was no objective change. She remained pleased to have had the partial Gasserian neurectomy.

Discussion

Hemiatrophy of the body is usually facial, and not always progressive and unilateral. Like other hereditary degenerations, most cases begin near adolescence. Wartenberg (1945) reviewed the subject thoroughly. The present case has the typical features of onset at puberty, scleroderma, fits, trigeminal neuralgia, muscular spasms, sweats, and slow progression.

There is no evidence of involvement of the internal organs in the hemiatrophy. The E.E.G. showed no evidence of epilepsy. The findings on myography are not clear. Possibly there may be a cord lesion on the right, and there is no evidence of an upper motor neurone lesion affecting the muscles of the left side. Cases in which this occurs have been described by Brain (1955), Reider and Player (1946), and Wartenberg (1945), who quoted several other instances. The operative results suggest that the cause of the trigeminal neuralgia was peripheral.

The segment of scleroderma on the opposite side of the body suggests that in this case the factor controlling the process is not confined to the right. It is assumed that the scleroderma and the hemiatrophy have a common origin on the evidence collected by Wartenberg (1945). Urechia and Retezeanu (1936) have reported a case of right hemi-facial atrophy with atrophy of the opposite arm, and Thoma (1954) mentions others in which the hemiatrophy was patchy and bilateral.

The spontaneous fractures of the jaw, with which this case presented, are believed not to have been described before in a case of hemiatrophy of the body. The cause was probably intense spasm of the right side of the face due to the trigeminal neuralgia, often found in this condition, acting on a jaw weakened by the hemiatrophy.

Summary

A case of progressive hemiatrophy is described, in which the patient twice suffered a spontaneous fracture of the edentulous mandible on the affected side, attributed to facial spasm associated with trigeminal neuralgia.

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Since the discovery of the curative powers of the sulphones, interest in leprosy has increased, campaigns have been launched bringing as many as possible of the estimated 10 to 12 million people affected throughout the world under treatment, and more doctors are engaged in research into the unsolved problems of the disease. W.H.O. has played a leading part in spreading knowledge of advances in treatment and in helping organize leprosy campaigns in countries where the disease is an important public-health problem, and its work is described in the booklet *International Work in Leprosy, 1948-59*, and is set against the background of a short history of leprosy, its incidence, its treatment, and the research and other problems it raises. (Available from H.M.S.O., P.O. Box 569, London, S.E.1.)

THE INHERITANCE OF ESSENTIAL PENTOSURIA

BY

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Essential pentosuria is a rare condition characterized by the constant excretion of xyloketose in the urine. Included by Garrod (1923) among the inborn errors of metabolism, the condition probably results from a block in the metabolism of xyloketose, one of the normal products of glucuronic acid metabolism (Enklewitz and Lasker, 1935; Flynn, 1955).

This report describes the finding of xyloketosuria in a mother and her two children.

Methods.—The following methods were used to investigate the reducing substance in the urine: (1) qualitative Benedict's test for urine reducing substance; (2) Bial's test for pentoses; (3) the Lasker-Enklewitz test for xyloketose (Lasker and Enklewitz, 1933); (4) osazone preparation; (5) paper chromatography (Horrocks and Manning 1949) using 3:2:1.5 *n*-butanol-pyridine-water solvent (Hough *et al.*, 1950), demonstrating the sugar spots with aniline hydrogen oxalate; (6) quantitative estimation of the urine xyloketose (Lasker and Enklewitz, 1933), using the factor of Greenwald (1930) for the calculation.

Cases

The propositus, a Jewish woman aged 40 (E.R.), was admitted to the National Hospital, Queen Square, complaining of unsteadiness of gait, giddiness, and nausea for two years. The symptoms dated from a hysterectomy for fibroids two years previously, and no organic cause for them was found. During investigation this patient was found to have a reducing substance in the urine; it transpired that this had been first noticed in 1939 and on a number of occasions since, resulting in three normal glucose-tolerance curves and the warning that she was "knocking on the door of diabetes."

On the present admission the urine showed a constant yellowish-green precipitate with Benedict's solution; a glucose-tolerance curve was normal (fasting blood-sugar 98 mg./100 ml.; half-hour, 155 mg.; 1 hour, 138 mg.; 1½ hours, 115 mg.; 2 hours, 112 mg.; 3 hours, 80 mg.), with no alteration of the amount of reducing substance in the urine during the test. The urine gave a positive Bial test for pentose and a positive Lasker-Enklewitz test for xyloketose. An osazone was prepared, showing fine needles similar to glucosazone, but smaller. Glucose-oxidase test paper ("clinitix"), specific for glucose, was negative. Paper chromatography with suitable markers showed a pentose

FIG. 1.—Results of paper chromatography.

in the xyloketose position (Fig. 1). Quantitative estimation showed 0.55 g./100 ml. of this reducing substance in the urine.

Family Investigation.—Investigation of the immediate family revealed xyloketosuria in the patient's two children, a boy aged 12 (H.R.) and a girl aged 9 (R.R.), the methods of testing being those used with the mother. Chromatography of the three positive urines is shown in Fig. 1. Quantitative estimation showed 0.30 g./100 ml. in the boy's urine and 0.13 g./100 ml. in the girl's urine. An

The recessive inheritance of the condition has been generally accepted by other writers, though Schultz (1938) described four cases in two generations—a father and his two sons, and a brother—and concluded that the condition was inherited as a dominant characteristic. Macumber (1949) found xyloketosuria in a father and two sons.

The findings in the present family are consistent with the recessive hypothesis, provided one assumes that the

mating of Case III 13 with Case III 14 (Fig. 2) is between a xyloketosuric who is homozygous with a heterozygote. Both these individuals are Jewish, the parents of Case III 13 (E.R.) coming from a small town near Warsaw and those of Case III 14 from Odessa and Leningrad. Lasker (personal communication) believes that the incidence of xyloketosuria among Jewish people originating from Central and Eastern Europe may be as high as 1 in 1,500. This means that the frequency of heterozygotes must be of the order of 5% of the population and the chance of a xyloketosuric marrying an unrelated heterozygote would not be remote. An attempt

was made by comparing family names to establish a connexion between the present family and some of Lasker's cases originating in Warsaw and Odessa, but no such connexion could be traced.

It is unfortunate that, with the increasing use of glucose-specific enzyme test papers for urine-testing in place of Benedict's or Fehling's tests, these interesting cases will not be recognized. With this type of glucose-specific test other reducing substances in the urine will also be missed, some of which—for example, galactose—are of direct clinical importance.

Summary

The unusual occurrence of xyloketosuria is reported in a mother and her two children. The family had originated from known pockets of high incidence for xyloketosuria in Poland and Russia. An extensive survey of the family showed no further cases.

I am grateful to Dr. Macdonald Critchley for permission to investigate a case under his care and to Professor J. N. Cumings for his advice and encouragement. I thank Dr. Margaret Lasker for most interesting information on her investigations of this condition.

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FIG. 2.—Genealogical tree of family showing xyloketosuria.

extensive survey of the family (Fig. 2), using Benedict's test and the Lasker-Enklewitz test for screening, revealed no further positive cases.

Discussion

Since the first description of xyloketosuria by Salkowski and Jastrowitz (1892), some 200 cases have been reported. In this country Cammidge and Howard (1920) recorded seven cases, but the exact diagnosis in these is uncertain; Flynn (1955) reported two unrelated cases proved by chromatography. Xyloketosuria is an apparently entirely benign condition, with a normal expectation of life (Lasker, 1955).

In the most comprehensive study of xyloketosuria, Lasker *et al.* (1936) reviewed the published cases and reported their own series of 37 individual cases in 20 families. They concluded from their survey that the condition was inherited and probably controlled by a simple recessive gene. They noted that xyloketosuria was very rare in the general population but often found in the families of known cases. Blatherwick (personal communication to Lasker *et al.*, 1936) found an incidence of 1 in 50,000 from Metropolitan Life Insurance records, but admitted that a number of cases excreting less than 0.25% of xyloketose may have been missed. On the other hand, Lasker *et al.* (1936) found 10 new cases on testing 34 sibs of xyloketosuric propositi; in one family 66 individuals were tested and seven cases found. The condition is almost, if not entirely, confined to Jews, though Barnes and Bloomberg (1953) described the condition in an apparently non-Jewish family.

In the series of Lasker *et al.* (1936) xyloketosuria was found in the children of 10 families in which neither parent was positive. In these 10 sibships 16 out of 38 members had xyloketosuria: deducting the eight propositi, there remained 8 out of 30 members. This series included one case, the child of first cousins, one parent being affected.