

# Stapedial Ankylosis in the Mayer-Rokitansky-Küster-Hauser Syndrome

Cor W. R. J. Cremers, MD, PhD; Ernst H. Strübbe, MD, PhD; Wim N. P. Willemsen, MD, PhD

**W**e describe a 21-year-old woman with congenital unilateral conductive hearing loss and an atypical form of the Mayer-Rokitansky-Küster-Hauser syndrome. To our knowledge, this is the first reported case of this syndrome in which surgery for congenital stapedial ankylosis was successful. Besides aplasia of the vagina and uterus, the patient also had various other anomalies, such as the Klippel-Feil syndrome, Sprengel's deformity, and congenital stapedial ankylosis. Congenital hearing loss is an associated characteristic of the Mayer-Rokitansky-Küster-Hauser syndrome (10% to 20% of cases), particularly in the atypical form. Against the background of the favorable results of surgery for isolated unilateral congenital stapedial ankylosis and other unilateral congenital anomalies of the middle ear that have been described in the literature and the significant advantages of bilateral hearing, we used stapedectomy to successfully treat this case of Mayer-Rokitansky-Küster-Hauser syndrome with unilateral congenital stapedial ankylosis.

(*Arch Otolaryngol Head Neck Surg.* 1995;121:800-803)

The congenital absence of the vagina and a rudimentary uterus are the most distinct characteristics of the Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome.<sup>1-3</sup> On the basis of laparoscopic findings, it is now possible to make a distinction between a typical and an atypical form of the syndrome.<sup>4-6</sup> The typical form is characterized by symmetrical muscular buds and normal fallopian tubes. The atypical form shows asymmetrical uterine remnants (aplasia of one or both muscular buds; one bud is smaller than the contralateral one) and/or abnormal fallopian tube development (aplasia/hypoplasia of one or both fallopian tubes).

An earlier study showed that hearing loss was an associated feature (10% to 20%) in patients with the MRKH syndrome.<sup>7</sup> Congenital deafness was found in five of 51 women with the MRKH syn-

drome who underwent systematic hearing tests.

Patients with the atypical form of MRKH syndrome are likely to show congenital hearing loss caused by atresia of the external auditory canal<sup>7,8</sup> or solely by fixation of the ossicular chain. One case of pure inner ear deafness was also found.<sup>7</sup> Other features that may be found in association with the atypical form of the MRKH syndrome include congenital anomalies of the uropoietic tract,<sup>6</sup> such as renal agenesis or ectopia, and congenital spinal abnormalities,<sup>9</sup> such as the Klippel-Feil syndrome, scoliosis, and other vertebral anomalies. Extravertebral features have also been noted, such as abnormalities of the upper extremities and the hands<sup>10</sup> and asymmetry of the face.<sup>8</sup> Congenital sensorineural deafness and congenital conductive deafness are associated features of the Klippel-Feil syndrome and the Wildervanck syndrome. Surgical results have been reported for these syndromes.<sup>11,12</sup> To our knowledge, this is the first article to present the surgical findings and results for congenital conductive deafness in a patient with the MRKH syndrome.

From the Departments of Otolaryngology (Dr Cremers) and Obstetrics and Gynecology (Dr Willemsen), University Hospital Nijmegen (the Netherlands), and the Department of Radiology, Rijnstate Hospital, Arnhem, the Netherlands (Dr Strübbe).



Figure 1. A 21-year-old woman with the Mayer-Rokitansky-Küster-Hauser syndrome. Note short neck and Sprengel's deformity.

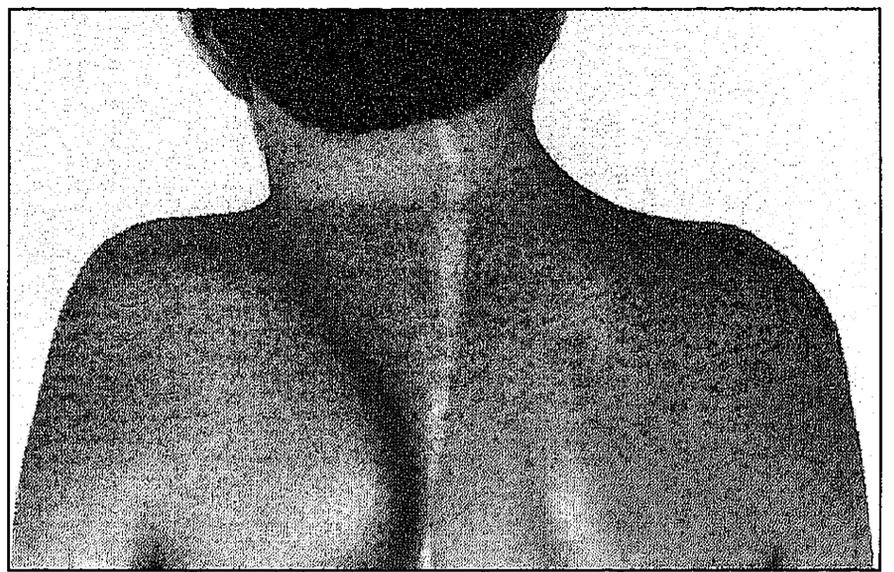


Figure 2. A 21-year-old woman with the Mayer-Rokitansky-Küster-Hauser syndrome. Note short neck. The scar is the result of a Woodward plasty at the age of 6 years.

## REPORT OF A CASE

A 21-year-old woman was referred to our department to take part in a systematic study on hearing function in patients with the MRKH syndrome (Figure 1 and Figure 2).

She had first consulted a gynecologist because of primary amenorrhea. She denied any cyclic lower abdominal complaints. It should be noted that the pregnancy that led to her birth was uneventful, and there was no exposure to medication. Gynecologic examination revealed a blind-ended vagina with a depth of 2 cm. During the rectal examination, no uterus was palpable.

Ultrasound scanning and subsequent laparoscopy confirmed the diagnosis of MRKH syndrome. The patient showed the symptoms of the atypical form of the syndrome. On the right, a rudimentary uterine bud was observed with a normal fallopian tube, whereas the left side lacked a uterine bud and a fallopian tube. The right ovary looked normal, but the left one was fusiform. Adjuvant secretory urography revealed renal agenesis on the left side.

The Klippel-Feil syndrome (congenital fusion of several vertebrae) and kyphoscoliosis owing to malformed vertebrae were visible on roentgenograms of the whole vertebral column. Sprengel's deformity was also present, but there was no radius aplasia or visible abnormalities of the hands.

The patient's face was symmetrical, without any sign of hemifacial microsomia. The auricles had developed normally. The function of

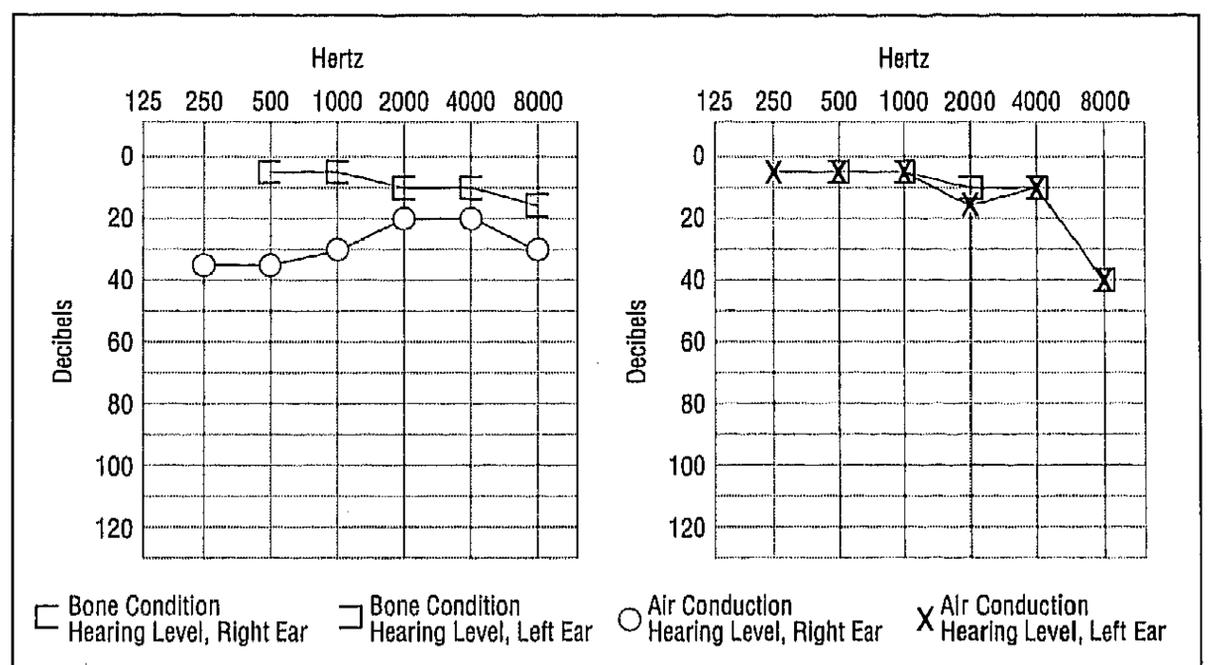


Figure 3. Preoperative tone audiogram.

the cranial nerves was normal, and there was no abducens paralysis. Otologic examination revealed that the tympanic membranes and the malleus handle were normal and that the middle ear had a pneumatized aspect. Tone audiometry demonstrated a large conductive hearing loss of almost 30 dB on the right side (Figure 3). Stimulation with 120 dB on the right ear produced contralateral stapedial reflexes in the left ear, while stimulation with 120 dB on the left ear produced no contralateral reflexes in the right ear for the four frequencies measured. Impedance curves showed normal movement of the middle ear system. Vestibular examination, including electronystagmography and the rotating chair test, and caloric stimulation showed normal stimulation of both labyrinths. No abnormalities were seen on the computed tomographic scans of the petrous bones. In particular, the form and diameter of the internal

auditory canal were normal (Figure 4).

Explorative middle ear surgery was performed because a congenital anomaly of the ossicular chain was suspected. It was possible to see the contours of the incus through the translucent tympanic membrane during inspection of the tympanic membrane with a microscope equipped with a bright light. Isolated stapedial ankylosis was suspected and was indeed found during surgery. The form and mobility of the malleus and the incus were normal. The chorda tympani was also normal, and care was taken to preserve it. The structure of the stapes was slender and correctly formed, but there was bony fixation of the footplate and it became thicker toward the posterior end. There were no other anatomical abnormalities in the middle ear. A stapedotomy opening was made in the middle of the footplate, and the crura

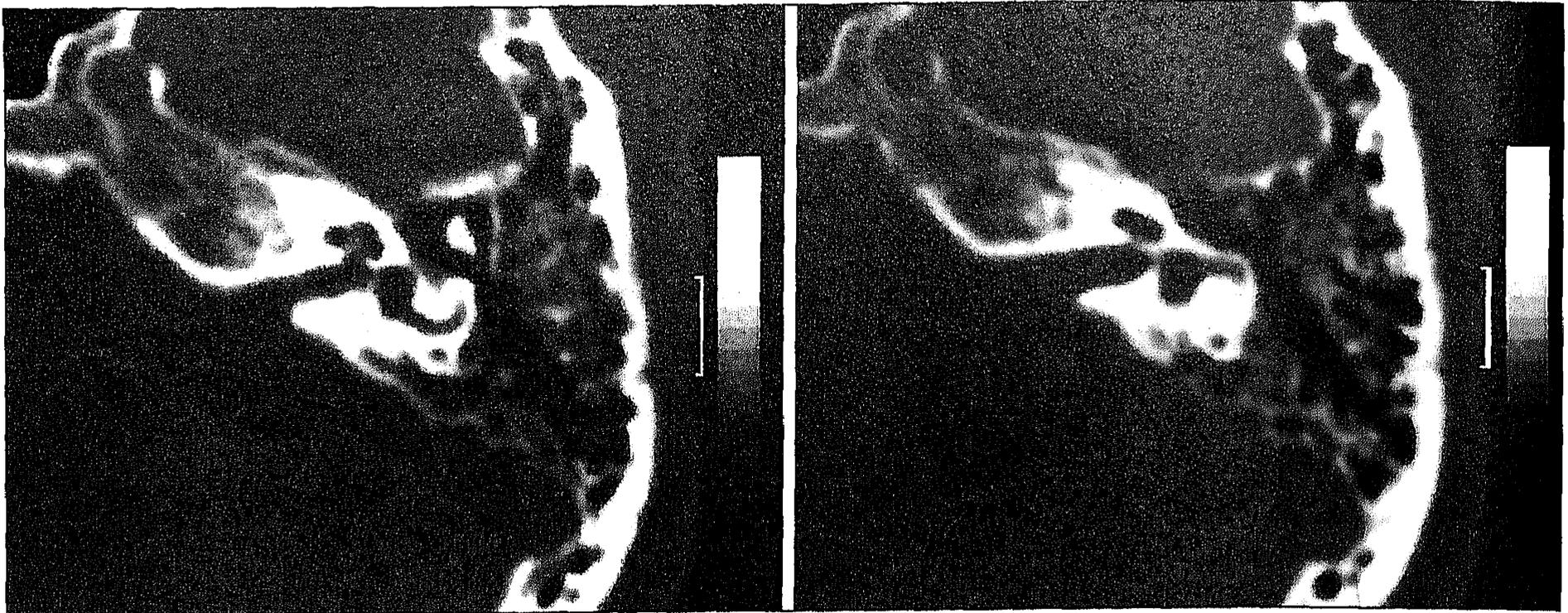


Figure 4. Two normal computed tomographic scans of the left internal acoustic canal, cochlea, and vestibule.

were removed. A 0.4-mm-thick Teflon-platinum piston with a length of 4.75 mm measured from the opening was fixed to the long process of the incus and protruded a sufficient distance into the vestibulum. This procedure led to considerable improvement in the patient's hearing, and at the 3-year follow-up, her air conduction component was less than 10 dB (Figure 5). She did not suffer from postoperative dizziness and was able to taste normally.

#### COMMENT

Isolated stapedial ankylosis was the cause of congenital conductive hearing loss in 44 (31%) of a series of 144 consecutive ears with a congenital middle ear anomaly.<sup>13</sup> Generally, this type of anomaly can be treated successfully by surgical intervention.<sup>13,14</sup> The most dreaded complication after the stapes footplate is opened is a severe stapes gusher, as was described previously in a patient with the Klippel-Feil syndrome.<sup>15</sup> On the basis of findings from the X-recessive hereditary syndrome with progressive mixed hearing loss,<sup>16</sup> it is assumed that the risk of encountering this complication can be traced by meticulous computed tomographic scanning of the internal auditory canal. Several studies have shown that the stapes gusher in this X-recessive syndrome is caused by flushing of cerebrospinal fluid into the vestibulum via the di-

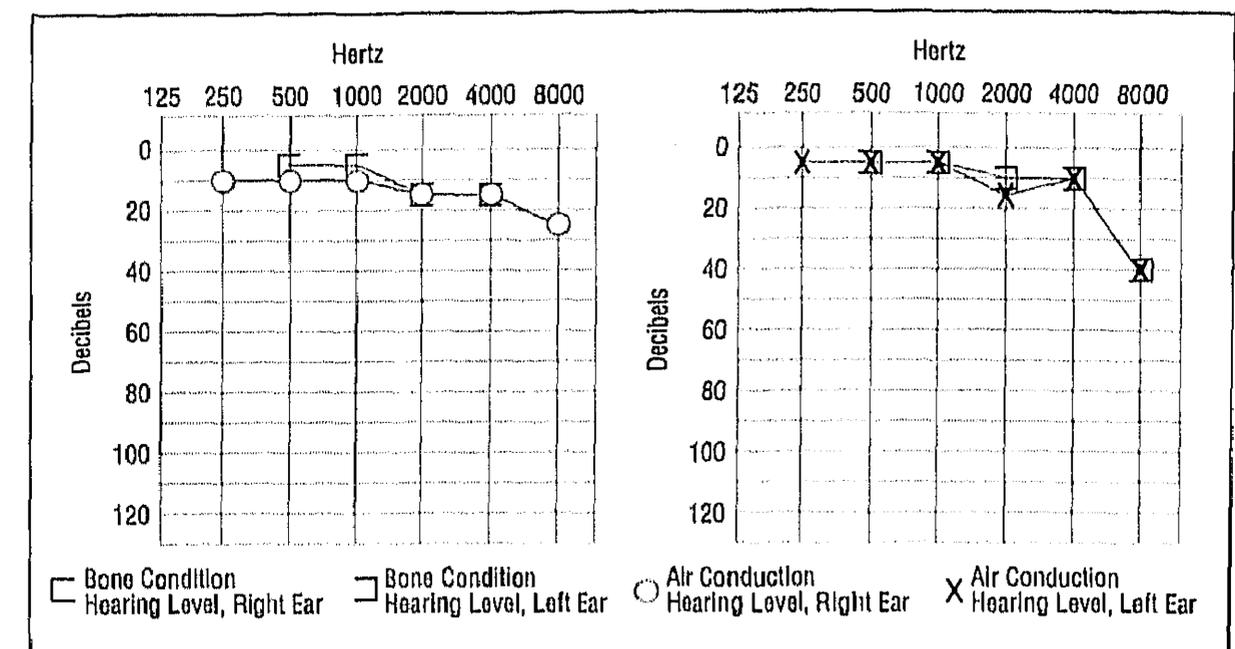


Figure 5. Postoperative tone audiogram obtained at 3-year follow-up.

lated internal auditory canal.<sup>17-20</sup> In cases where unilateral congenital conductive hearing loss is corrected successfully by surgery, it appears that the treated ear not only hears well, but that it also makes a considerable contribution to binaural hearing.<sup>21,22</sup> Such observations are an inducement for otologists to offer patients with unilateral congenital conductive hearing loss the chance of reconstructive middle ear surgery, even if the hearing loss is associated with the Klippel-Feil syndrome and whether or not it is associated with other congenital anomalies, on the condition that the results of thorough preoperative investigation, including radiologic examination of the internal auditory canal, are favorable.

Accepted for publication February 14, 1994.

Reprint requests to Department of Otolaryngology, University Hospital, PO Box 9101, 6500 HB Nijmegen, the Netherlands (Dr Cremers).

#### REFERENCES

- Chervonak FA, Stangol JJ, Nemecek M, Amin HK. Mayer-Rokitansky-Küster-Hausner syndrome: congenital absence of vagina. *N Y State J Med.* 1982; 82:23-26.
- Griffin JE, Edwards C, Madden JD, et al. Congenital absence of the vagina: the Mayer-Rokitansky-Küster-Hausner syndrome. *Ann Intern Med.* 1976;85:224-236.
- Willemsen WNP. *Neovaginoplastiek met peritoneumtranspositie.* Nijmegen, the Netherlands: University of Nijmegen; 1982. Thesis.
- Willemsen WNP, Dony JMJ. Een deconium ervaring met de behandeling van hypo- en aplasia van de vagina met de neovaginoplastiek vol-

- gens Davydov en met de (niet operatieve) methode van Frank. *Ned Tijdschr Geneesk.* 1988; 132:1199-1202.
5. Schmid-Tannwald I, Hauser G. Deutung der 'atypischen' Formen des Mayer-Rokitansky-Küster Syndrom. *Geburtshilfe Frauenheilkd.* 1977; 37:386-392.
  6. Strübbe EH, Willemsen WNP, Lemmens JAM, Thijn CJP, Rolland R. Mayer-Rokitansky-Küster syndrome: distinction between two forms based on excretory urographic, sonographic and laparoscopic findings. *AJR Am J Roentgenol.* 1993;160:331-334.
  7. Strübbe EH, Cremers CWRJ, Dijkers FG, Willemsen WNP. Hearing loss and the Mayer-Rokitansky-Küster-Hauser syndrome. *Am J Otol.* In press.
  8. Willemsen WNP. Renal-skeletal-ear and facial anomalies in combination with the Mayer-Rokitansky-Küster (MRK) syndrome. *Eur J Obstet Gynecol Reprod Biol.* 1982;14:121-130.
  9. Strübbe EH, Lemmens JAM, Thijn CJP, et al. Spinal abnormalities and the atypical form of the Mayer-Rokitansky-Küster-Hauser syndrome. *Skeletal Radiol.* 1992;21:459-462.
  10. Strübbe EH, Thijn CJP, Willemsen WNP, Lappöhn R. Evaluation of radiographic abnormalities of the hand in patients with the Mayer-Rokitansky-Küster-Hauser syndrome. *Skeletal Radiol.* 1987;16:227-231.
  11. Rijn van P, Cremers C. Surgery for conductive deafness in Klippel-Fell syndrome. *Ann Otol Rhinol Laryngol.* 1988;97:347-352.
  12. Cremers C, Hoogland G, Kuljpers W. Hearing loss in the cervico-oculo-acoustic (Wildervanck) syndrome. *Arch Otolaryngol.* 1983;110:54-57.
  13. Teunissen E, Cremers CWRJ. Classification of minor congenital ear anomalies: a report of 144 ears. *Ann Otol Rhinol Laryngol.* 1993;102:606-612.
  14. Teunissen E, Cremers CWRJ, Huygen PLM, Pouwels TPBM. Isolated congenital stapes ankylosis: surgical results in 32 ears and a review of the literature. *Laryngoscope.* 1990;100:1331-1336.
  15. Danilidis J, Maganaris T, Dimitriadis A, Lladas T, Manolidis L. Stapes gusher and Klippel-Fell syndrome. *Laryngoscope.* 1978;88:1178-1183.
  16. McKusick VA. *Mendelian Inheritance in Man.* 10th ed. Baltimore, Md: Johns Hopkins University Press; 1992.
  17. Glasscock ME. The stapes gusher. *Arch Otolaryngol.* 1973;98:82-91.
  18. Cremers CWRJ, Hombergen GCJH, Scaff JJ, Huygen PLM, Volkers WS, Pinckers AJLG. X-Linked progressive mixed deafness with perilymphatic gusher during stapes surgery: preoperatively identifiable specific features. *Arch Otolaryngol.* 1985; 111:249-254.
  19. Schuknecht HF, Reisser C. The morphologic basis for perilymphatic gushers and oozers. *Adv Otorhinolaryngol.* 1988;39:1-12.
  20. Michel O, Breunsbach J, Mattias R. Das angeborene Liquordrucklabyrinth. *HNO.* 1991;39:486-490.
  21. Snik AFM, Teunissen E, Cremers CWRJ. Frequency resolution after successful surgery in congenital ear anomalies. *Scand Audiol.* 1991;20: 265-267.
  22. Snik AFM, Teunissen E, Cremers CWRJ. Speech recognition in patients after successful surgery for unilateral congenital ear anomalies. *Laryngoscope.* In press.

## Only in JAMA . . .

*Theme issues concentrate on critical issues in medicine today. Recently, JAMA: The Journal of the American Medical Association has explored these pertinent health topics, always from a clinical perspective and frequently in the full context of the social, economic, and political factors that affect them:*

- Genetics and Molecular Medicine
- Perioperative Myocardial Ischemia
- Immunization
- Tobacco
- Medical Education
- HIV/AIDS
- Guidelines for Cardiopulmonary Resuscitation and Emergency Cardiac Care
- Violence
- Mental Health
- Allergic and Immunologic Diseases

*Plus, each year JAMA publishes its important CONTEMPO issue. This valuable issue highlights the latest developments in over 40 key medical specialties.*

Don't miss a single issue. For important scientific medical information you cannot afford to miss, subscribe to JAMA today!

**For information about subscribing, call 1-800-AMA-2350.**