

Akhenaten and the Strange Physiques of Egypt's 18th Dynasty

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Akhenaten was one of Egypt's most controversial pharaohs, in part because of his strange appearance in images produced after he had declared Aten, the Sun-disc, his one-and-only god. Whether these were symbolic representations or realistic ones that indicate a deforming genetic disorder is the subject of continuing debate. The authors present evidence that the bizarre physical features portrayed in these images are not only realistic but were shared by many members of Egypt's 18th Dynasty. The features are best

explained by either 2 different familial disorders—the aromatase excess syndrome and the sagittal craniosynostosis syndrome—or a variant of the Antley-Bixler syndrome caused by a novel mutation in one of the genes controlling the P450 enzymes, which regulate steroidogenesis and cranial bone formation.

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When Akhenaten, also known as Amenophis IV, ascended Egypt's Horus Throne of the Living in 1377 BCE, he was ill-prepared to rule the most powerful empire on earth. He had previously been excluded from court functions, possibly because of his strange physique. Although some speculate that excessive inbreeding caused his deformities, others point out that he was the product of a gene pool that had not been corrupted by close intermarriage for at least 2 generations (1).

A CASE SUMMARY

The distinctive physical features of Akhenaten depicted in statues and reliefs are at once odd, strikingly diverse, and inconsistent. Likenesses produced at the beginning of his reign depict him in traditional pharaonic guise, with a relatively normal face and physique. However, after he created his radical new religion that focused on Aten (the Sun-disc) and became history's first monotheist, his images became floridly androgynous, with an elongated head, almond-shaped eyes, lantern-like jaw, protruding teeth, and large ears (Figure 1). Some representations show similar distortions of the head, body, and extremities in Akhenaten's children and his principal wife, Nefertiti (Figure 2).

Only a bit more is known of the health of Akhenaten or of his family. He had 6 children by Nefertiti; all were daughters. Many members of his family died during a plague that ravaged his kingdom. Akhenaten survived the plague only to die in the summer of 1359 BCE under obscure circumstances (1).

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THE HERETIC PHARAOH

Amenophis was born in ancient Egypt's royal city of Memphis in approximately 1385 BCE. His father, Amenophis III, ruled an empire stretching nearly 2000 miles from the central Sudan to the mountains of Anatolia. An elder brother, named Thutmose after his grandfather, stood in line to inherit the throne but died suddenly in the third decade of his father's reign, leaving Prince Amenophis the heir apparent.

As a youth, Prince Amenophis was assigned undistinguished tutors. When his father became ill, the ailing king moved his court south to the more salubrious climate of Thebes, where the prince seems to have been hidden from public view until becoming Pharaoh.

Amenophis III died in his 38th year on the throne (around 1377 BCE) and was immediately succeeded by Prince Amenophis (now Amenophis IV). At first, little changed. The new king continued to reside at Thebes in his father's palace and, shortly after his accession, married a strikingly beautiful woman named Nefertiti. Some believe she was a distant relative. In the first or second year of their marriage, she bore a daughter, Meret-aten.

Sometime during the new king's second year on the throne, he experienced a revelation regarding the gods; their temples; their cult images; and the sacred, prescriptive literature. This new insight induced him to decree all the gods of the Egyptian pantheon nonexistent except one, the solar deity. Pursuant to this revelation, he forbade the depiction of any god or goddess except the falcon-headed human figure bearing a large sun-disc on its head. He then had the great house of the Sun-disc at Karnak transformed into a new, open-air temple, wholly devoid of cultic apparatus except for the large altar on which offerings to the sun could be made. He diverted the incomes of the other temples to the new establishment, which caused all other shrines to close their doors. In decorating his new structures, the king commissioned a new artistic style which depicted him in a greatly distorted form as something akin to a "humanoid praying mantis" (1), with all of the bizarre and effeminate features we have enumerated.

Key Summary Points

Akhenaten (also known as Amenophis IV) was one of ancient Egypt's most controversial pharaohs because of his devotion to Aten, the Sun-disc, as his one-and-only god; he has a strange appearance in images produced after the introduction of his radical new religion.

Whether the floridly androgynous images of Akhenaten were symbolic representations, meant to concretize the concept of him as the father and mother of all mankind, or realistic representations indicative of some deforming genetic disorder is the subject of continuing debate.

The bizarre physical features portrayed in these images (specifically, gynecomastia and cranial abnormalities) were shared by many members of Egypt's 18th Dynasty and are therefore realistic rather than symbolic.

The physical abnormalities of Egypt's 18th Dynasty are best explained by either 2 different familial disorders—the aromatase excess syndrome and the sagittal craniosynostosis syndrome—or a variant of the Antley–Bixler syndrome.

Explanations for these strange images have followed 2 opposing lines. One maintains that they were purely symbolic and intended to concretize the concept of Amenophis IV as the father and mother of all mankind. The other holds that they were realistic and conformed, in the main, to what the king actually looked like. With regard to the former hypothesis, Egyptian art is replete with hermaphroditic representations; however, none is of the character of the images of Amenophis IV.

In the fifth year of his reign, conflict with the priesthood of Amun induced the king to move to a new site in Middle Egypt, near the modern-day village of Amarna, and build a new capital (Akhet-aten, the “Horizon of the Sun-disc”). On leaving Thebes, he closed the Temple of Amun-Re, king of the gods, and expunged the deity's representations and name from all monuments. He also discarded Amenophis (“Amun is satisfied”) in favor of a new title, Akhenaten (“useful for the Sun-disc”). Once settled in Akhet-aten, he seems never to have traveled abroad, apparently content to closet himself in his new capital, oblivious to the economic calamity his new belief system brought about.

Something of Akhenaten's personality can be discerned from his diplomatic correspondences and the few inscriptions that survived his later anathematization. He was not an evangelical and had no wish to proselytize or convert his people. He confined his teaching to an inner circle of courtiers, all of whom remarked on the beauty of his mellifluous voice. In curious contrast to the single-

mindedness with which he pressed his new belief system, he was frequently irresolute about the outside world and procrastinated constantly in his negotiations with foreign ambassadors.

Akhenaten died during his 17th year on the throne. The cause of his death is unknown. Contemporary texts speak of a plague that ravaged the eastern Mediterranean, Anatolia, and (undoubtedly) Egypt, in which many of the Amarna faithful are thought to have perished. Akhenaten died some time later and was interred in an unknown location, possibly in an Amarna tomb he had had carved for himself. Some suspect that shortly after his capital was abandoned, his remains were moved to KV 55 in the Valley of the Kings (2–6), although they have yet to be found.

THE DYNASTIC DISORDERS

Akhenaten's gynecomastia and androgynous appearance have previously been attributed to a host of genetic disorders, including the Marfan syndrome, Frohlich syndrome (adiposogenital dystrophy), Klinefelter syndrome, and androgen insensitivity syndrome (1, 7, 8). Although each of these has features in common with the abnormalities depicted in the Amarna images, each also has features

Figure 1. Alabaster statuette of Akhenaten.



Reproduced with permission from Bildarchiv Preussischer Kulturbesitz/ Art Resource, NY Aegyptische Museum, Staatliche Museen zu Berlin, Berlin, Germany.

Figure 2. Relief of Akhenaten, Nefertiti, and their 3 eldest daughters.



The second daughter, Meketaten, sitting on Nefertiti's lap, is 5 to 7 years old and has prominent breasts and wide hips. Reproduced with permission from Vanni/Art Resource, NY Aegyptisches Museum, Staatliche Museen zu Berlin, Berlin, Germany.

inconsistent with what we know of Akhenaten's clinical history. The Marfan syndrome is not associated with either gynecomastia or an androgynous habitus, and patients with

Table. Images Depicting Endocrine and Cranial Abnormalities in the Principal Rulers of Egypt's 18th Dynasty

Ruler	Gynecomastia (Reference)	Cranial Abnormality (Reference)
Amosis	No data*	Absent (10 [plate 11])
Amenophis I	Absent (11)	Absent (12)
Thutmose I†	Present (13 [page 101])	Present (10 [plates 20–22])
Thutmose II	Absent (14)	Present (10 [plates 23 and 24])
Queen Hatshepsut	Not applicable	Present (15)
Thutmose III	No data*	Present (10 [plate 28])
Amenophis II	Present (16)	No data*
Thutmose IV	Present (13 [page 34])	Present (10 [plates 29 and 30])
Amenophis III	Present (13 [page 44])	Present (10 [plate 34])
Amenophis IV/Akhenaten	Present (13 [page 108] and 17 [page 93])	Present (Figure 1)
Smenkhkare	Present (18‡)	Present (10 [plate 36])
Tutankhamun†	Present (13 [page 180])	Present (13 [page 7])
Aye	Absent (18)	Absent (18)
Horemheb	Absent (18, 19)	Absent (18, 19)

* We could not establish presence or absence of cranial or endocrine abnormality in any statuary, paintings, mummies, or reliefs.

† Patrilineal line of Akhenaten begins with Thutmose I and ends with Tutankhamun.

‡ Click on "18th Dynasty" in the left sidebar of this Web site to see images of all the kings of the 18th Dynasty.

each of the other syndromes are nearly always sterile (Akhenaten fathered 6 daughters).

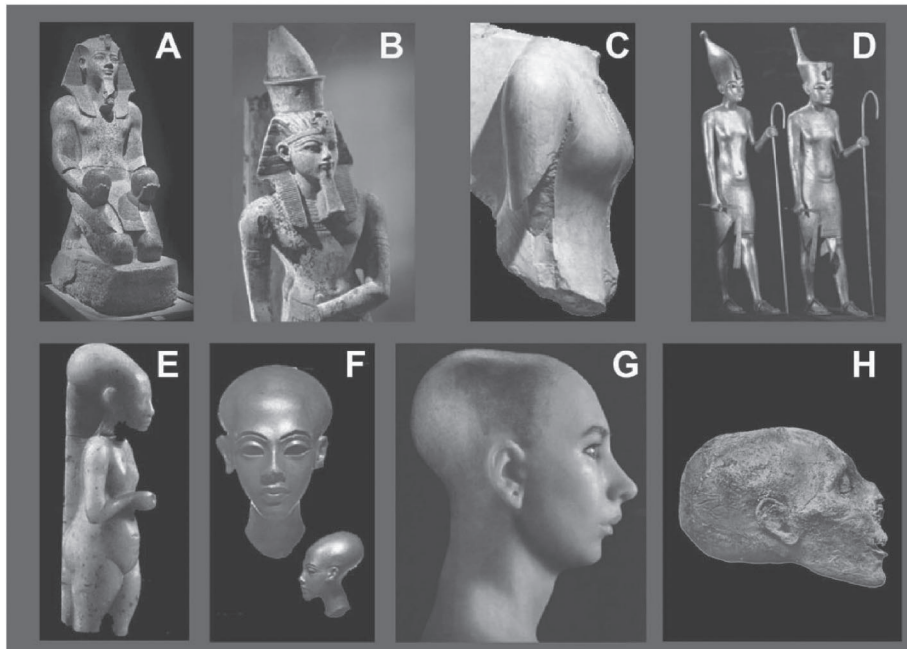
In 1980, Dr. Bernadine Paulshock called attention to evidence of familial gynecomastia in Egypt's 18th Dynasty, Akhenaten's extended family (9). She diagnosed familial gynecomastia, mechanism unknown but probably dominantly transmitted, on the basis of presence of the physical finding in images of Amenophis III (Akhenaten's father), Amenophis IV (Akhenaten), and Tutankhamun and Smenkhkare (his brothers) (Table) (10–19). She also detected evidence of gynecomastia in a sculpture of Thutmose IV (his grandfather) but apparently was unaware that a statue of Thutmose I, the patriarch of Akhenaten's family, also shows evidence of breast enlargement. We believe the presence of gynecomastia in no fewer than 4 generations of the 18th Dynasty without apparent impairment of male fertility is best explained by the aromatase excess syndrome, a dominantly transmitted disorder first described in 1977 (20).

Aromatase (estrogen synthetase) is one of many P450 enzyme systems. In the aromatase excess syndrome, aromatase converts the C19 steroids androstenedione, testosterone, and 16-hydroxyandrostenedione into estrone, estradiol, and estriol, respectively. This occurs not just in gonadal tissues but also in extragonadal tissues, such as subcutaneous fat and skin fibroblasts. In affected males, mutations of the aromatase gene (*CYP19* on chromosome 15q21) and promoters from adjacent genes modulate *CYP19* to produce increased aromatase, which results in gynecomastia, a eunuchoid habitus with normal fertility, and (potentially) the mellifluous (female) voice Akhenaten is purported to have had. Affected females have isosexual precocity and, in some instances, macromastia (21–23).

Gynecomastia in images of the patrilineal line of the 18th Dynasty is both striking and widespread (Figure 3, A through D); however, isosexual precocity in the females is less so. Nevertheless, a relief believed to represent Akhenaten's second daughter, Princess Meketaten (Figure 2), a child of no more than 5 to 7 years of age, depicts her with both enlarged breasts and prominent hips and buttocks. Two statues of Akhenaten's daughters, one of which is shown in Figure 3 (panel E), depict similar evidence of isosexual precocity (17, 24). Only the aromatase excess syndrome causes these distinctive familial anomalies. Testing for the aromatase gene in tissues from the 3 relevant mummies of the 18th Dynasty that are available (Thutmose I, Smenkhkare, and Tutankhamun) could verify or disprove the diagnosis.

The other unusual anatomical feature of Akhenaten is the long narrow face with prominent chin, long neck, and presumed elongated occiput (Figures 1 and 2). Because no drawings or busts of Akhenaten without a crown exist and his mummy has never been located, we cannot be certain of the shape of his head. However, 5 of his 6 daughters are depicted with long necks, markedly elongated skulls with overhanging occiputs, and (in at least 1 case) broadening of

Figure 3. Images of members of Egypt's 18th Dynasty.



A. Statue of Amenophis II (Akhenaten's great-grandfather), showing gynecomastia, making an offering from the Temple of Amun at Thebes. Reproduced from reference 16 with permission. B. Votive figure of Amenophis III (Akhenaten's father), showing gynecomastia. Reproduced from reference 13 with permission. C. Fragment of a statue of Akhenaten, showing gynecomastia. Reproduced from reference 17 with permission. D. Tutankhamun (Akhenaten's brother), as the King of Upper and Lower Egypt, showing gynecomastia. Reproduced from reference 13 with permission. E. Alabaster statuette of a daughter of Akhenaten, showing elongated head, cranial (parietal bone) widening, breast development, and widened hips and buttocks. On the basis of facial development, she is 3 to 5 years of age. Reproduced from reference 24 with permission. F. Daughter of Akhenaten as a teenager or young adult, showing elongated and widened skull. Reproduced from reference 17 with permission. G. Forensic reconstruction of Tutankhamun from computed tomography scan data of his skull. Reproduced from reference 13 with permission. H. Elongated head of the mummy of Thutmose III. Reproduced from reference 10 (public domain).

the parietal area (Figure 2 and Figure 3, E and F). We could find no image of the sixth daughter. Craniosynostosis of the sagittal suture, the most common inherited craniosynostosis, produces this type of dolichocephaly without affecting either cognition or intracranial pressure (25, 26). Recent computed tomographic imaging of Tutankhamun's skull shows the same overhanging occiput and long neck (Figure 3, G) (13). Thutmose III (Figure 3, H), Akhenaten's great-great-grandfather, also has an elongated skull with overhanging occiput, as does every pharaoh in Akhenaten's patrilineal line for whom information exists (Table). Many have prominent chins as well. These findings suggest a second familial disorder, one associated with a cranial and possibly cervical spine abnormality. Likewise, Nefertiti is depicted with an elongated neck and a facial profile that suggests dolichocephaly, but again, she is never seen without a crown. Her relationship to the 18th Dynasty is unclear, although some authorities believe her to have been a distant relative. If she was derived from that genetic pool, she and Akhenaten might both have passed on genes responsible for cranial abnormalities to their daughters.

The genes responsible for the deformities in most of the classic craniosynostosis syndromes—*TWIST*, *FGFR1*, and *FGFR2*—have been identified (26). Because tissues are available from 5 relevant mummies of the 18th Dynasty with cranial abnormalities (Thutmose I, Thutmose II, Queen Hatshepsut, Tutankhamun, and Smenkhkare), a search for these genes among members of the dynasty is feasible.

The Antley-Bixler syndrome, a rare genetic disorder with cranial and endocrine abnormalities analogous to those we have described, involves a combination of deficient estrogen steroidogenesis and abnormal cranial bone formation. The former produces abnormal genitalia in both sexes, and the latter produces craniosynostosis that results in brachycephaly and midface hypoplasia. Although the syndrome was initially believed to be caused by mutations in the *FGFR2* gene, Flück and colleagues (27) recently reported that a mutation in the *POR* gene encoding P450 oxidoreductase is the actual defect responsible for the impaired steroidogenesis and craniofacial abnormalities. In view of these observations, it is possible that the 2 separate familial disorders that affected Egypt's 18th Dynasty actu-

ally represent a unitary disorder akin to the Antley–Bixler syndrome, caused by a novel mutation in one of the genes controlling the P450 enzymes that regulate steroidogenesis, which resulted in excessive rather than deficient synthesis and abnormal cranial bone formation.

SUMMARY

Although some have argued that the images commissioned during the later years of Akhenaten's reign are symbolic rather than realistic, many of the most striking anomalies depicted (the gynecomastia and androgynous physique) are prevalent in images of other pharaohs of the 18th Dynasty but not those of other dynasties. This finding, in conjunction with evidence of isosexual precocity in representations of his daughters, strongly suggests that Akhenaten's bloodline was riddled with the aromatase excess syndrome, which might have contributed to or simply reinforced Akhenaten's belief that he was the incarnation of the father and mother of all mankind. Cranial abnormalities prevalent in royal images uncovered at Amarna also seem to be realistic representations that are best explained by a second familial disorder of the 18th Dynasty, the sagittal craniosynostosis syndrome. Recent findings also suggest that a yet-to-be-discovered mutation in the P450 isoform enzyme system was responsible for both the cranial and endocrine abnormalities of Egypt's 18th Dynasty.

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