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Alice Vance ("Das Bärenweib"): A Historical Case of Nievergelt Syndrome

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Several malformed individuals were presented at the World Exhibition in Antwerp in 1894. Among them was Mrs. Alice Vance from Mount Pleasant, Texas, with congenital limb defects, and Mr. Eugen Berry, who had asymmetrical, monstrous enlargement and macrodactyly of the feet, i.e., Proteus syndrome. After the World Exhibition Mrs. Vance presented herself to the public in Castan's Panopticon imitating a bear. She became famous under the stage name "Das Bärenweib" ("the bear-like woman") and was examined by several German clinicians, and her malformations were considered to be of high scientific interest. Mrs. Vance had mesomelic dwarfism and her mother was known to have similar malformations. Her limb deficiencies were generally considered a unique congenital condition those days, and the diagnosis of "a maternally inherited malformation of the forearms and the shanks" [Daffner 1898: Munch Med Wochenschr 25:782] was made. Virchow [1897: Verh Berl Ges Ethnol Urgeschichte 29:624], feeling attacked by a daily newspaper stating that the physicians as well as the police of Berlin had missed the diagnosis of an "English disease," eventually exercised his authority and diagnosed Alice Vance as a "phocomelic." Clearly, she was not a phocomelic according to past and current definition of this term. Thus, from a historical point of view, the story illustrates how pressure from the daily press altered the definition of an up-to-then precisely defined medical term for decades. According to the clinical data and an X-ray report available from the literature, Alice Vance had a dominantly inherited type of mesomelic dwarfism. We propose the diagnosis of Nievergelt syndrome. Am. J. Med. Genet. 76:145–149, 1998.

KEY WORDS: Nievergelt syndrome; phocomelia; mesomelic dysplasia; mesomelic dwarfism; Proteus syndrome; history of medicine; Virchow

INTRODUCTION

Nievergelt syndrome is an autosomal-dominant disorder comprising marked mesomelic dwarfism with a characteristic rhomboid shape of the rudimentary bones of the leg, normal facial appearance, and normal intelligence. The condition was initially reported as an example of how to use malformations for determining paternity, when Nievergelt [1944] showed a malformed man to be the father of 3 similarly affected sons of 3 different women (male to male transmission in three instances). This family was later restudied by Hess et al. [1978]. A few further sporadic cases [Solonen and Sulamaa, 1958; Young and Wood, 1974; Petrella et al., 1990] have been added since Nievergelt's report [reviewed in Petrella et al., 1990].

We review the case of the 24-year-old Alice Vance from Mount Pleasant, Texas, who was among the malformed individuals presented at the World Exhibition in Antwerp in 1894. Her malformations, along with those of Mr. Eugen Berry from Ohio (who evidently had Proteus syndrome, see Fig. 1), were noted to be of high scientific interest at the end of the nineteenth century [see Henning, 1895].

Mrs. Vance (Fig. 2) had mesomelic dwarfism. After the World Exhibition she was displayed in Castan's Panopticon in Berlin, where she imitated a bear. She became famous under the stage name "Das Bärenweib" ("the bear-like woman," Fig. 3A–C). Since her case was under scientific discussion at the end of the nineteenth century, several accounts on her malformations [Henning, 1895; Maass 1895, 1897; Grunmach, 1897; Virchow, 1897, 1898; Daffner, 1898; Krüger, 1906] are available from the literature, allowing for a retrospective diagnosis.

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Fig. 1. The 22-year-old Eugen Berry from Ohio at the World Exhibition in Antwerp in 1894 [reproduced from Henning, 1895]. Note marked asymmetric hyperplasia of the legs and feet, macrodactyly, and syndactyly of the third and fourth toes of the less severely affected left foot. Mr. Berry was born to normal parents and had several normal sibs [Henning, 1895]. Our diagnosis is Proteus syndrome. Curiously enough, Mr. Berry stated that his feet were absolutely normal at birth [Henning, 1895]. Intriguingly, Virchow [1895] had already stumbled on that clue but diagnosed him as "ein ausgezeichnetes Beispiel der *sonst gewöhnlich als congenital bekannten* Hyperplasie der Füße," i.e., "an excellent example of hyperplasia of the feet, *which is commonly known as a congenital condition*" [Virchow, 1895, emphasis ours].

REVIEW OF CLINICAL DATA

Mrs. Alice Vance was born in 1874 [Daffner, 1898].[1] Her mother was said to earn her living by displaying similar malformations in New York [Virchow, 1898; Daffner, 1898]. Mrs. Vance's father and her 2-year-old sister had normal body proportions [Daffner, 1898]. Parental consanguinity is unlikely, because her mother was a mestiza, while her father was a "Neger" [Maass, 1895; Daffner, 1898], i.e., an African-American. Mrs. Vance married (her husband is shown in Fig. 2) and had a normal child, who died at age 10 months. The sex of this child is uncertain. Daffner [1898] says that Alice Vance gave birth to a daughter, while Virchow [1898] mentions a son.

[1]This date is controversial. According to Henning [1895] and Maass [1895] she was already aged 23 and 24 years, respectively. In our impression, however, Daffner [1898] published the most accurate case report.

Mrs. Vance was an intelligent woman [Henning, 1895; Maass, 1895, 1897; Daffner, 1898] and was able to embroider and to sew ("Alice Vance spricht . . . sehr gut Englisch, näht und stickt und macht überhaupt den Eindruck von Intelligenz" [Henning, 1895]). She had bilateral deficiencies of the forearms and of the shanks and, hence, disproportionate short stature, i.e., mesomelic dwarfism. Her body length was 110 cm. On clinical examination retrogenia was noted [Daffner, 1898], but her face was otherwise normal (Figs. 2, 3A). Examination of the skeleton was carried out by radiography [Grunmach, 1897] and by palpation [Daffner, 1898]. She had bilateral shortness and thickness of the radial, ulnar, tibial, and fibular bones, whereas the humeri and the femora were reported as normal. On X-ray films, the ulnae and radii were "2–3 cm in length, width and depth" [Grunmach, 1897]. Rotational movement of both forearms was preserved, indicating absence of radioulnar synostoses. The hands had 5 fingers each. The range of finger movement was limited (apparently because of shortness of the forearm muscles). Mrs. Vance was unable to fully extend her fingers except for the index finger [Daffner, 1898], which has an additional extensor muscle. X-ray studies showed a "rudimentary carpus." The metatarsals and the phalanges were "complete" [Grunmach, 1897]. Her rudimentary lower legs and her feet where laterally displaced so that Alice walked on the condyles of her femora [Maass, 1895, 1897; Daffner, 1898]. On radiographs, two bones of the legs measuring "4–5 cm in

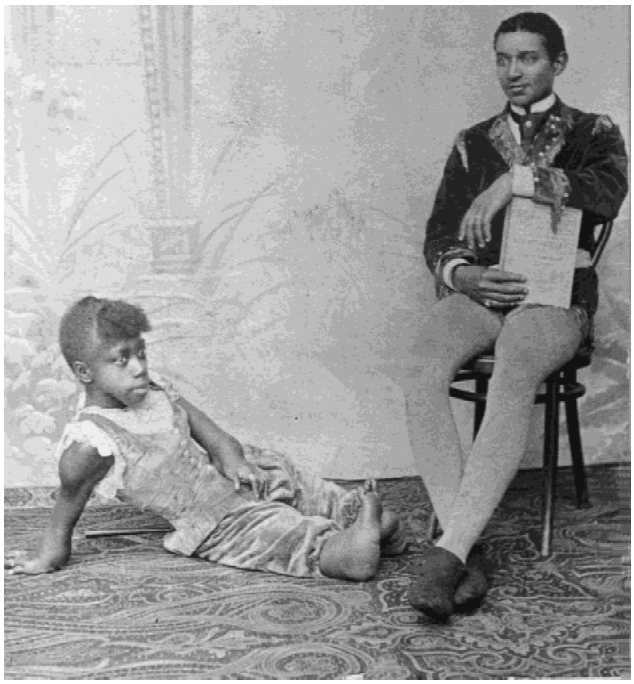

Fig. 2. Mrs. Vance and her husband at the World Exhibition in Antwerp in 1894 (reproduced from photocopy P 3358, Berliner Gesellschaft für Anthropologie, Ethnologie und Urgeschichte, Archiv, Schloß Charlottenburg, Langhansbau, 14057 Berlin). This photograph, labelled as "Das sog. Bärenweib," was probably sent to that society from Antwerp by Henning [1895]. Our assumption is based on a note in his paper: "Der auf mitfolgender Photographie sitzend abgebildete junge Mann, ist ihr Gatte!" (The young man shown on the photograph in a sitting position is her husband!)

Fig. 3. **A:** Mrs. Alice Vance as "the bear-like woman" [reproduced from Krüger, 1906]. Note shortness of both forearms. During her performances in Castan's Panopticon in Berlin, Mrs. Vance came out of a cave, crawling about on all fours [Maass, 1895]. Here she is wearing a bear-fur. There was no hypertrichosis. **B:** Newspaper clipping of an advertisement page (*Berliner Zeitung* of July 28, 1895). "Das Bärenweib" was the main attraction of Castan's Panopticon in Berlin. **C:** "Das Bärenweib" was later shown in Castan's Panopticon in Dresden. Advertisement in the daily newspaper *Dresdner Anzeiger* of November 18, 1896.

length, width and depth" [Grunmach, 1897] were made out. In homology to the upper limb findings, the tarsus was also "rudimentary" on X-ray films and the metatarsals and phalanges were reported as "complete" [Grunmach, 1897]. Daffner [1898] palpated the "fibulae," which were markedly thickened at the upper ends. The X-ray appearance of the ribs, the thoracic spine, the sternum, and the scapulae was normal [Grunmach, 1897].

DISCUSSION

Alice Vance: A Historical Case of Nievergelt Syndrome

In summary, Mrs. Vance had normal intelligence, normal facial appearance except for retrognathia, severe mesomelic dysplasia probably associated with carpal and tarsal coalition, and disproportionate short stature. She had always been of good health [Daffner, 1898]. Thus, she showed no evidence of any associated malformations. The family history is consistent with a dominantly inherited condition.

Differential diagnosis includes all dominantly inherited types of mesomelic dysplasia, i.e., dyschondrosteosis [Léri and Weill, 1929], Nievergelt syndrome [Nievergelt, 1944], "mesomelic shortening and hereditary nephritis" [Funderburk et al., 1976], Osebold-Remondini syndrome [Osebold et al., 1985; Optiz and Gilbert, 1985], "dominant mesomelic dysplasia, ankle, carpal, and tarsal synostosis type" [Kantaputra et al., 1992], and a further condition reported by Leroy et al. [1975]. In dyschondrosteosis and in the condition reported by Funderburk et al. [1976], the limb deficiencies are mild and have other morphological characteristics such as Madelung deformity and bowing of bones. In Osebold-Remondini syndrome there are additional anomalies (e.g., absence of second phalanges, irregular synostoses of digits, and deformities of fingers) which were apparently not present in Mrs. Vance. The condition reported by Leroy et al. [1975] is different from her malformations in that it predominantly affects the radii and the fibulae. "Dominant mesomelic dysplasia, ankle, carpal, and tarsal synostosis type" predominantly affects the ulnae and, to a lesser extend, the radii; unlike the bear-like woman, the legs are just "somewhat short" and there is mildly short stature only [Kantaputra et al., 1992].

The available clinical data and the X-ray report best fit the characteristics of Nievergelt syndrome. The ru-

dimentary mesomelic bones in Nievergelt syndrome are of abnormal thickness and there usually is a characteristic "rhomboid" shape of the fibulae and tibiae on X-ray films. In retrospect, such unusually shaped structures might have been palpated as "fibulae" by Daffner [1898]. The X-ray report of Grunmach [1897], at least, suggests abnormal thickness of the tibiae and the fibulae. The diagnostic value of these X-ray films taken in August 1897[2] might have been limited, because a rather long time of exposure was necessary for obtaining a proper radiograph. Radioulnar synostoses were ruled out clinically [Daffner, 1898]. This anomaly was present in 3 of the 8 patients with Nievergelt syndrome reported so far [Petrella et al., 1990]. All of them were members of the family studied by Nievergelt [1944] and Hess et al. [1978]. Remarkably, the one relative lacking that sign was the one who was most severely affected [Petrella et al., 1990]. Therefore we need not expect it in an individual with radii and ulnae of 2–3 cm in length, width, and depth. Retrognathia has up to date not been reported in Nievergelt syndrome.

Pathogenetically, mesomelic dysplasia is a defect of a secondary epimorphic field involving the forearms and the shanks, which are homologous parts of the upper and lower limbs [for review of developmental field theory, see Opitz, 1996]. Perhaps the mandible is a further component of a polytopic developmental field. If so, retrognathia may reflect the severity of the lesion rather than another differential diagnosis. The combination of marked shortness of the mesomelic parts and retrognathia was reported in individuals with Langer-type mesomelic dwarfism [Langer, 1967]. This condition is the homozygous state of dyschondrosteosis [Espiritu et al., 1975] and is hence ruled out in Mrs. Vance because of her family history.

Daffner [1898] made the diagnosis of "a maternally inherited malformation of the forearms and the shanks." The reports of Henning [1895] and Maass [1895, 1897] imply the same conclusion. Those interpretations are in line with our diagnosis, but the genetic implications of the family history were not understood in those days [discussed in Urban et al., 1997].

How Alice Vance Became a Phocomelic

On November 18, 1896, the *Dresdner Anzeiger* announced that the police had prohibited any further presentation of the "Bärenweib" in Castan's Panopticon in Dresden. According to that daily newspaper an examination of Mrs. Vance carried out on official instruction had brought to light that she was not at all affected by what would be "eine unerklärliche Abnormität" (an inexplicable anomaly). Rather, she was found only to be crippled by earlier disease, probably the "English disease,"[3] and otherwise she would have been a quite normal woman ("[eine] durchaus normale Frauensperson"). And to cap it all, the newspaper emphasized that the "Bärenweib" had been displayed in Berlin for months, but neither the physicians of Berlin nor the police had caught on to that "swindle" [*Dresdner Anzeiger* of November 18, 1896, as cited in Maass, 1897]. Apparently, such actions undertaken by the police were not too unusual, because it was their job to make sure that only "proper miracles" were shown in public. In this context Gruber [1955] mentioned that just 20 years ago he had received a human fetus with an artificial dicephaly confiscated by the police. That particular specimen was an attraction shown at fairs by a touring company and had aroused suspicion on account of different hair color on its two heads.

The substance of the article published by the *Dresdner Anzeiger* was that Mrs. Vance's malformations had been discussed twice before the "Berliner Gesellschaft für Anthropologie, Ethnologie und Urgeschichte." How embarrassing that incident must have been to Virchow and his colleagues. In an emotionally charged session on December 18, 1897, Maass again demonstrated the "Bärenweib" and Grunmach presented his X-ray results. As Maass pointed out, this was to "firmly reject that accusation of that newspaper from Dresden saying that the physicians of Berlin would be unable to accurately classify congenital anomalies, and in order to make clear, that there is no need for instruction from Dresden" (our translation) [Maass, 1897]. The apparently furious Virchow emphasized that he had never had any problems with that case and that he had made the diagnosis of "phocomelia." He additionally promised to give an overview on phocomelics in the following session. As a final word, he wondered why the colleagues from Dresden had assumed an acquired condition in that case and, what was even more surprising to him, how so poorly informed a newspaper could be "that impolite" [Virchow, 1897].

Redefinition of Phocomelia

In the following session on January 15, 1898, Virchow reported "on phocomelics and the bear-like woman." At the beginning he cited Isidore Geoffroy St.-Hilaire's classification of certain limb defects referred to as "monstres ectroméliens" into "phocomelia" (limbs virtually consisting of hands or feet only), "hemimelia" (incomplete limbs with terminal stumps), and "ectromelia" (absent or almost absent limbs). Since Virchow realized that there was a continuous spectrum of limb deficiencies, making an accurate classification of a certain case almost impossible, he lumped these three categories and used the term "phocomelia" for them all,

[2]Roentgen discovered what he referred to as "X-Strahlen" (X-rays) on October 8, 1895. He published his observations on January 23, 1896, in his paper "Über eine neue Art von Strahlen."

[3]"English disease" was an old term for rickets, i.e., "rhachitis," originally an ancient term referring to spinal complaints. Francis Glisson (1597–1677) is credited for restricting the term "rhachites" to its modern meaning in 1650. Thus, "English disease" already referred to an acquired condition. In order to reject the diagnosis favored by the *Dresdner Anzeiger*, however, Virchow [1898] had to face some terminological difficulties. An old, ill-defined term, "rhachitis congenitia," vaguely referred to severe congenital osteodysplasias, and was still in use those days. An early instance for the use of this term is in the 1763 *Dissertatio Inauguralis Medica* of Johann Heinrich Klein, i.e., "Casum rhachitidæ congenitæ observatæ in infante varie monstroso."

i.e., synonymous with St.-Hilaire's "monstres ectroméliens." He commented that too many technical terms may provoke confusion instead of contributing to clarification of the matter. However, strictly speaking, mesomelic dysplasia does not fit with even one of the categories of Geoffroy St.-Hilaire's classification lumped together by Virchow, in which (proper) phocomelia was the *mildest* defect. Furthermore, Mrs. Vance was not at all a "seal-like woman" in the impression of the visitors of the Castan's Panopticon but was successfully imitating a quite different animal. Anyhow, Virchow's definition of phocomelia also included mesomelic dysplasia, apparently on account of the diagnosis he made in the preceding session. His definition influenced later research for several decades until it was eventually rejected by Wepler from Göttingen in 1937 [see Urban et al., 1997].

To best of our knowledge, the case of Alice Vance is the second report on a familial occurrence of Nievergelt syndrome. Up to now, the basic genetic defect has not been identified. We emphasize that the limbs of Mrs. Vance did not look like those of a seal, but to complete the picture we need to add that, according to Daffner [1898], her limbs also did not resemble those of a bear.

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