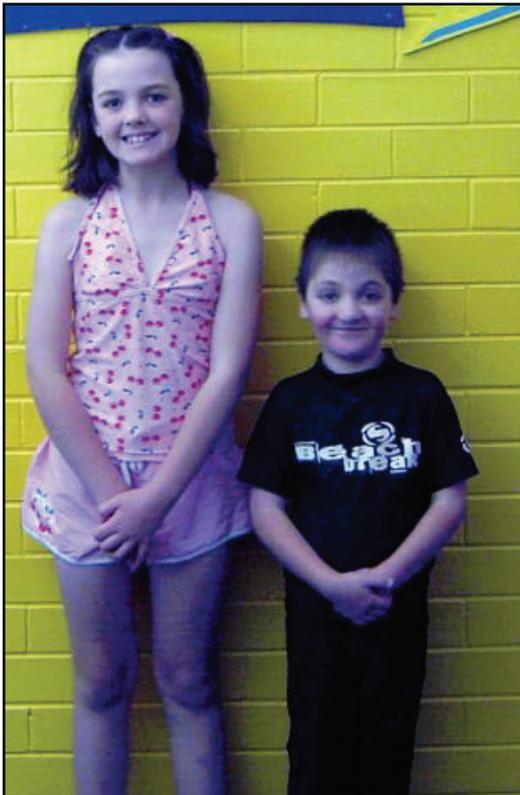
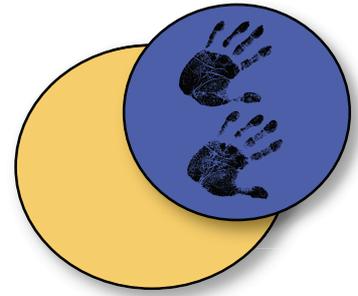
Behaviors

- extreme need to know what to expect throughout the day and exact schedule of events (about 60% according to survey done by KSN)
- repeating of questions over and over
- difficulty waiting
- interrupting often
- talking to self (about 60% according to survey done by KSN)

What does this mean for the child with Kabuki?

It is important to know that developmental delay in general can be accompanied by several types of symptoms and behaviors that one sees with autism (speech and language delay, self-stimulatory behaviors, social impairment, inappropriate behavior). It is true that autism is more easily recognized today and if a child fits into a set of criterion, a diagnosis of autism may come about. This is not to say that the autistic diagnosis is permanent or that it conflicts with the Kabuki diagnosis. With skill development and ongoing intervention, a child may mature and gain ground in an area so that they no longer 'fit' into the autism heading. The fact that our children have Kabuki syndrome is the reason they are demonstrating autistic-like tendencies in the first place. Autism is not necessarily a separate label. More than likely, ALL of our children at some point or other are demonstrating behaviors that could be considered autistic-like. Whether our children have been given an autism label or not, the types of therapy and intervention that we would seek to assist with their areas of need are the same. Many autism treatment approaches are very beneficial for all children facing issues in any of these functional areas.

Growth



Twins Hannah and Zachary on 10th Birthday

Postnatal short stature is one of the cardinal features of Kabuki Syndrome. It is still unclear as to what extent growth hormone deficiency contributes to this characteristic.

Although birth weight and length are generally normal, growth delay often starts during the first year of life. Poorly coordinated sucking & swallowing, reflux, recurrent infections, cardiac defects, and hypotonia may all be contributing factors. Although growth hormone levels are in the normal range for most children, a significant number have a partial or complete deficiency. Obesity seems to be a common problem during puberty years. The adult with Kabuki will be shorter than the norm – two or more standard deviations below the mean.

