



# KABUKI SYNDROME 101

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1. Kabuki syndrome was first described in Japan in 1967 by Dr. Niikawa and in 1968 by Dr. Kuroki. Each identified unique malformations in several other patients over the next decade.

2. Kabuki syndrome became a formal diagnosis in 1981.

3. While some syndromes are linked to a chromosome abnormality, Kabuki syndrome is linked to a change at the gene level.

4. There were five cardinal traits originally used to clinically diagnose syndrome.

5. Kabuki syndrome affects approximately 1 in 32,000 births worldwide.

6. Mutations in two genes are currently identified as a cause for Kabuki syndrome: KMT2D (formerly known as MLL2) and KDM6A.

7. Kabuki syndrome was originally referred to as Kabuki make-up syndrome (KMS); Niikawa-Kuroki syndrome would be a second, alternate name. Kabuki make-up syndrome would later be shortened to Kabuki syndrome.

8. Individuals with Kabuki syndrome can have an excellent memory in certain areas, despite experiencing intellectual delays.

9. Seizures can occur in Kabuki syndrome.

10. One of the features of Kabuki syndrome is beautiful, long eyelashes and large eyes.

11. Flat block feet are a common Kabuki trait.

12. The first of five characteristics used to clinically diagnose Kabuki syndrome was skeletal abnormalities.

13. The second characteristic used to clinically diagnose Kabuki syndrome was dermatoglyphic (fingers, hands and feet) abnormalities.

14. The third characteristic used to clinically diagnose Kabuki syndrome was postnatal short stature.

15. The fourth characteristic used to clinically diagnose Kabuki syndrome was mild to moderate intellectual disabilities. Approximately 5% of individuals with Kabuki syndrome have no intellectual impairment.

16. The fifth characteristic that was used to clinically diagnose Kabuki syndrome was facial features. Facial features continue to be an important aspect in identifying patients with Kabuki syndrome. Individuals with Kabuki syndrome often have long palpebral fissures with eversion of the outer third, arched eyebrows with sparse outer half, prominent eyelashes, prominent or misshaped ears and a depressed nasal tip.

17. Persistent fetal pads on the fingers is a common trait found in Kabuki syndrome. It is considered a dermatoglyphic abnormality.

18. The scrunchy face is not a characteristic used for diagnosing, but a trait frequently seen in individuals with Kabuki syndrome.

19. 75% or more of individuals with Kabuki syndrome will have a mutation on the KMT2D (MLL2) gene.

20. If a person does not show a mutation on the KMT2D gene, it does not mean they do not have Kabuki syndrome. It may indicate that another gene is mutated.

21. Feeding issues are almost always an issue early in life.

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

23. Excessive drooling in the early years is common in Kabuki syndrome.

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

25. Renal (kidney) anomalies can occur in Kabuki syndrome.



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

27. Kabuki syndrome was first publicly referred to as Kabuki make-up syndrome because the elongated eyes resembling those seen in Japanese Kabuki theatre actors.

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

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

30. Hearing loss can be seen in those diagnosed with Kabuki syndrome. This may include the need for hearing aids.

31. Amblyopia (lazy eye) is a visual impairment that can occur in Kabuki syndrome. Amblyopia is not correctable with glasses or contacts and causes decreased vision.

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

33. Short digits (fingers and toes), especially the fifth digit, is common in Kabuki syndrome.

34. Precocious puberty can occur in Kabuki syndrome.

35. Individuals with Kabuki syndrome often present with autistic-like behaviors.

36. It is not uncommon for individuals with Kabuki syndrome to have an unusually high-pitched voice.

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

38. Approximately 5% of those diagnosed with Kabuki syndrome have mutations on the KDM6A gene.

39. Spina bifida occulta can also occur in Kabuki syndrome.

40. 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.

41. Gastroesophageal reflux is common in the early years of Kabuki syndrome, but often improves with age.

42. Physical Therapy is often needed with Kabuki syndrome.

43. Children with Kabuki syndrome often learn to walk much later than typical children. ATK's parent survey suggests the average age to be two.

44. Speech Therapy is usually needed with Kabuki syndrome.

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

46. It is not uncommon for children with Kabuki syndrome to have a high arched palate.

47. Scoliosis is another condition that can be seen in those with Kabuki syndrome.

48. The KMT2D gene is located on the long arm of chromosome 12.

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

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

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

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

53. Individuals with Kabuki syndrome typically have loose ligaments. They tend to be very flexible.

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



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55. Horseshoe kidneys can occur in Kabuki syndrome.

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

57. Cleft palate abnormalities can also be part of Kabuki syndrome. They can affect speech, eating and even hearing.

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

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

60. Most individuals with Kabuki syndrome have a happy disposition.

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

62. It is not uncommon for those with Kabuki syndrome to sleep with their eyes partially open. This is called nocturnal lagophthalmos.

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

64. Equine Therapy can be used to help with conditions of Kabuki syndrome.

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

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

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

68. Ptosis (drooping upper eyelid) is a condition seen in Kabuki syndrome.

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

70. Aversions to certain textures is common in Kabuki syndrome.

71. Obsessive tendencies can be seen in Kabuki syndrome.

72. Malrotation of the intestines can occur in Kabuki syndrome.

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

74. Abnormal development of the eustachian tubes is often seen in Kabuki syndrome. This can contribute to ear infections and hearing loss.

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

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

77. 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.

78. 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 questions, etc.

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

80. Constipation and diarrhea are common issues seen in Kabuki syndrome.

81. Mild webbing between fingers can occur in Kabuki syndrome.

82. Infants with Kabuki syndrome often have difficulty coordinating the suck, swallow, breathe process.



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83. Sleep apnea can be an issue in Kabuki syndrome.

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

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

86. Children with Kabuki syndrome tend to talk later than usual. The average age Kabuki kids talk, according to parent data on ATK's survey, is four years old.

87. Toe and fingernails may be of a different thickness and grow with a sharp, upward curve.

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

89. Abnormal sleep patterns can occur in Kabuki syndrome.

90. Autoimmune hemolytic anemia is another condition that can be seen in Kabuki syndrome. This means antibodies directed at red blood cells cause the life of the red blood cell to be reduced.

91. Low muscle tone (hypotonia) 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.

92. Males with the KDM6A mutation are more severely affected than females.

93. In girls with Kabuki syndrome, early breast development can occur, and less commonly early puberty.

94. Individuals with Kabuki syndrome have been known to have an unusually high pain threshold.

95. Spatial awareness issues are seen in Kabuki syndrome.

96. Anxiety is often seen in individuals with Kabuki syndrome. First reported in older children, parents are reporting signs of anxiety much earlier than usual.

97. Early intervention is the key to ensuring a child with Kabuki syndrome has the resources needed to overcome the obstacles that often come with Kabuki syndrome.

98. Kabuki syndrome is considered a spectrum disorder. While the majority of individuals share a mutation in the same gene (KMT2D, the variant is often different.

99. Kabuki syndrome affects each individual differently.

100. Eczema and keratosis pilaris are commonly reported in Kabuki syndrome.

101. Kabuki syndrome itself does not shorten an individual's life span, however underlying conditions may.

*Thank you for taking the time to learn about Kabuki syndrome. This document was produced for educational purposes only. Always consult a medical professional for advice. \* We invite you to join our efforts to advocate for Kabuki syndrome and those affected by this rare genetic disorder. Additionally, we encourage families to participate in research efforts as they become available. Together we can make a difference and affect change for the future for our children!*



SOURCES: Most of the facts listed in Kabuki 101 come directly from parents raising children affected by Kabuki syndrome. Additional sources include: [rarediseases.org](http://rarediseases.org), [nih.gov](http://nih.gov), and [kabukisynndrome.com](http://kabukisynndrome.com).