# Congenital Anomalies in Ancient Japan as Deciphered in the *Nihon shoki* (Chronicles of Japan)

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#### Introduction

Congenital anomalies are structural or functional abnormalities that occur during prenatal development. According to the World Health Organization (WHO), an estimated 6 percent of newborns have some congenital abnormality, resulting in hundreds of thousands of related deaths. Systematic studies of human congenital anomalies started in the 1960s, triggered by the thalidomide cases. At that time, as many as 10,000 cases were reported worldwide, and the risk of congenital morphological abnormalities stemming from drugs taken by the mother during pregnancy became known. This led to research on cases of congenital anomalies and their factors. In recent years, with the progress of developmental biology, molecular biology, and genome analysis techniques, a host of both environmental and genetic factors have now been identified that can cause various congenital abnormalities.

Before congenital anomalies were systematically studied, the epidemiological status of human congenital anomalies (the kinds of cases and the extent to which these occurred across a population) could be ascertained only by collecting case reports published in medical journals. For example, the earliest case of the human tail—a congenital anomaly in which a newborn possesses an excess of tail-like structures—was reported in 1884 in the *New England Journal of Medicine*.<sup>2</sup> Similarly, other malformations either characterized by a striking appearance or

<sup>&</sup>lt;sup>1</sup> See the WHO website: <a href="https://www.who.int/health-topics/congenital-anomalies">https://www.who.int/health-topics/congenital-anomalies</a>.

<sup>&</sup>lt;sup>2</sup>Tojima and Yamada, "Classification of the 'Human Tail."

associated with fatality have also been reported in medical journals. However, the number of cases caused by postnatal disease or by accidents is generally much larger than those possibly caused by the prenatal situation. Thus, it was difficult to grasp the epidemiological status of congenital anomalies even after the 1800s. For this reason, it has been challenging to elucidate the actual situation of congenital anomalies before the spread of modern medical science. This study, however, demonstrates the potential of historical documents to help us in understanding the epidemiology of congenital anomalies in ancient times, using Japan as an example.

As in other countries, systematic study of human congenital anomalies in Japan started in the 1960s, and modern medical journals began to be published only in the late 1870s. Consequently, it was challenging to grasp the degree of incidence or the kinds of congenital abnormalities in pre-modern Japan.

However, numerous ancient historical documents are preserved in Japan. Careful reading of these historical sources reveals several descriptions suggesting congenital morphological or functional abnormalities. The oldest official historical chronicle that survives in Japan is the *Nihon shoki* 日本書紀, which is said to have been completed in 720. It is a chronological text whose compilation was ordered by the emperor (tennō 天皇)—the world's longest-running imperial family. The Nihon shoki consists of a mythological part as well as a record of the genealogy and achievements of forty-one generations of emperors, from the first emperor, Jinmu 神武, to Emperor Jitō 持統. It is written in Literary Sinitic and in annalistic style, with years and dates during each emperor's reign being recorded according to the Chinese sexagenary calendrical cycle.

The *Nihon shoki* is considered to contain more legendary elements than the five subsequent official national histories. In particular, post-World War II historiography tended to reject pre-war studies and thought, and several historians proposed theories questioning the historicity of all accounts in the *Nihon shoki*. However, recent archaeological excavations of the ancient capital and research on wooden documents called *mokkan* 本簡 have shown that the descriptions in the *Nihon shoki* (not the entirety but a part of its descriptions at least) accurately reflect what actually happened.<sup>3</sup> The existence of the emperors mentioned in this document has been proven by the archaeological record as well. An inscription on an iron sword excavated from the Inariyama 稲荷山 burial mound in 1968, for example, proves the existence of the twenty-first emperor, Yūryaku 雌略,<sup>4</sup> which means that the *Nihon shoki* is not a work of fiction but a chronicle of actual events. Many descriptions in this document, especially those recounting events after the seventh century, have been verified by the findings of such archaeological excavations.

<sup>&</sup>lt;sup>3</sup> Endō, Rikkokushi.

<sup>&</sup>lt;sup>4</sup> Hayakawa et al., "Saitama Inariyama kofun."

The *Nihon shoki* is an important historical document that plays a central role in understanding Japanese history; and it is also highly valuable from the perspective of East Asian history. Therefore, it has been a subject of study mainly in history fields. Noteworthily, it contains many descriptions of natural phenomena, including records of disasters (such as earthquakes, heavy rain, drought, and storms) and accounts of astronomical events (such as meteors, planetary motions, and solar or lunar eclipses). Recently, some of its astronomical records have been corroborated by modern science,<sup>5</sup> suggesting that these details were not incorporated as fictional creations but were duly recorded as important matters related to the functioning of the nation. The *Nihon shoki* also contains numerous descriptions of humans: not only of members of the imperial family and people subordinate to them, but even of the people opposed to the emperor. Some of these descriptions possibly refer to congenital anomalies.

While the *Nihon shoki* contains medically valuable descriptions as noted above, it cannot be treated in the same way as ordinary medical records. This is because it is a historical document based on the social background at that time, and its contents must be handled with a strict critical awareness. The historical background and intentions of the period in which the text was composed, along with the degree of truth and exaggeration in its descriptions, must always be considered, instead of simply understanding those descriptions as facts.

Thus, this study attempted to elucidate the epidemiology of congenital anomalies in ancient Japan—which to date has remained largely unexplored—through a cross-disciplinary approach combining medical and historical perspectives.

#### Materials and Methods

## The Nihon shoki Used in This Study

No original copy of the *Nihon shoki* has survived to the present. Nonetheless, since it has been considered an important record of national history since its completion, its contents have been repeatedly hand-copied at the imperial court, and over the years many manuscript copies have been produced; most of these are designated as National Treasures or Important Cultural Properties of Japan.

In this study, we used the edition of the *Nihon koten bungaku taikei* 日本古典 文学大系 series as the main source text for our reading and data collection. Additionally, taking into account the possibility that some characters may have been transcribed incorrectly in the process of hand-copying or been damaged with age (through deterioration or by insect bite), we have referred also to as many major extant manuscript copies as possible, to confirm the text of those descriptions we identified as potentially representing congenital anomalies (for a list of the manuscript copies of the *Nihon shoki* referenced in this study, see **Table 1**).

<sup>&</sup>lt;sup>5</sup> Tanikawa, Sōma, and Qu, "Short-Term Variations."

<sup>&</sup>lt;sup>6</sup> Sakamoto et al., Nihon shoki, vols. 1–2.

Table 1. Copies of the Nihon shoki Referenced in This Study

	日本古典文学大系 (底本:卜部兼方本)	岩崎本	前田家本	図書寮本	熱田本	北野本
	Nihon Koten Bungaku Taikei (based on Urabe Kanekata's copy)	Iwasaki bon	Maeda-ke bon	Zushoryō bon	Atsuta bon	Kitano bon
	13c	10–11c	11c	12c	14c	14–16c?
Jinmu 神武 (1st)	0				0	0
Suinin 垂仁 (11th)	$\circ$				$\bigcirc$	$\bigcirc$
Keikō 景行 (12th)	$\circ$				$\bigcirc$	$\bigcirc$
Chūai 仲哀 (14th)	$\bigcirc$				$\bigcirc$	$\bigcirc$
Empress Jingū 神功皇后 (14.5th)	0				$\circ$	$\circ$
Ōjin 応神 (15th)	$\circ$			$\bigcirc$	$\bigcirc$	$\bigcirc$
Nintoku 仁徳 (16th)	$\bigcirc$		$\bigcirc$			$\bigcirc$
Hanzei 反正 (18th)	$\bigcirc$			$\bigcirc$	$\bigcirc$	$\bigcirc$
Yūryaku 雄略 (21st)	$\bigcirc$		$\bigcirc$	$\bigcirc$	$\bigcirc$	
Seinei 清寧 (22nd)	$\bigcirc$			$\bigcirc$	$\bigcirc$	$\bigcirc$
Buretsu 武烈 (25th)	$\bigcirc$			$\bigcirc$		$\bigcirc$
Kinmei 欽明 (29th)	$\bigcirc$					0
Bidatsu 敏達 (30th)	$\circ$		$\bigcirc$			$\bigcirc$
Suiko 推古 (33rd)	$\circ$	$\bigcirc$		$\bigcirc$		$\bigcirc$
Kōgyoku 皇極 (35th)	$\circ$	$\bigcirc$		$\bigcirc$		$\bigcirc$
Seimei 斉明 (37th)	$\circ$					$\bigcirc$
Tenji 天智 (38th)	$\circ$					$\bigcirc$
Tenmu 天武 (40th)	$\bigcirc$					$\circ$
Jitō 持統 (41st)	$\circ$					$\circ$

Note: For bibliographic information on each of the referenced manuscripts, see the reference list.

Quotations of the *Nihon shoki* below have been excerpted from the main source text. In cases where the quoted passage included Chinese characters in traditional form, we have changed these to their modern equivalents for convenience. This article contains many emperors' names as well as other Japanese names and terms. For these, the original characters have been supplied, and their readings given as well, for example, Emperor Jinmu 神武. Additionally, in this article, we use the term "Emperor" for any person who is described as having ascended to the throne as *tennō*, regardless of his or her biological sex. At the same time, the term "Empress" is not used to refer to a female emperor, but rather to a woman who is described as the legitimate wife of a male emperor.

#### Data Collection from the Nihon shoki

As for the existence of the emperors mentioned in the *Nihon shoki*, there are several theories and it is particularly difficult to verify the existence of the emperors preceding the twenty-first emperor, Yūryaku, from an archaeological perspective. It is therefore also challenging to positively confirm or deny, at this point, the historical veracity of the *Nihon shoki'*s descriptions, and discussions of the question fall outside the scope of this article. Since the purpose of this study was to analyze descriptions of human congenital anomalies, the mythological section of the work was excluded. The analysis in this study covered the period from the first emperor, Jinmu, to the 41st emperor, Jitō, an era whose emperors are clearly identified as being humans rather than gods in the *Nihon shoki*.

We collected descriptions of humans possessing abnormal physical and mental characteristics. Cases clearly describing postnatally-acquired diseases (e.g., the cases described as 病, meaning "illness") were excluded from this analysis. Based on the information available in each case, the possibility of congenital anomalies was examined, and a diagnosis of the disease was attempted to the extent possible.

### Case Diagnoses: From Medical and Historical Points of View

The *Nihon shoki* is not a medical text but a historical document recording the national history of Japan. As background to the compilation of the *Nihon shoki*, the threat of the Chinese empire, which at the time exercised a powerful hegemony in the East Asian region, needs to be underlined. In the second year of the reign of the thirty-eighth emperor, Tenji (Tenchi) 天智, Japan suffered a major defeat in a battle against the combined forces of Tang and Silla in the Korean peninsula (i.e., the battle of Baekgang 白江, Jp. Hakusukinoe 白村江, 663). The changing balance of power, both on the continent and in the Korean peninsula, significantly impacted the formation of the *Nihon shoki*. All the statements it contains, in other words, must be carefully examined to assess whether they offer us fact, exaggeration, or metaphor. Accordingly, this study (a) conducted a medical analysis, to the extent possible, to determine what the names and etiologies of the diseases would be if the statements represented actual symptoms and (b) also undertook a consideration of the historical record to determine whether these various diseases could, in fact, have actually occurred.

### Records of Phenomena Possibly Related to Maternal Nutritional Status

Regarding the relationship between maternal nutritional status and congenital anomalies, a deficiency in folic acid is well-known to be directly related to neural tube defects.<sup>7</sup> However, most studies to date have focused on the relationship between individual nutrients and disease, and not much research has been focused

<sup>&</sup>lt;sup>7</sup>Smithells, Sheppard, and Schorah, "Vitamin Deficiencies"; Smithells et al., "Apparent Prevention of Neural Tube Defects."

on multiple nutrients, energy levels, and birth defects. In the longer term (not in terms of birth defects immediately after birth, but in terms of fetal growth retardation [FGR]), "Barker's Hypothesis," stating that maternal undernutrition during pregnancy increases the incidence of future adult diseases in the child, is a plausible effect of undernutrition.

In ancient societies, epidemics, crop failures, and famines due to climatic irregularities and disasters were considered to be situations directly related to the deterioration of maternal nutritional status. Since these events were emergencies in the management of the nation, several clear descriptions of them are given in the *Nihon shoki*. Therefore, we examined the possibility of a chronological correlation between the timing of these events and various descriptions of what might be thought to be congenital anomalies. We also collected and summarized data on the occurrence of disasters such as climatic irregularities (extreme temperatures [cold summers, warm winters, hail, etc.], abnormal rainfall [drought, long rains], typhoons, and earthquakes) as possible causes of the deterioration of nutritional status, even if the character \$\mathbb{I}\$ (meaning "starved") is not explicitly mentioned.

#### Results

Among the records for forty-one generations of emperors, a total of thirty-three cases of human abnormalities were found (**Table 2**). Except during the period stretching from the reign of the second emperor, Suizei 綏靖, to the that of the ninth emperor, Kaika 開化—known as the "generations without history" for their lack of detailed descriptions—descriptions regarding abnormalities were found almost universally, and there was no significant chronological bias in their distribution. These thirty-three cases, the subject of this study, included descriptions of the emperors themselves, of members of their families, and also of non-imperial people. These cases can be classified into the following five types.

## Type 1: Abnormal Height

This type appeared most frequently in the *Nihon shoki*. It could be divided into two patterns: unusually tall and unusually short statures. Descriptions of unusually tall stature were seen only for members of the imperial family (four out of ten cases). In two of these cases, there were specific descriptions of the person's stature. Yamato Takeru no Mikoto日本武尊, who was a son of the twelfth emperor, Keikō 景行, is described as being about 3 meters (1  $j\bar{o}$  文) tall. His son, the fourteenth emperor, Chūai 仲哀, is also described as a tall person, 3 meters (10 *shaku* 尺) in height. An indirect description indicates that Emperor Yūryaku

<sup>&</sup>lt;sup>8</sup> Abu-Saad and Fraser, "Maternal Nutrition and Birth Outcomes."

<sup>&</sup>lt;sup>9</sup> Barker, "Fetal Origins of Cardiovascular Disease."

<sup>&</sup>lt;sup>10</sup> Sakamoto et al., Nihon shoki, vol. 1, pp. 282–283.

<sup>&</sup>lt;sup>11</sup> Ibid., pp. 320-321.

	Number of Cases			
	Imperial	Non-imperial	Total	
Type 1: Abnormal height	4	6	10	
-Abnormally tall	4	0	4	
-Abnormally short	0	6	6	
Type 2: Excess tissue or organ formation	1	5	6	
Type 3: Speech or behavioral abnormality	4	2	6	
Type 4: Different facial or body feature	0	6	6	
Type 5: Pigment anomaly	2	1	3	
Other	0	2	2	

Table 2. Possible Cases of Congenital Anomalies in the Nihon shoki

was also tall. In the scene where this emperor encounters a deity named Katsuragi no Hitokotonushi 葛城一言主, the height of the deity is described as being tall in a manner similar to that of the emperor.<sup>12</sup>

Meanwhile, descriptions of unusually short stature were found only in the non-imperial peoples (six out of ten cases). The earliest description of short stature is about the people called Tsuchigumo 土蜘蛛, in the chronicle of Emperor Jinmu's reign. According to this description, the Tsuchigumo had short bodies and long limbs similar to the Hikihito 侏儒.<sup>13</sup> The term Hikihito refers not to specific individuals but rather to a so-called "dwarf" people. In the six descriptions of short stature, four cases related to them. The term 侏儒 also appears in the Chinese historical work Records of the Three Kingdoms (Ch. Sanguozhi 三国志; in the "Book of Wei" (魏書), within the chapter "Encountering the Dongyi" (烏丸鮮卑東夷伝), "Section on the Wa 倭 people," commonly known in Japanese as "Gishi wajinden" 魏志倭人伝), to which the original editors of the Nihon shoki themselves referred. 4 According to this Chinese work, the Hikihito people were about 90-120 centimeters (3-4 shaku) tall. Their occupation can also be inferred from the descriptions in the Nihon shoki. In the chronicles of the twenty-fifth emperor, Buretsu 武烈, and the fortieth emperor, Tenmu 天武, onward, the Hikihito people are described as participating in a number of recreation activities held by the emperors. 15

There was also a description of a such a "dwarf" person not of the Hikihito people. During the reign of Emperor Tenji, the chronicle mentions Nakatomibe no Wakako 中臣部若子 of Hitachi 常陸 Province (present-day Ibaraki Prefecture), who is said to have been only about 48 centimeters (1 *shaku* and 6 *sun*) tall at the

<sup>&</sup>lt;sup>12</sup> Sakamoto et al., Nihon shoki, vol. 1, pp. 466–467.

<sup>&</sup>lt;sup>13</sup> Ibid., pp. 210–211.

<sup>&</sup>lt;sup>14</sup>Chen and Pei, Sangokushi, vol. 30; Sakamoto et al., Nihon shoki, vol. 1, pp. 350–353.

<sup>&</sup>lt;sup>15</sup> Sakamoto et al., *Nihon shoki*, vol. 2, pp. 16–17, 416–417, 460–461.

age of sixteen and who was presented to the emperor, perhaps because of his or her unusual stature.<sup>16</sup>

# Type 2: Excess Tissue or Organ Formation

Among the descriptions of excess tissue or organ formation, only one case related to the emperor himself. This is the fifteenth emperor, Ōjin 応神, who is described as congenitally having excess tissue as follows: at birth, he had raised flesh on his arms, shaped like a *tomo* 鞆—a device worn on the left forearm when using a bow and arrow (**Figure 1**).<sup>17</sup> The *Nihon shoki* also notes that Emperor Ōjin had a long reign and a long life<sup>18</sup>, and these descriptions strongly suggested a lack of other severe morphological or functional abnormalities.

Other than the one imperial case, there were five descriptions of non-imperial individuals with various kinds of excess tissue or organs. The earliest such records appeared in the chronicle of Emperor Jinmu's reign. The emperor is said to have encountered two people with tails in the Yoshino 吉野 region (in present-day Nara Prefecture). The first of these was named Ihika 井光 and is described as the founder of a tribe living in the region. This person is also described as having emerged from a well (*i* 井) and as possessing a body that glowed. The second person with a tail introduced himself to the emperor as Ishioshiwaku no Ko 磐排別之子 and appeared in front of the emperor after pushing his way through a huge rock. This tailed person is also described as the founder of a local tribe in the Kuzu 国栖 district of the Yoshino region.

In the reign of the eleventh emperor, Suinin 垂仁, there is a description of a man with a horn (or horns) on his forehead, Tsunuga Arashito 都怒我阿羅斯等.<sup>20</sup> He was the son of the king of the Kara 韓 kingdom on the Korean peninsula, and is said to have come to Japan by ship during the reign of the tenth emperor, Sujin 崇神, eventually serving two emperors.

While the people in the three cases above were friendly to the court and the emperor, the following two cases describe forces opposed to the imperial court. In the first year of Empress Jingū's 神功皇后 regency (after the death of her husband, Emperor Chūai 仲哀), there was a man named Hashirokumawashi 羽白熊鷲 in present-day Kyushu.<sup>21</sup> He was robust, had wings, and could fly high. The imperial court had him subdued because he was a violent robber and disobeyed imperial orders. A similar case is recorded in the sixty-third year of the reign of the sixteenth emperor, Nintoku 仁徳—the court subdued a person named Sukuna 宿儺 in present-day Gifu Prefecture for similarly disobeying imperial commands.

<sup>&</sup>lt;sup>16</sup> Sakamoto et al., Nihon shoki, vol. 2, pp. 376–377.

<sup>&</sup>lt;sup>17</sup> Ibid., vol. 1, pp. 362–363.

<sup>&</sup>lt;sup>18</sup> Ibid., pp. 380–381.

<sup>&</sup>lt;sup>19</sup> Ibid., pp. 198–199.

<sup>&</sup>lt;sup>20</sup> Ibid., pp. 258-259.

<sup>&</sup>lt;sup>21</sup> Ibid., pp. 332–333.

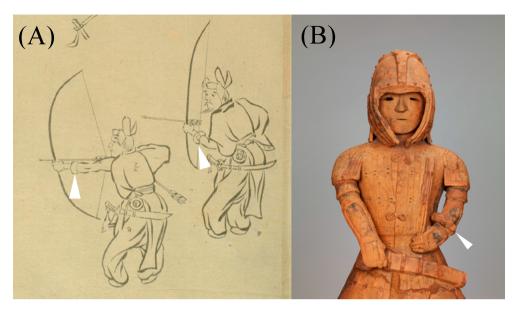


Figure 1. Depictions of tomo 鞆 (indicated by white arrowheads) worn on the left forearm. (A) Nenjū gyōji emaki 年中行事絵巻, by Fujiwara no Mitsunaga 藤原光長, copied by Tani Bunchō 谷文晃. National Diet Library Digital Collection. <a href="https://doi.org/10.11501/2591106">https://doi.org/10.11501/2591106</a> (image 14). (B) Haniwa Keikō no bujin 埴輪 挂甲の武人 (terracotta tomb figure of an armored man). Tokyo National Museum. e-Museum: National Treasures & Important Cultural Properties of National Institutes for Cultural Heritage, Japan. <a href="https://emuseum.nich.go.jp/detail?langId=&content\_base\_id=100200&content\_part\_id=001&content\_pict\_id=033">https://emuseum.nich.go.jp/detail?langId=&content\_base\_id=100200&content\_part\_id=001&content\_pict\_id=033</a>.

Sukuna is described as possessing one body but two faces dorsoventrally. The top of the head was fused and he had no nape, and there were eight limbs (the number of the limbs was enough for two people); he had knees but no popliteal fossa.<sup>22</sup>

# Type 3: Speech or Behavioral Abnormality

This type was found in six cases: four were about members of the imperial family, and two were about non-imperials. All involve functional abnormalities, but the cases can be divided into those of speech impairment and those of behavioral disability.

Two cases of speech impairment were found in the imperial family. Homutsuwake no Miko 營津別王 (命, 皇子) the son of Emperor Suinin, is said to have cried like a baby and been unable to speak even though he was thirty years old with a long beard. Later, however, he apparently developed the ability to speak when he became interested in a swan.<sup>23</sup> Something similar was seen in the case of Takeru no

<sup>&</sup>lt;sup>22</sup> Sakamoto et al., Nihon shoki, vol. 1, pp. 414–415.

<sup>&</sup>lt;sup>23</sup> Ibid., pp. 256–257, 267–269.

Miko 建皇子, the son of Emperor Tenji, of whom it is clearly stated that "he could not speak and was dumb [sic]". He died prematurely at the age of eight.<sup>24</sup>

Descriptions of behavioral abnormality referred specifically to the tendency to become easily incensed and use violence. Emperor Yūryaku is described as often killing people based on misunderstandings, and as having once been condemned as "a very bad emperor" by the people. However, he improved his behavior after being admonished by the empress. In the chronicle of Emperor Buretsu, a grandson of Yūryaku, there are more descriptions of violent tendences, and those were not for the execution of criminals but rather for pleasure: incidents such as ripping open the belly of a pregnant woman to see her fetus, or making people climb trees and then shooting them down. Although this emperor had an empress, he had no sons or daughters. There is no clear statement in the Nihon shoki about Buretsu's age at his death, and there are various theories based on descriptions in later history books. The Fusō ryakki 扶桑略記 and the Mizukagami 水鏡 from the eleventh to twelfth century state that he died at the age of eighteen, the reiō hennenki 帝王編年記 and the Kōdaiki 皇代記 from the fourteenth century describe his age at death as fifty-seven. The second second

Another such description related to an individual who was a royal but not a member of the Japanese imperial family: King Mata 末多 of Paekche on the Korean peninsula, who was recommended by the aforementioned Emperor Yūryaku to become a king, is also described as a tyrant who mistreated his people.<sup>30</sup>

Among this type, only one case was about a non-imperial person. In the fourth year of the reign of Emperor Tenmu, a certain man is said to have climbed up a hill, uttered something mysterious and thereupon died after beheading himself.<sup>31</sup>

#### Type 4: Different Facial or Body Features

Of the thirty-three total cases, six were of type 4. These all concerned non-imperial people living either in remote areas controlled by the imperial court at the time or even further beyond, whose facial features appeared to be "different." This type of description first appears in records of the reign of Emperor Keikō. There it is described how he launched a large-scale expedition to the Kyushu area because several tribes there called Kumaso 熊襲 or Tsuchigumo had not paid tribute. During the expedition, the emperor is said to have killed two Kumaso

<sup>&</sup>lt;sup>24</sup> Sakamoto et al., *Nihon shoki*, vol. 2, pp. 331, 368–369.

<sup>&</sup>lt;sup>25</sup> Ibid., vol. 1, pp. 464–465.

<sup>&</sup>lt;sup>26</sup> Ibid., vol. 2, pp. 14–17.

<sup>&</sup>lt;sup>27</sup> Higo et al., "Kunchū Fusō ryakki 3."

<sup>&</sup>lt;sup>28</sup> Kuroita, Fusō ryakki, Teiō hennenki; Kuroita, Mizukagami, Ōkagami.

<sup>&</sup>lt;sup>29</sup> Kuroita, Fusō ryakki, Teiō hennenki; Hanawa, Gunsho ruijū 3.

<sup>&</sup>lt;sup>30</sup> Sakamoto et al., *Nihon shoki*, vol. 1, pp. 497–499, vol. 2, pp. 14–15.

<sup>&</sup>lt;sup>31</sup> Ibid., vol. 2, pp. 420–421.

leaders named Hanatari 鼻垂 and Mimitari 耳垂 and a Tsuchigumo leader named Tsutsura 津頬.<sup>32</sup> These were all personal names but also describe physical characteristics: Hanatari and Mimitari respectively mean "large, drooping nose" and "large, drooping ears," while Tsutsura means "spiky face."

During the reign of the twenty-ninth emperor, Kinmei 欽明, there is a description of the people from the Mishihase 粛慎 region, an area to the north of ancient Japan's (i.e., the imperial court's) sphere of influence at that time, as "demons (oni 鬼魅), not people (hito ni arazu 非人)."<sup>33</sup> The following thirtieth emperor, Bidatsu 敏達, summoned a person named Ayakasu 綾柏. The leader of the Emishi 蝦夷 people, a tribe inhabiting the region to the north of the area controlled by the court, he had been brought to court because thousands of Emishi were ravaging the frontier. Ayakasu is described as an ōemishi 大毛人, the Chinese characters for which mean "hairy person."<sup>34</sup> The more definitive description of Emishi as having a face or appearance different from that of the people at court can be found in the chronicle of the thirty-seventh emperor, Saimei 斉明. In the fifth year of this emperor's reign, Japanese envoys sent to the Tang dynasty in China presented two Emishi people (a man and a woman) to the Emperor Gaozong 高宗. The Tang emperor is to have commented that he found the faces and bodies of the Emishi very strange.<sup>35</sup>

# Type 5: Pigment Anomaly

There were three descriptions of what appear clearly to be pigmentary anomalies. Two of the three cases related to the imperial family. The most obvious case was that of the twenty-second emperor, Seinei 清寧. It is clearly stated that the emperor was born with silver hair.<sup>36</sup> He seems not to have an empress or children.

Indirect description of hair streaked with gray was found regarding Yamashiro no Ōe no Miko 山背大兄王 during the reign of the thirty-fifth emperor, Kōgyoku 皇極. He was the son of a great regent named Umayato no Toyotomimi no Miko 厩戸豊聡耳皇子 and a grandson of the thirty-first emperor, Yōmei 用明. He was eventually attacked by a minister named Soga no Iruka 蘇我入鹿 in a dispute over the succession to the throne, and this attack led him to suicide. A satirical rhyme recorded in the *Nihon shoki* uses the term *kamashishi* 山羊 to refer to the prince.<sup>37</sup> The word is thought to refer to the Japanese serow, whose fur is streaked with gray.

Another description of a supposed pigmentary abnormality of non-imperial

<sup>&</sup>lt;sup>32</sup> Sakamoto et al., *Nihon shoki*, vol. 1, pp. 288–289, 294–295.

<sup>&</sup>lt;sup>33</sup> Ibid., vol. 2, pp. 91–93.

<sup>&</sup>lt;sup>34</sup> Ibid., p. 141.

<sup>&</sup>lt;sup>35</sup> Ibid., pp. 340–341.

<sup>&</sup>lt;sup>36</sup> Ibid., vol. 1, pp. 502–503.

<sup>&</sup>lt;sup>37</sup> Ibid., vol. 2, pp. 252–253.

people was found in the twentieth year of the reign of the thirty-third emperor, Suiko 推古. People who had come to Japan from Paekche were said to have had vitiligo on their faces and bodies. They were almost marooned on an island in the sea because of their appearance, but they were allowed to come to Japan after they appealed, citing their special skills in building landscapes.<sup>38</sup>

Other: Descriptions of Disability Possibly Including Cases of Congenital Anomalies
The expression atsuehito 篤癃 appears twice in the chronicle of Emperor Jitō.<sup>39</sup>
This term corresponds to tokushitsu 篤疾, a category of disabled people in the ritsuryō 律令 code, a set of laws enacted around the time the Nihon shoki itself was compiled (ritsu 律 refers to criminal law, ryō 令 to administrative law). Generally, the term ritsuryō contains both the Taihō 大宝 Ritsuryō enacted in 701–702 and the Yōrō 養老 Ritsuryō (a revision of the former Taihō Ritsuryō) enacted in 757. In the ritsuryō system, there were three categories of disability, based on severity: zanshichi 残疾, haishichi 癈疾, and tokushitsu 篤疾. Tokushitsu referred to people with critical illnesses such as leprosy, epilepsy, disability in two of four limbs, or blindness in both eyes, and such people were exempted from all duties and given one attendant to care for them. 40 Although it is impossible to determine the specific symptoms and cause of disease from a description of atsuehito alone, some conditions so categorized may have been due to congenital abnormalities.

#### Discussion

In the *Nihon shoki*, we found numerous descriptions that might be attributed to congenital anomalies. For each of the abovementioned types 1–5, we examined whether the cases surveyed could have been caused by a congenital abnormality and tried to diagnose the disease.

## Type 1: Abnormal Height

In the *Nihon shoki*, three emperors were mentioned as being tall. Similarly, in the *Kojiki* 古事記, a historical text compiled around the same time as the *Nihon shoki*, there are additional descriptions of two further tall emperors: Emperor Keikō (about 3.6 meters) and Emperor Hanzei 反正, the eighteenth emperor. Combining the descriptions in both texts, we might conclude that Emperor Keikō, Yamato Takeru no Mikoto, and Emperor Chūai represented three generations of tall height, and that such height was thus likely a genetic trait. However, the heights given for these emperors are too high to be factual. Since the specific figures themselves are unlikely to be accurate, it is more realistic to consider

<sup>&</sup>lt;sup>38</sup> Sakamoto et al., *Nihon shoki*, vol. 2, pp. 197–199.

<sup>&</sup>lt;sup>39</sup> Ibid., pp. 500–501, 532–533.

<sup>&</sup>lt;sup>40</sup> Kishi, Ryō no shūge.

<sup>&</sup>lt;sup>41</sup> Kurano and Takeda, Kojiki, Norito, pp. 188–189, 288–289.

them as exaggerations of the actual height. It is worth noting that descriptions of the height of an emperor are usually accompanied by expressions praising their appearance. Furthermore, since the emperors do not appear with the depictions of short stature described below, focus upon their tall stature may be a way of expressing prestige, fighting prowess, or esteem. It is also possible that the descriptions represent a combination of actual tall stature and exaggeration.

Medically, unusual tallness is sometimes caused by congenital anomalies. For example, Klinefelter syndrome, a sex chromosome abnormality, is one possible cause. <sup>42</sup> In the emperors' cases, however, all the tall emperors are described as having multiple children. Thus, it is highly likely that their tall height resulted not from a congenital abnormality but rather from a genetic trait. The only exception would be Emperor Chūai, who is said to have died suddenly at the age of fifty-two.

Such an association of tall stature and sudden death reminded us of another causative congenital anomaly: Marfan syndrome. 43 This is a genetic disease, and tallness is its characteristic symptom. Additionally, in affected adults, cardiovascular complications and sudden death are highly likely due to aortic dissection.<sup>44</sup> Furthermore, this anomaly is also associated with poor eyesight due to myopia and lens deviation. 45 In the chronicle of Emperor Chūai, one account states that the emperor could not see and did not recognize the existence of a certain "country on the sea" that a deity wanted to show him. 46 The emperor died shortly after this event, with the chronicle indeed attributing his early death to his failure to heed the deity's words. Of course, it is evident that such a narration contains mythical elements; and it would be practically difficult to see the Korean peninsula with the naked eye from atop a high hill near Kashii no Miya 橿日宮 Palace (in present-day Fukuoka Prefecture), where the emperor is said to have been at the time. However, that another country existed beyond the sea was, setting aside the deity, probably a fact known to the people of that time. It is highly likely that there were already political, economic, and cultural exchanges between Japan and the southern Korean peninsula in the third century, when Emperor Chūai is believed to have reigned. To take one indicative example, a series of excavations since the 1990s<sup>47</sup> have uncovered a group of ancient "keyhole-shaped" burial mounds (zenpō-kōen-fun 前方後円墳) in the southern part of the Korean peninsula. Burial mounds of such shape were characteristic of ancient Japan at

<sup>&</sup>lt;sup>42</sup> Klinefelter, Reifenstein, and Albright, "Syndrome Characterized by Gynecomastia"; Ford et al., "The Chromosomes in a Patient"; Jacobs and Strong, "A Case of Human Intersexuality."

<sup>&</sup>lt;sup>43</sup> Marfan, "Un cas de déformation congénitale."

<sup>&</sup>lt;sup>44</sup> de Beaufort et al., "Aortic Dissection in Patients"; Flynn et al., "Systematic Review and Meta-Analysis"; Pyeritz, "Etiology and Pathogenesis."

<sup>&</sup>lt;sup>45</sup> Jones, Rodriguez, and Bassnett, "Targeted Deletion of Fibrillin-1."

<sup>&</sup>lt;sup>46</sup> Sakamoto et al., Nihon shoki, vol. 1, pp. 326–327.

<sup>&</sup>lt;sup>47</sup>Choi, "Kanhantō no Eisankō-ryūiki."

the time. In any case, even if the statement that the emperor "could not see" really was an expression of his weak eyesight, it would be consistent with the symptoms of Marfan syndrome.

Meanwhile, a typical case of short stature was the Hikihito people. Based on the description of the Tsuchigumo people, who they are said to have resembled, the Hikihito would seem to have possessed long limbs compared to their trunk. Two main possible causes of genetic short stature are chondrodysplasia<sup>48</sup> and growth hormone deficiency.<sup>49</sup> However, since the trait of long limbs cannot occur in the case of chondrodysplasia, if there is any truth in such a representation, the Hikihito possibly suffered from growth hormone deficiency and genetic short stature.

#### Type 2: Excess Tissue or Organ Formation

At birth, Emperor Ōjin 応神 is described as having a soft, fleshy mass on his forearm. Since the emperor had a long life according to the chronicle, we ruled out the possibility that this node of his was malignant. The most likely cause, then, would be vascular malformation.<sup>50</sup> Such anomalies often occur in the extremities and are not fatal diseases.<sup>51</sup> Vascular malformation can be venous, lymphatic, and a mixture of the two, and it has been reported that venous malformation in the limbs mostly causes pain in the muscles, tendons, and bones.<sup>52</sup> According to the *Nihon shoki* description, however, this emperor enjoyed hunting several times, and these statements suggested that he did not feel much pain in the arm with the mass.<sup>53</sup> Thus, the mass would more likely be a lymphatic rather than a venous malformation.

While the descriptions about Emperor Ōjin are highly likely to be caused by a congenital anomaly, the other cases found are not likely to have been factual. The tailed people recorded during Emperor Jinmu's reign may have had a congenital anomaly known as a human tail.<sup>54</sup> Even if they had this anomaly, it would be difficult to confirm in an individual wearing clothing unless the tail-like structure itself were exposed. Thus, regarding these tailed people, the descriptions are highly likely to be metaphors for other facts (e.g. symbols of power, specific forms of clothing or other local customs) rather than representing the symptoms of some congenital anomaly.

In another case, there is a description of a Korean man named Tsunuga Arashito with a horn on his forehead. The *Nihon shoki* notes that he was a member

<sup>&</sup>lt;sup>48</sup> Horton, Hall, and Hecht, "Achondroplasia."

<sup>&</sup>lt;sup>49</sup> Şıklar and Berberoğlu, "Syndromic Disorders with Short Stature."

<sup>&</sup>lt;sup>50</sup> Legiehn and Herans, "Venous Malformations."

<sup>&</sup>lt;sup>51</sup> Maftei et al., "The Surgical Management"; Merouane et al., "Cystic Lymphangioma of the Elbow."

<sup>&</sup>lt;sup>52</sup> Rikihisa et al., "Evaluation of Pain Incidence."

<sup>&</sup>lt;sup>53</sup> Sakamoto et al., *Nihon shoki*, vol. 1, pp. 370–371, 374–375.

<sup>&</sup>lt;sup>54</sup>Tojima and Yamada, "Classification of the 'Human Tail"; Tojima, "A Tale of the Tail."

of the royal family on the Korean peninsula and that he had made a voyage from there to Japan.<sup>55</sup> In such a situation, his health condition must have been strong enough to survive both what would have been an arduous journey during ancient times, as well as a long stay in Japan. As such, one would not expect him to have had headaches or other problems in association with the "horn." Interpreting this description as a case of hard, palpable tissue formation in the frontal region of the head, the most likely cause would then be a benign skull or subcutaneous tumor, such as an osteoma or calcifying epithelioma.<sup>56</sup> Other anomalies, such as intraosseous meningioma, which causes a mass in the frontal part of the skull, might also be possible.<sup>57</sup> In this particular case, however, several Japanese historical studies have hypothesized that this does not represent an actual physical description.<sup>58</sup>

Hashirokumawashi, who revolted against Empress Jingū, is described as having had wings on his body. The closest available realistic interpretation of a "wing-like" structure on the trunk would be a winged scapula.<sup>59</sup> This is an anomaly in which the medial border of the scapula rises when the arm is raised, giving the appearance of angel wings or folded bird wings. However, this condition does not match the *Nihon shoki's* clear depiction of Hashirokumawashi's high mobility, such as in his "flying high." Winged scapula can be caused by paralysis of the serratus anterior muscle due to palsy of the long thoracic nerve, or by paralysis of the trapezius muscle due to paranodal nerve injury. In other words, when a winged scapula occurs, flexion of the shoulder joint to raise the arm forward is restricted, and athletic ability rather reduced.

As for Sukuna, who fought Emperor Nintoku, there are many morphological descriptions, but just as with Hashirokumawashi, comparable cases hardly even exist. First, the characteristic of having one body, two faces, four arms, and four legs in the literal sense can only realistically be true of conjoined twins (craniopagus twins). In the case as described, however, facial orientation of the dorsum seems to be present, which is rare. The descriptions about fused parietals and being without any nape suggest that the head also would be fused to a significant degree. Moreover, the phrase "possessing knees but no popliteal fossa" is challenging to interpret. Additionally, if Sukuna were so severely deformed, it would be difficult to even survive to adulthood, let alone fight with a sword or bow and arrow. Therefore, it is more likely that this Sukuna, as well as the abovementioned Hashirokumawashi, did not actually have any such deformity, and that the descriptions thereof are rather merely metaphorical expressions.

<sup>&</sup>lt;sup>55</sup> Sakamoto et al., Nihon shoki, vol. 1, pp. 258–259.

<sup>&</sup>lt;sup>56</sup> Farah et al., "Giant Frontal Sinus Osteoma."

<sup>&</sup>lt;sup>57</sup> Agrawal et al., "Intraosseous Intracranial Meningioma"; Kim et al., "Two Cases of Primary Osteolytic Intraosseous Meningioma."

<sup>&</sup>lt;sup>58</sup> Sakamoto et al., Nihon shoki, vol. 1, p. 589.

<sup>&</sup>lt;sup>59</sup> Fiddian and King,"The Winged Scapula"; Kitamura et al., "Tōka ni okeru yokujō kenkō shōrei no kentō."

<sup>60</sup> Walker and Browd, "Craniopagus Twins."

The descriptions of excess tissue or organ formation in non-imperial individuals share several commonalities. First, all five examples were about leaders of local or frontier-region communities. Furthermore, the formation of organs such as tails, horns, and wings, which humans do not typically possess, and the two examples of Hashirokumawashi and Sukuna, who revolted against the imperial court, are accompanied by descriptions suggesting superhuman physical abilities, such as flight and immense power and agility. These may be expressions of contempt for the frontier-region people who defied the imperial court, disparaging them as barbarians even while granting the strength of their power.

## Type 3: Speech or Behavioral Abnormality

Of the two cases suggesting speech impairment, the cause of the problem seems to be different in the cases of Homutsuwake, the son of Emperor Suinin, and Takeru no Miko, the son of Emperor Tenji. The major differences between the two lie in whether they were adults or infants, and in whether they eventually became able to speak.

Regarding Homutsuwake, the expression "he cries like a baby all the time" suggests that he had a speech impediment, not a problem with vocalization. Additionally, description of his age (thirty years old), and of his having a long beard, indicates that he was an adult physically. He also eventually became able to speak, after he became interested in swans, which he also enjoyed playing with. Cumulatively, these descriptions suggested that his symptoms were likely caused by autism. <sup>62</sup> If so, this would explain his survival to adulthood, both his dysphasia as well as his ultimate ability to speak, and even his strong attachment to a particular object, in this case the swan.

Conversely, in the case of Takeru no Miko, there is only the statement that he was unable to speak, leaving it unclear whether he had a problem with speech or with articulation. The major difference between him and Homutsuwake is that Takeru no Miko died at the age of eight, before he learned to speak. The cause of his death is not mentioned, and it is not difficult to imagine that the infant mortality rate was higher in ancient times than today. Yet while it is also unclear whether there was a connection between his speech impediment and his premature death, if there was a connection, Takeru may have had some severe intellectual disability (mental retardation). It is known that many childhood diseases can cause both mental disability and premature death. Arima syndrome, which causes renal failure, is one such example.

Next, we discuss examples of impulsive murder and the tendency to become enraged. Emperor Yūryaku was described as one who summarily executed his

<sup>&</sup>lt;sup>61</sup> For example, on the Kuzu district, see Haraguchi, "Kuzu no utabue sōjō."

<sup>&</sup>lt;sup>62</sup> Mody and Belliveau, "Speech and Language Impairments."

<sup>63</sup> Katō, "Seishin chitaijisha no hassei yōin."

<sup>&</sup>lt;sup>64</sup> Arima et al., "Nō keisei ijō"; Kumada et al., "Renal Disease in Arima Syndrome."

subjects whenever their words or actions deviated from what he expected. Given the disproportion between the triggering events and the violence expressed by the emperor on such occasions (quite extreme in degree), together with the frequent occurrence of such descriptions, he might have had an intermittent explosive disorder, a type of impulse control disorder.<sup>65</sup>

The chronicle of Emperor Buretsu, the grandson of Emperor Yūryaku, also has many descriptions of abuses against his subjects, but his abuses differ in nature. In contrast to Emperor Yūryaku, who responded to the actions of his subjects with violence, many of Emperor Buretsu's abuses can only be understood as having been conducted for his own interest or pleasure, without any apparent fault on the part of the victim. However, several hypotheses have been proposed to date by Japanese historians suggesting that descriptions of Emperor Buretsu as a tyrant are not accurate. Tsuda's study in 1947 noted that Emperor Buretsu's death without a successor severed the male lineage that had existed since the reign of Emperor Nintoku. 66 Furthermore, he believed that the Nihon shoki's editors, based on Confucian ideology, considered Buretsu evil—precisely because he had severed this lineage—and fabricated such an account accordingly. Another theory suggests that the editors of the Nihon shoki fabricated the story of Buretsu's tyranny in order to legitimize the accession of Emperor Keitai 継体 as his successor.<sup>67</sup> Although the authenticity of the descriptions in the Nihon shoki remains uncertain, from a medical point of view, since impulse control disorders can be inherited, it is possible to decipher the presence of genetic influences in these successive descriptions of tyranny from both Emperors Yūryaku and Buretsu, respectively grandfather and grandson.

Finally, regarding the person who committed suicide after uttering something mysterious during the reign of Emperor Tenmu, from a medical point of view, the possibility of schizophrenia might be raised.<sup>68</sup>

#### Type 4: Different Facial or Body Features

In conclusion, we believe that descriptions of this type are not related to congenital anomalies. We presume that these descriptions result from a combination of both anthropological morphological differences within Japan at that time and feelings of contempt for the barbarians outside the area of imperial dominion. This speculation is based on the statements that the people described as having "different appearances" all lived in areas geographically distant from the Kansai region where the imperial court was located—for example in the present Kyushu and Tōhoku regions.

Regarding the facial morphology of the ancient northeastern people called

<sup>&</sup>lt;sup>65</sup> Schreiber, Odlaug, and Grant, "Impulse Control Disorders."

<sup>66</sup> Tsuda, Nihon jōdai-shi no kenkyū.

<sup>&</sup>lt;sup>67</sup> Inoue, Shinwa kara rekishi e.

<sup>&</sup>lt;sup>68</sup> Hor and Taylor, "Suicide and Schizophrenia"; Sher and Kahn, "Suicide in Schizophrenia."

Emishi, Takigawa's study in 2012 used excavated human skeletal remains and suggested that there may have been a mix of groups in the region, some with strong Jōmon-period characteristics and others who were immigrants from the Kansai region. <sup>69</sup> Several human remains excavated from the Yamoto Tunnel Burials (from the mid-seventh to the early ninth century) <sup>70</sup> and from the Tekiana Cave (from the eighth to the first half of the tenth century) <sup>71</sup>—both located in Miyagi Prefecture—exhibit Jōmon-period facial features, indicating that they were morphologically different from the Kansai-dwelling population at that time. The people who strongly retained these Jōmon-period characteristics were likely the Emishi, who were considered "different in appearance." However, such descriptions may also have been influenced by the dominant viewpoint of the time, centered on the imperial court, which regarded remote areas and areas outside the court's control as barbaric and primitive.

Thus, descriptions of this type combine both differences in physical forms or lifestyles that actually existed, and also more exaggerated representations rooted in the court-centrism of the time.

#### Type 5: Pigment Anomaly

Since this type includes only simple descriptions of pigmentary anomalies without involving any other superhuman characteristics, for such cases congenital anomalies could well be the most reasonable explanation. Particularly, it is clearly stated that the silver hair of Emperor Seinei was congenital, raising the possibly that he had albinism, an anomaly caused by reduced melanin pigment biosynthesis.<sup>72</sup>

In the chronicle of Emperor Kōgyoku, it is suggested that Yamashiro Ōe no Miko had gray-streaked hair, similar to that of a Japanese serow. However, since it is unclear how old he was at the time of this description, and from the fact that he suffered the serious stress of being attacked, it is difficult to determine whether his gray hair was congenital, or acquired due to aging or to stress.

The group of Korean people who came to Japan during the reign of Emperor Suiko was most likely afflicted with vitiligo.<sup>73</sup> Its cause is unclear, but according to the description, they were skilled workers who arrived in Japan together and may have shared some genetic traits. Since there is also a description of them being regarded as peculiar because of white patches, they might have been discriminated against for their appearance also in their original homeland, prompting their move to Japan.

<sup>&</sup>lt;sup>69</sup> Takigawa, Kotsukōkogaku to Emishi, Hayato.

<sup>&</sup>lt;sup>70</sup> Takigawa and Satō, "Hokuhen no yokoanabo kodaijin."

<sup>71</sup> Yamaguchi and Ishida, "Human Skeletal Remains."

<sup>&</sup>lt;sup>72</sup> King, "Albinism"; King and Summers, "Albinism"; Oetting and King, "Molecular Basis of Albinism"

<sup>73</sup> Pande, Suggu, and Bhalla, "Rare Case of Congenital Vitiligo."

## Trends and Factors of Congenital Abnormalities in the Nihon shoki

We found several descriptions of structural or functional abnormalities in the *Nihon shoki* that, from the viewpoint of modern medical science, might appear to result from congenital anomalies. Although there are various causes of such congenital anomalies, systematic studies on congenital anomalies since the 1960s have revealed that they can result from a deteriorated prenatal environment as well as from genetic factors. Therefore, we also examined whether there are any descriptions in the *Nihon shoki* of such possible environmental (i.e., non-genetic) causes of birth defects, as described below.

# Relation to the Occurrence of Epidemics, Famines, and Disasters

The *Nihon shoki* records four instances of pandemic disease: in the fifth and twelfth years of Emperor Sujin's reign<sup>74</sup>, in the thirteenth year of Emperor Kinmei's reign<sup>75</sup>, and in the fourteenth year of Emperor Bidatsu's reign.<sup>76</sup> The last of these pandemics is specifically described as being smallpox.

However, there seemed to be no clear causal relationship between the occurrence of large-scale epidemics and congenital anomalies in Japan. There was no clear description of congenital anomalies in the chronicle of Emperor Sujin (although there is a record in the chronicle of Emperor Sujin's reign). There is also only one case each to be found in the chronicles of Emperors Kinmei and Bidatsu. Furthermore, as both the Kinmei and Bidatsu descriptions relate to the Emishi (type 4), the possibility of a congenital anomaly in either case is low.

Other environmental factors that may lead to the deterioration of maternal nutritional status include the occurrence of famine. Definitive descriptions of severe famine were recorded three times: in the twenty-eighth year of Kinmei's reign (great flood in the country, many people starving, some resorting to cannibalism)<sup>77</sup>; in the thirty-third year of Suiko's reign (prolonged period of rain from the third to the seventh month, with all people starving and particularly many elders, infants, and mothers being said to have starved to death)<sup>78</sup>; and in the eighth year of the reign of the thirty-fourth emperor, Jomei 舒明 (severe drought, the whole country starving).<sup>79</sup> As can be seen in these cases, poor weather significantly impacted grain yields.

From our results, it became clear that descriptions of congenital anomalies did not occur frequently in the *Nihon shoki* at times when plagues and crop failures were recorded (**Table 3**). Therefore, it is difficult to find a clear causal relationship

<sup>&</sup>lt;sup>74</sup> Sakamoto et al., Nihon shoki, vol. 1, pp. 248–249.

<sup>&</sup>lt;sup>75</sup> Ibid., vol. 2, pp. 102–103.

<sup>&</sup>lt;sup>76</sup> Ibid., pp. 148–149.

<sup>&</sup>lt;sup>77</sup> Ibid., pp. 126-127.

<sup>&</sup>lt;sup>78</sup> Ibid., pp. 212–213.

<sup>&</sup>lt;sup>79</sup> Ibid., pp. 230–231.

	Total number of disasters*	Famine or crop failure**	Climate distur- bance***	Earth- quake	Heavy rain or flooding		Storm or strong wind	Ash fall
Sujin 崇神 (10th)	1		1					
Ingyō 允恭 (19th)	2		1	1				
Kinmei 欽明 (29th)	1	1			1			
Sushun 崇峻 (32nd)	1				1			
Suiko 推古 (33rd)	6	2	3		2	1		
Jomei 舒明 (34th)	7	1	1		3	1	3	
Kōgyoku 皇極 (35th)	24		11	3	4	2	5	
Kōtoku 孝徳 (36th)	1				1			
Tenji 天智 (38th)	3			1	1		1	
Tenmu 天武 (40th)	35	2	2	18	2	7	4	2
Jitō 持統 (41st)	12			1	2	9		

<sup>\*</sup>Not counting separately those cases where multiple descriptions were thought to represent one continuous disaster, or where the phenomena described were considered to be secondary manifestations of a larger single disaster.

between the two, and at the very least, the descriptions of congenital anomalies collected in this study can be considered to have occurred independently of plague or famine.

# Differences in Disease Incidence/Description between Emperors and Non-emperors

Not only some congenital anomalies but also other genetic diseases for which the causative genes have been identified are familial, as the genes are inherited from one generation to the next. European royal families are known to historically have many consanguineous marriages, in which case the disease genes of their lineages are assumed to have been passed on at a high rate, causing diseases

<sup>\*\*</sup>Number of records clearly describing famine or crop failure.

<sup>\*\*\*</sup>Includes temperature extremes, excessive snowfall, and hail.

such as hemophilia.<sup>80</sup> Additionally, about half of all congenital anomalies are multifactorial. Even in diseases where the specific causative genes have not been identified (including cleft or palate lip<sup>81</sup> and some congenital heart diseases<sup>82</sup>) there are higher recurrence rates among people with a blood relationship to carriers of such diseases than among the general population. It would therefore be unsurprising if, once found in the Japanese imperial family, such diseases came to appear repeatedly.

In the long history of Japanese emperors, there have been several generations of consanguineous marriages to maintain the lineage. 83 It would thus be expected if the incidence of congenital anomalies and the expression of associated symptoms were more prevalent in the imperial family than among non-imperials. However, no clear trend specific to the imperial families was found. The incidences of congenital anomalies are difficult to estimate accurately in either imperial or non-imperial people. This is largely because the *Nihon shoki* was written to record the emperors' achievements. Given its nature as a national history compiled by the order of the emperor, there are inevitably many descriptions of imperial family members. At the same time, even information about imperial children is not necessarily recorded correctly in the Nihon shoki: Emperor Keikō is said to have had more than eighty children, yet their names are not recorded.84 It is also difficult to estimate the number of non-imperial citizens at that time, especially before the family register system had been introduced. Therefore, for both emperors and non-imperials (whose population size is unknown), the calculation of incidence rates for various congenital anomalies is impossible.

Among descriptions regarding the members of the imperial family, some potential genetic traits were found: the tall stature recorded for Yamato Takeru no Mikoto and Emperor Chūai, and the descriptions of Yūryaku and Buretsu's tyrannical behavior. However, these are only potential genetic traits, and there are no cases where incestuous marriage seems to have been the leading cause.

It is only in the case of Emperor Ōjin that the maternal environment, rather than heredity, could not be ruled out as a possible influence. It is written that Empress Jingū, Emperor Ōjin's mother, went to the Korean peninsula to fight in Silla while pregnant, dressed as a man and leading an army.<sup>85</sup> If hypothetically this were true, it might be imagined that the empress was under severe stress during her pregnancy. As described above, we have speculated that the fleshy mass on the emperor's arm was a lymphatic or vascular malformation. Recent studies have shown that the incidence of infantile hemangioma is higher when

<sup>80</sup> Stevens, "The History of Haemophilia."

<sup>81</sup> Mossey and Modell, "Epidemiology of Oral Clefts 2012."

<sup>82</sup> van der Bom et al., "The Changing Epidemiology."

<sup>83</sup> Sakamoto et al., Nihon shoki, vols. 1, 2.

<sup>84</sup> Ibid., vol. 1, pp. 286–287.

<sup>85</sup> Ibid., pp. 334–335.

the mother has allergic conjunctivitis or hay fever.<sup>86</sup> Thus, we believe that such a high-stress state as in Empress Jingū's case, during a pregnancy, could well have been a factor in producing something like Emperor Ōjin's condition.

#### Conclusion

In this study, we attempted to understand the epidemiology of congenital anomalies in ancient times, which until now have been considered a complete mystery. This study showed that by reading the *Nihon shoki* from both medical and historical perspectives, we were able to catch a glimpse into an aspect of ancient times that until now had remained invisible. In addition to the *Nihon shoki*, many other valuable documentary texts exist in Japan. These include manuscripts and anecdotes related to the history of other regions in East Asia, sometimes even records that no longer survive in their countries of origin. As with the *Nihon shoki*, it is impossible to interpret all the descriptions in such historical documents as fact. However, by expanding in scope the application of a method like ours, we can expect to learn more about ancient congenital anomalies, their pathologies, and the responses of people to the same long before the spread of modern medicine, not only in Japan but also in East Asia as a whole. We believe this study has shown significant results in demonstrating the potential of these new methods for deciphering ancient epidemiology from historical documents.

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<sup>86</sup> Mizawa et al., "Infantile Hemangioma and the Risk Factors."

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