Kabuki syndrome has its founding in Japan. In 1967 Dr. Niikawa and in 1968 Dr. Kuroki each identified unique malformations in their patients. Over the next decade, they identified similar malformations in several others. Kabuki syndrome became a formal diagnosis in 1981.

Some syndromes are linked to a chromosome abnormality, but Kabuki syndrome is linked to malformation at the gene level.

There are five cardinal traits used to clinically diagnose Kabuki syndrome.

Kabuki syndrome affects approximately 1 in 32,000 births.

Mutations in two genes are currently identified as a cause for Kabuki syndrome. One is the KMT2D (also known as MLL2) and KDM6A.

Kabuki syndrome was originally referred to as Niikawa-Kuroki syndrome after the doctors who discovered the syndrome. It was first referred to as Kabuki Make-Up syndrome because the elongated eyes resembling those seen in Japanese kabuki theater actors, as was later shortened to Kabuki syndrome.

There are five cardinal traits used to clinically diagnose Kabuki syndrome. One of them being skeletal abnormalities.

The second characteristic for a clinical diagnosis is dermatoglyphic abnormalities.

The third characteristic for a clinical diagnosis is postnatal short stature.

The fourth characteristic for a clinical diagnosis is mild to moderate intellectual disabilities. Approximately 5% of persons with Kabuki syndrome have no intellectual impairment.

The fifth characteristic for a clinical diagnosis is facial features. Facial features are very important in clinically diagnosing Kabuki syndrome. Those with Kabuki syndrome have long palpebral fissures with eversion of outer third, arched eyebrows with sparse outer half, prominent eyelashes, prominent and/or misshaped ears, and a depressed nasal tip.

Persistent fetal pads on the fingers is a common trait of Kabuki syndrome. It is considered a dermatoglyphic abnormality (See Fact #8).

Not a trait used for diagnosing, a “scrunchy face” when smiling big is one of the wonderful traits about Kabuki syndrome.

Flat block feet are a common Kabuki trait.

As many as 75% of persons with Kabuki syndrome will have a mutation on the KMT2D (aka MLL2) gene.

If a person does not show a mutation on the KMT2D (MLL2) gene, it does not mean they do not have Kabuki syndrome. It indicates that another gene is mutated.

Feeding issues are almost always an issue early in life.

Oral fixation is another trait of Kabuki syndrome. The need for oral stimulation goes beyond the normal “hand to mouth” seen in young children.

One of the features of Kabuki syndrome is beautiful, long eyelashes and large eyes.
20. Hypotonia (low muscle tone) is usually found in Kabuki syndrome.

21. Another condition of Kabuki syndrome is hip dysplasia. This condition, like others, is not present in all cases.

22. The first gene recognized to be responsible for Kabuki syndrome was discovered in 2010 at the University of Washington.

23. Kabuki syndrome was first referred to as Kabuki Make-Up syndrome because the elongated eyes resembling those seen in Japanese kabuki theater actors.

24. Another feature of Kabuki syndrome is misshapen ears. Kabuki ears can be larger than usual, protruding, and each can look different from the other.

25. It is common for those with Kabuki syndrome to have sensory integration issues.

26. Hearing loss can be seen in those diagnosed with Kabuki syndrome. This includes the need for hearing aids.

27. Conditions of the eyes can be part of Kabuki syndrome. One of the eye conditions seen is strabismus. This condition does not allow the eyes to line up correctly.

28. Another feature of Kabuki syndrome is having a depressed nasal tip.

29. Joint laxity is common in Kabuki syndrome.

30. Short digits, especially the fifth digit, is common in Kabuki syndrome.

31. Precocious puberty can occur with Kabuki syndrome.

32. Many with Kabuki syndrome display autistic like characteristics.

33. Misshaped teeth, missing teeth, and other dental issues are common in Kabuki syndrome.

34. An estimated 50% of children with Kabuki syndrome will have a cardiovascular malformation.

35. Approximately 5% of Kabuki syndrome is caused by mutations on the KDM6A gene.

36. Occupational Therapy is one of several therapies that is commonly needed with Kabuki syndrome.

37. Gastroesophageal reflux is common in the early years of Kabuki syndrome.

38. Physical Therapy is often needed with Kabuki syndrome.

39. Speech Therapy is usually needed with Kabuki syndrome.

40. Children with Kabuki syndrome often learn to walk much later than typical children.

41. Potty training is a slow and lengthy process in Kabuki syndrome.

42. Renal anomalies can occur in Kabuki syndrome.

43. Cleft palate is one issue associated with Kabuki syndrome.

44. Scoliosis is another condition that can be seen in those with Kabuki syndrome.
45. Spinal bifida occulta can also occur in Kabuki syndrome.

46. The KMT2D (MLL2) gene is located on the long arm of chromosome 12.

47. The KDM6A gene is located on the short arm of the X chromosome.

48. Chronic ear infections are another condition seen in Kabuki syndrome.

49. Coloboma is a condition that can occur in Kabuki syndrome. This condition is caused by a hole, or missing tissue, in the structure of the eye.

50. Seizures can occur in Kabuki syndrome.

51. Children with Kabuki syndrome often experience delayed bone growth. Bone age is confirmed through x-rays.

52. One of the heart conditions seen in Kabuki syndrome is juxtaductal coarctation (narrowing) of the aorta.

53. Feeding therapy can be needed with Kabuki syndrome. It is usually administered by an Occupational Therapist or Speech Language Pathologist.

54. Cleft palate abnormalities can also be part of Kabuki syndrome. It affects speech, eating and even hearing.

55. Children with Kabuki syndrome often have the need to seek sensory input. This is called stimming.

56. Horseshoe kidneys can occur in Kabuki syndrome.

57. A heart murmur (abnormal sound) is a condition often seen in Kabuki syndrome.

58. Most children with Kabuki syndrome have a happy disposition.

59. Kabuki syndrome is found globally and is not limited to a specific race or ethnicity.

60. It is not uncommon for those with Kabuki syndrome to sleep with their eyes partially open.

61. Those born with Kabuki syndrome are generally born at a normal height and weight. Growth delays occur after birth.

62. Equine therapy can be used to help with conditions of Kabuki syndrome.

63. Sacral dimpling (a small depression in the skin, just above the buttocks) can occur with Kabuki syndrome.

64. Lax ligaments can be one of the conditions seen in Kabuki syndrome.

65. Strabismus (abnormal alignment) is an eye condition seen in Kabuki syndrome.

66. Fetal pads can be persistent, not only on fingertips but also on toes.

67. Ptosis (dropping upper eyelid) is a condition seen in Kabuki syndrome.

68. Long palpebral fissures (refers to the opening of the eye lid) are seen in Kabuki syndrome.

69. Aversions to certain textures is common in Kabuki syndrome.
Gastroesophageal reflux is common in the early years of Kabuki syndrome, but often improves with age.

Obsessive tendencies can be seen in Kabuki syndrome.

Hundreds of mutations have been identified with the KMT2D (MLL2) gene, where only a handful have been identified with the KDM6A gene. KMT2D is the more prevalent gene associated with Kabuki syndrome.

Abnormal development of the eustachian tubes can be seen in Kabuki syndrome, and can contribute to ear infections and hearing loss.

Dental abnormalities, such as the absence of permanent teeth, are common in Kabuki syndrome.

Those with Kabuki syndrome have a tendency to take language literally. They may not pick up on visual cues with facial expression or varying tones of voice (like sarcasm).

Those with Kabuki syndrome can have an excellent memory in certain areas (i.e. remembering song lyrics or grocery lists.)

An Individualized Education Plan (IEP) is an important tool used for children with Kabuki syndrome. It can be useful at all stages of education, including transitioning into adulthood.

Children with Kabuki syndrome are prone to repetitive behavior such as watching the same shows, reading the same book, singing the same song, asking the same question.

Those with Kabuki syndrome typically have loose ligaments. They can be very flexible.

Ectopic kidney is also found in Kabuki syndrome. This means one or both kidneys are not located where they should be.

Constipation and diarrhea can be issues in Kabuki syndrome.

Infants with Kabuki syndrome often have difficulty coordinating sucking and swallowing.

Idiopathic Thrombocytopenic Purpura can occur in Kabuki syndrome. It is a condition in which there is a low platelet count because platelets are being destroyed.

Certain noises can bother children with Kabuki syndrome. They may react by covering their ears or having panic like reactions.

As children with Kabuki syndrome develop and grow, behaviors such as autistic characteristics can be seen.

Malrotation of the intestines can occur in Kabuki syndrome.

Toe nails and finger nails may be a different thickness, and grow with an upward curve.

Mild webbing between fingers can occur with Kabuki syndrome.

Kabuki syndrome with KMT2D mutation is inherited autosomal dominate, which means one copy of the altered gene in each cell will cause the syndrome.

Sleep apnea can be an issue with Kabuki syndrome.

Autoimmune hemolytic anemia is another condition that can be seen in Kabuki syndrome. This means antibodies directed at red blood cells causes the life of the red blood cell to be reduced.
92. Some children with Kabuki syndrome will head rock. This can be done when experiencing emotions or to self soothe.

93. Fingerprints with unusual features can occur in Kabuki syndrome.

94. Obesity is common during the puberty years in Kabuki syndrome. This is a change from the early years when gaining weight is often difficult.

95. Low muscle tone is part of Kabuki syndrome. It does not mean there is a lack of muscle definition but rather a lack of stamina and ability to respond timely.

96. Abnormal sleep patterns can occur in Kabuki syndrome.

97. Sensory Processing Disorder is often seen in Kabuki syndrome. Aversions may include oral, touch, and noise.

98. In girls with Kabuki syndrome, early breast development can occur, and less common early puberty.

99. Nystagmus is an eye condition that can occur in Kabuki syndrome. It is an involuntary eye movement.

100. Kabuki syndrome itself does not shorten a persons life span.

101. Kabuki syndrome comes with many challenges but it brings with it, an abundance of joy and blessings!

SOURCES:
The majority of the facts listed in Kabuki 101 come directly from families raising children with Kabuki syndrome. Additional sources include:

www.rarediseases.org/rare-disease-information/rare-diseases/byID/920/viewFullReport
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