# MULTIPLE PILOMATRICOMAS OF THE SCALP

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A 41-year-old white woman initially presented in 1982 with myotonic dystrophy and multiple skin nodules (Fig. 1). She first noted the onset of these nodules at age 16. Over an 8-year period, 19 nodules were removed, and all were histologically found to be pilomatricomas. Records obtained from an outside hospital document the prior removal of five additional pilomatricomas. The majority were described as 1 to 2 cm, firm, subcutaneous nodules that were extremely painful to the patient (Fig. 2). In two cases, the patient presented with a red, inflamed nodule that, upon incision and drainage, discharged serosanguinous fluid along with white chalky material. At the time of surgery, the majority of specimens were white, chalky, and easily fragmented (Fig. 3). One lesion "recurred" or more likely several fragments were left behind, and the area had to be reopened. (This lesion was only counted once during data analysis.)

The diagnosis of myotonic dystrophy was made at age 30 by electromyography and clinical findings, although the mother had suspected some sort of abnormality for years. The patient did not learn to walk until 16 months of age and was never as active as the other children (e.g., a poor runner). Upon formal testing, she was found to be "borderline" mentally retarded, and she slept excessively. It was not until the mother learned that an uncle on the patient's father's side had myotonic dystrophy that the correct diagnosis was made. No other relatives appear to be affected by myotonic dystrophy, and the uncle is not known to have any skin nodules. The patient's other medical problems include cataracts, nocturnal enuresis, chest pain, history of herpes zoster, and congestive heart failure.

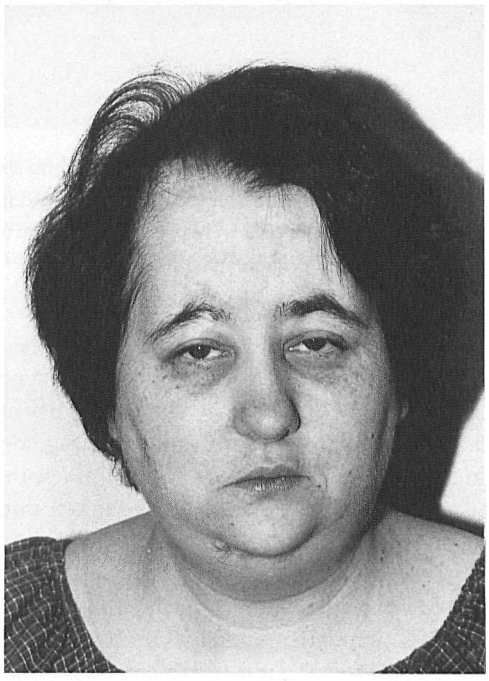

Figure 1. Note the ptosis and expressionless face.

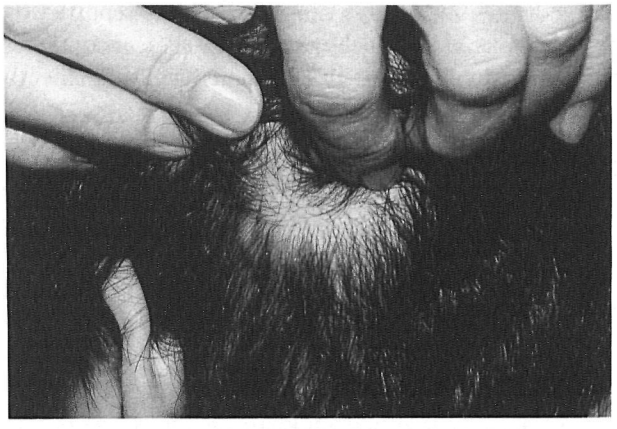

Figure 2. Tender, subcutaneous, hard nodule of the scalp.

## DISCUSSION

Myotonic dystrophy is an autosomal dominantly inherited disease characterized by myotonia or the inability to relax a muscle normally after contraction. Cantwell and Reed[1] were the first to report an association between pilomatricomas and myotonic dystrophy in 1965. Our case represents the highest number of biopsy-proven pilomatricomas in a patient with myotonic dystrophy reported in the English language literature. It also illustrates the tendency for these tumors to cluster within the scalp.

A search of the English language literature from 1965 to 1990 revealed a total of 20 cases of myotonic dystrophy and biopsy-proven pilomatricomas. The data are summarized in Tables 1, 2, and 3.

Myotonic dystrophy is inherited in an autosomal dominant fashion with a highly variable penetrance

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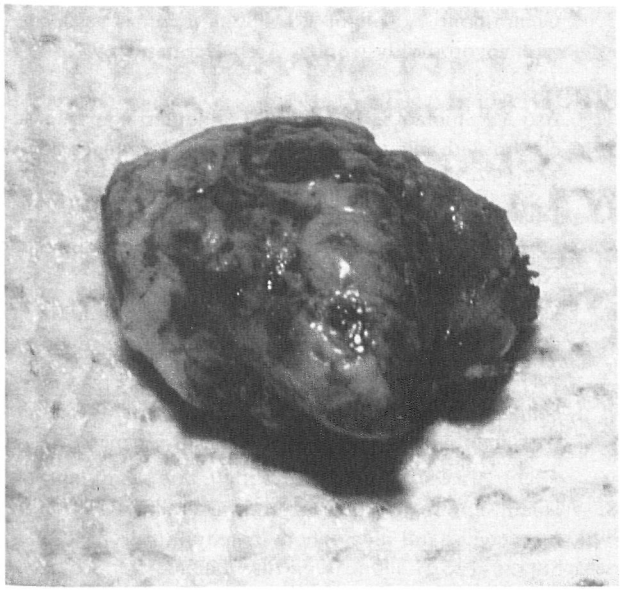

**Figure 3. Irregular, hard, white 2-cm nodule.**

and expressivity.[10,11] The gene defect has been mapped to chromosome 19. Characteristic features include a long, lean, expressionless face with ptosis, mental retardation, cataracts, frontal balding, gonadal dysfunction, hypersomnolence, and muscular abnormalities.

The conventional pilomatricoma is an adnexal tumor with differentiation toward hair cortex, which occurs most commonly in the first or second decade.[9] Sites of predilection include the head (40%) and extremities (38%). Multiple tumors are uncommon, with an incidence of 3.5%.[9] When occurring in association with myotonic dystrophy, several of the above characteristics change. The patients tend to be older, with an average age of onset of 24 years. The majority of patients have a multiplicity of tumors (75%), with 55% having multiple tumors of the scalp. The clinical appearance is not altered, nor is the histology.

The symptoms of myotonic dystrophy usually have their onset in the second or third decade. When pilomatricomas have occurred, they often have appeared within a year or so of the onset of the symptoms of myotonic dystrophy. In several unusual cases, the nodules have preceded by several years the manifestations of myotonic dystrophy. In other cases, a patient's neuromuscular complaints may go undiagnosed for years (as with our patient) allowing the dermatologist to suggest the correct diagnosis.

The reason why approximately 4% of patients with myotonic dystrophy develop pilomatricomas is not known.[2] It has been suggested that this tendency is a further pleiotropic effect of the myotonic dystrophy gene, much like the tendency to develop cataracts, frontal balding, or testicular atrophy.[2] The predilection for the scalp may somehow be related to the high density of hair follicles in that region.

**Table 1. Pilomatricomas and Myotonic Dystrophy**

| *Reference Number* | *Onset of First PM* (years)* | *Total No. of PMs* | *Sex* | *Site (N)* |
|---|---|---|---|---|
| 1 | 50 | 4 | M | Scalp (2),arm (2) |
| 2 | | 1 | F | Arm (1) |
| | | 1 | F | Scalp (1) |
| | | 16 | M | Scalp (13),arm (2), back (1) |
| | | 1 | F | Scalp (1) |
| | | 2 | F | Face (1), neck (1) |
| | | 2 | M | Back (2) |
| | | 2 | M | Scalp (2) |
| 3 | 22 | 5 | M | Scalp (5) |
| | 32 | 2 | F | Scalp (2) |
| 4 | 40 | 7 | F | Scalp (6), neck (1) |
| 5 | 15 | 1 | M | Buttocks (1) |
| | 20 | 14 | M | Scalp (10), neck (1), limb (1), pubic (1), head (1) |
| | 14 | 8 | M | Scalp (8) |
| | 3 | 1 | F | Neck (1) |
| 6 | 31 | 11 | M | Scalp (10), pubic (1) |
| | 14 | 8 | M | Scalp (8) |
| 7 | 39 | 3 | M | Arm (2), neck (1) |
| 8 | 19 | 5 | F | Scalp (1), face (1), neck (3) |
| † | 16 | 24 | F | Scalp (11), face (6), back (1), neck (2), breast (1), abdomen (1), arm (1), leg (1) |

* PM = pilomatricoma; † present case; N, number of PM at that site.

**Table 2. Summary of Pilomatricomas Occurring in Myotonic Dystrophy**

| | |
|---|---|
| Total number of pilomatricomas | 118 |
| Total number of patients | 20 |
| Average per patient | 5.9 |
| Man to woman ratio | 11:9 |
| Percent with multiple tumors | 75% (15/20) |
| Percent with multiple tumors of the scalp | 55% (11/20) |
| Average age onset | 24 years |

**Table 3. Distribution of Pilomatricomas in Myotonic Dystrophy**

| *Site* | *Percentage (N)** | |
|---|---|---|
| Scalp | 68 | (80) |
| Neck | 8 | (10) |
| Face | 7 | (8) |
| Arm | 7 | (8) |
| Back | 3 | (4) |
| Pubic region | 2 | (2) |
| Breast | 1 | (1) |
| Abdomen | 1 | (1) |
| Buttocks | 1 | (1) |
| Leg | 1 | (1) |
| Not fully specified | 2 | (2) |

*Total number of pilomatricomas = 118.

## CONCLUSIONS

This patient illustrates the striking tendency for some patients with myotonic dystrophy to develop pilomatricomas. This characteristic of the myotonic dystrophy gene has a low penetrance, as only a small percentage of patients develop these tumors. The clinical presentation of these patients with multiple, hard, and sometimes tender subcutaneous nodules, especially of the scalp, may be quite impressive and may require extensive surgical intervention. Thus, the dermatologist may be in a unique position to participate in the management of the myotonic dystrophy patient. Additionally, in rare instances, the dermatologist may be able either to predict the onset or recognize the presence of this unusual neuromuscular disease.

## REFERENCES

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### Venereal Disease Clinics

In fact, there were in 1919 only 154 special clinics throughout the country involved in the treatment of syphilis and gonorrhea, and many private practitioners continued to refuse to treat patients suffering from venereal disease. One of the consequences of the war to which Winslow made reference was the enactment of the Chamberlain-Kahn Bill by the U.S. Congress in 1918; it provided for the creation of an Interdepartmental Social Hygiene Board at the federal level to carry out its mandate. Funds were made available to the states on a matching basis; through this vehicle most of the states initiated programs for the treatment of patients with venereal disease. As of 1922, U.S. physicians were administering six million doses of arsphenamine annually. And by 1934 there were four times as many venereal disease clinics in operation throughout the country as there had been in 1919. However, it was only after the passage of the National Venereal Disease Control Act in 1938 that a comprehensive assault upon this scourge was undertaken. Wartime developments shaped the postwar convictions of the two founders of the Public Health Institute and influenced the course they pursued. *From Seipp C. Organized medicine and the Public Health Institute of Chicago. Bull Hist Med 1988;62:429–449.*