FAMILIAL DEAFNESS: A POSSIBLE EXAMPLE.*†

C. C. BUNCH, PH. D., St. Louis.

The complete chain of facts definitely proving the hereditary transmission of deafness is rarely if ever secured. Regardless of this fact, certain types are, on the basis of the evidence in the literature, usually considered hereditary, and when other possible causes have been eliminated, the otologist is compelled to consider heredity the etiological factor even though the actual proof is wanting. For example, much evidence is available which indicates rather strongly that otosclerosis is a disease transmitted by heredity.[1] In fact, many otologists will make a diagnosis of otosclerosis only where the evidence of hereditary transmission is secured.

Another type of deafness usually considered hereditary is that appearing in the children of deaf-mute parents. But these two types of deafness are not easily mistaken. That resulting from otosclerosis is insidious in its onset, appears most frequently in late adolescence or early adult life and quite consistently yields the Bezold triad in the functional tests of hearing. The deafness appearing in children of deaf-mute parents is usually profound and is evident very early in life. But in neither instance is the evidence of hereditary transmission definitely established, for many children born of otosclerotic and deaf-mute parents have normal hearing. Of the two, Tinkel[2] states that in 38 family history charts for cases of otosclerosis "a larger proportion are affected with this defect . . . than suffer from deafness in families of deaf-mutism."

Another type of deafness which is sometimes considered hereditary is that so classically described by Hutchinson, the deafness of the so-called hereditary syphilis. This type should be mentioned in the same category as those above. It may, however, be more rightly called congenital than hereditary.

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*From the Department of Oto-Laryngology, Oscar Johnson Institute, Washington University Medical School.

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The accuracy of the term *hereditary* in describing these types of deafness is open to criticism. Genetic studies, such as that of Davenport and others, reveal strikingly the difficulties in the way of the solution of the problem. The use of the term *familial* is perhaps more accurate of description, but it adds little or nothing to our knowledge of the etiology.

A fourth type of deafness of somewhat similar character is less frequently encountered, is recognized with greater difficulty and may be improperly diagnosed even with most thorough study. Dr. L. W. Dean and the writer,[3] in 1923, reported a study of the auditory acuity in three brothers, 8, 11 and 16 years of age, from a family of seven children. The deafness in each case was slight when discovered and appeared to be slowly progressing. The parents were not deaf nor was their marriage consanguineous. The Wassermann tests on the boys and their father gave negative results. No treatment instituted had any effect on the progress of the deafness. The diagnosis was neurolabyrinthitis, etiology unknown. The same study contained a report of similar deafness in two children and in their mother and maternal grandmother. The diagnosis was hereditary neurolabyrinthitis.

Macfarlan,[4] in 1927, under the title, "Identical Hearing in Identical Twins," showed the audiograms secured from tests on middle-aged twin women with this comment: "The hearing rises and falls across the pitch range in nearly exact correspondence one with the other," but he makes no comment as to the probable etiology. Shambaugh and Shambaugh[5] also reported cases of deafness in identical twin women, 31 years of age, and in twin men, 69 years of age. Concerning the deafness in the women, they state that "both gave the functional reactions of primary nerve deafness." Concerning the etiology of the deafness in the men, the writers conclude that it is "also of primary nerve type" and since there was a history of similar deafness in other members of the family, they were led "to question the possibility of some of these cases being the labyrinthine type of otosclerosis."

Rodin[6] in the same year also reported similar deafness in twin girls, 15 years of age. The Rinné test gave negative results and the audiograms showed quite uniform loss for all tones. He concluded tthat the diagnosis was otosclerosis.

This study concerns the hearing of five sisters, 6, 10, 12, 14 and 17 years of age at the time of the first examinations. The father is a factory employe, 42 years of age. The mother is 39 years of age.

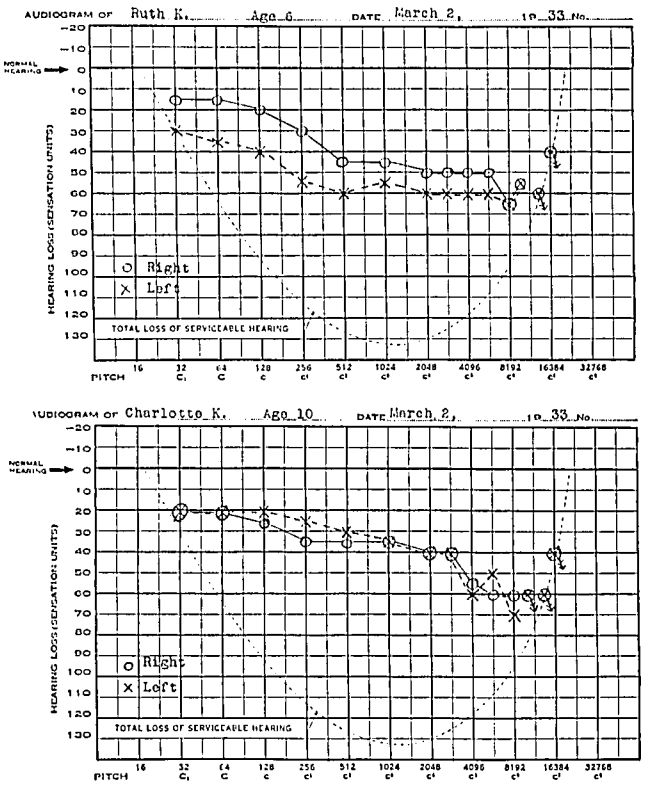

Fig. 1. Audiograms of two sisters, Ruth, age 6, and Charlotte, age 10, at the time of the first examination. Low tones are heard well by both, but the acuity diminishes for tones of higher pitch.

Ruth, age 6 years, and Charlotte, age 10 years, were brought to the Oto-Laryngologic Clinic of Washington University on the advice of Dr. Max A. Goldstein and Dr. Allan Potter. They presented audiograms secured in tests by Dr. Goldstein and his associates. The hearing tests were repeated in the clinic and found to be essentially the same as those secured by Dr. Goldstein. The results of these tests are shown in Fig. 1. The results show that the loss in acuity is bilateral. Low tones are heard well in both ears of each child, and the deafness

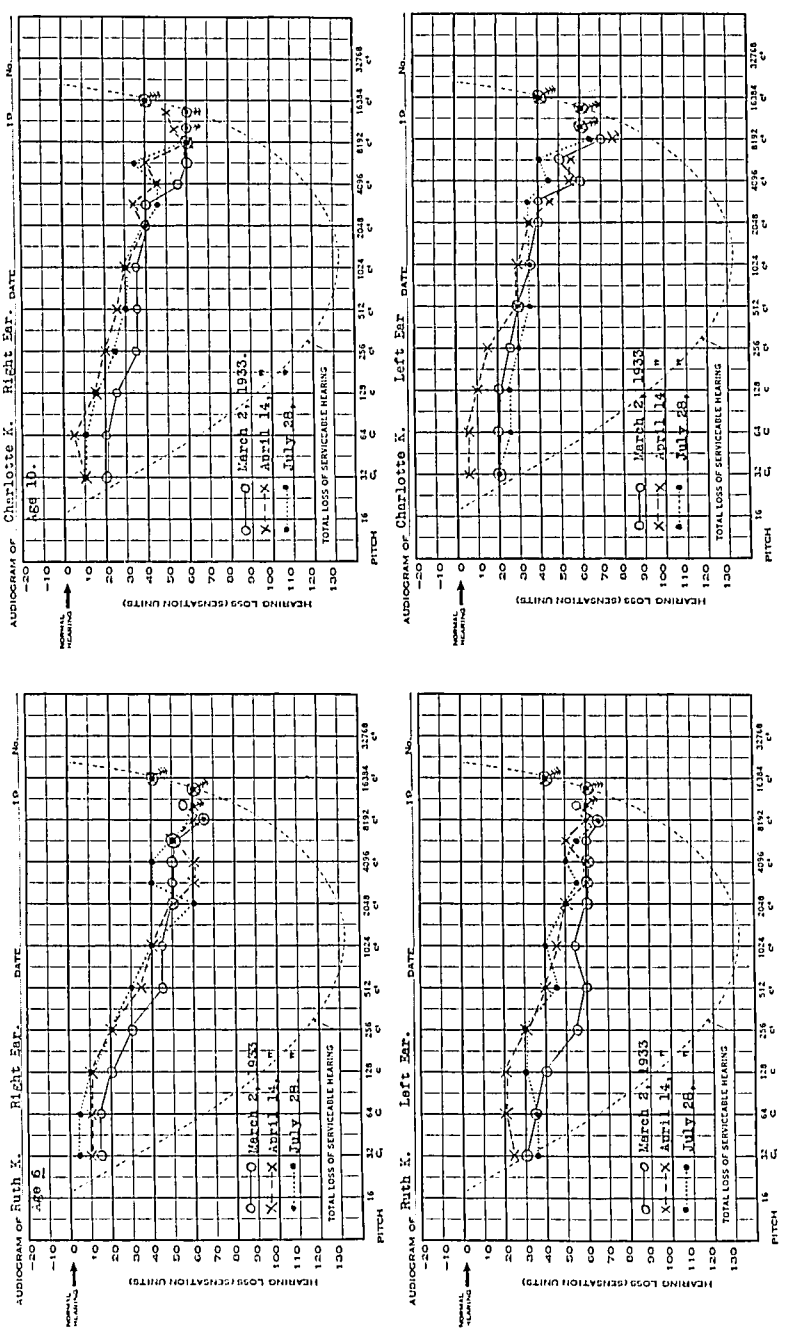

Fig. 2. Audiograms of Ruth and Charlotte showing improvement in hearing on the first test (April 14) after the removal of tonsils and adenoids. The third test (July 28) shows less definite improvement.

increases with the frequency, becoming quite marked for tones of very high pitch. The Rinné test gave the positive result and the Weber test gave no lateralization. The laryngologic examination revealed very bad tonsils and adenoids in each case. A diagnosis of acoustic neuritis was made and it was recommended that the diseased tonsils and adenoids be removed with the thought that the deafness was resulting from some toxic focus, most probably located in the diseased tonsils and adenoids. When the children entered the hospital to have this work done, it was found that the older child had some infection of the maxillary antra and her urine showed four plus albumin, but there were no red blood cells or casts. The Wassermann tests were negative. Both children had their tonsils and adenoids removed, and in the older child antral windows were made for irrigation and to secure better drainage. Their course in the hospital was uneventful and both were discharged in a few days. Six weeks after the first test had been made in the clinic, the children returned for examination and to have the hearing tests repeated in order to determine if there had been any change in acuity of hearing resulting from the removal of the toxic foci. Fig. 2 shows the records of the tests made before and after the operative work, together with the results of a third test made six weeks later. These charts show that the best record was made on April 14, a short time after the operative work. Changes appearing between April 14 and July 28 are too small to be definite. When the second test showed slight but apparently definite improvement, it was considered that the correct diagnosis has been made and that the improvement would continue. But when the results shown by the third test were secured, considerable doubt was expressed.

About this time, the children's mother reported that she thought her oldest daughter's hearing (Anna Marie, age 17 years) might perhaps not be as acute as it should be. She was requested to bring this daughter to the clinic for study. This was done and the results of the hearing tests are shown in Fig. 3. This girl had completed her high school course and was taking an advanced course in business training. The very evident progress she had made in her school work seemed to indicate that her deafness was of recent origin.

When this record was secured, more doubt than ever was expressed as to the accuracy of the original diagnosis in the

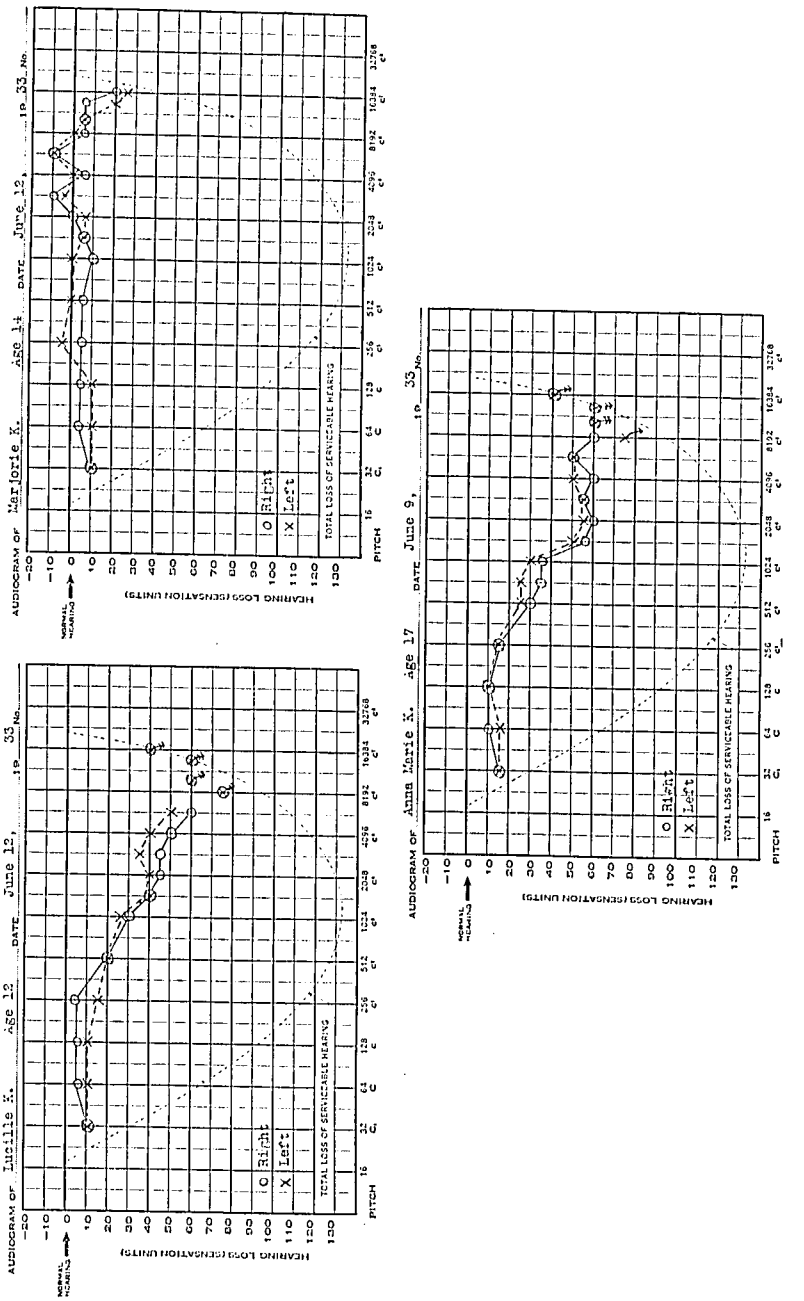

Fig. 3. Audiograms of Lucille, age 12; Marjorie, age 14, and Anna Marie, age 17, sisters of Ruth and Charlotte, whose records are shown in Figs. 1 and 2. Note that Marjorie has essentially normal hearing.

case of the two younger children. The records of all three showed the same general type of hearing loss and the question was then raised as to whether or not we were dealing with some form of familial deafness. The mother was asked if there were other children in the family, and replied that there were two more girls, but that their hearing appeared

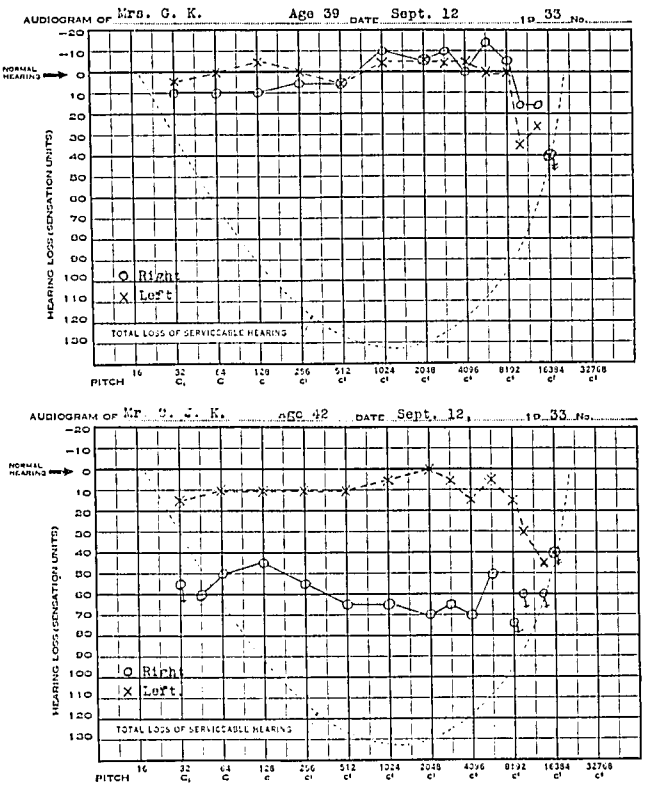

Fig. 4. Audiograms of the mother and father of the children whose records are shown in the preceding figures.

to be normal. She agreed, however, to bring them for examination. The audiograms showing the results of the tests of these two are also shown in Fig. 3 (Lucille, age 12 years, and Marjorie, age 14 years). That of Lucille shows in general the same type of hearing loss as that shown by Ruth, Charlotte and Anna Marie. The record of Marjorie, while showing a very slight loss for almost all tones, is practically normal, as is her record for the 4A (phonographic) audiometer.

In order to secure all the information possible as to the probable etiology of this deafness, audiometric tests were done on the father and mother. These records are shown in Fig. 4. The curves in the case of the mother show that the hearing for low tones in the right ear is slightly less acute than that in the left, but differences of this extent and magnitude are frequently encountered and are usually considered insignificant. The loss shown in the father's right ear is accounted for by a history of frequent attacks of otitis media in childhood and youth, and the record showing a rather marked loss for all tones in this ear confirms this history. Neither the father or the mother show losses similar to those secured from the tests of the four children.

Summary: Records secured by means of audiometric tests are given, showing a rather marked bilateral loss of auditory acuity in four girls of a family of five. The similarity of the auditory fields, the lack of a similar loss in records secured from tests of the parents and the absence of a history of deafness in other generations of the family in so far as could be determined have necessitated the use of the term *familial* deafness rather than *hereditary* deafness. The fifth child of the family had practically normal hearing. It is possible that something neither familial or hereditary may be the etiological factor in these cases, but thorough examinations by pediatricians in two of them, internists in one and nasopharyngeal in all four, have not revealed any common etiology. There is no history of common illness, the use of drugs or indications of dietary deficiency. For these reasons the deafness has been called *familial.*

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Scott and Euclid Avenues.