Psychiatric Clinic, St. Jörgen Hospital, University of Göteborg (Sverige)
(Head: Prof. H. Forssman)

Cutis Verticis Gyrata Thyroaplasia and Mental Deficiency

Hans Olof Åkesson

McDowall was the first to report a combination of cutis verticis gyrata and severe mental deficiency. He described his cases as far back as 1893, but only about thirty more cases have been reported up to 1964. The combination can hardly be as rare as would appear from this, however, for in a census study of all the institutionalized mentally deficient in Sweden I uncovered not less than 47 cases of cutis verticis gyrata (Åkesson, 1964).

Cutis verticis gyrata is not a disease in itself, but a symptom, apparently occurring in several different syndromes. Most commonly, however, when it occurs together with mental deficiency it seems to belong to a hereditary syndrome, which in complete form is also characterized by epilepsy, cerebral palsy and eye defects (Åkesson, 1964).

We know little about the etiologic background of this hereditary syndrome, but several observations point to it being caused by a form of endocrine disorder. This prompts me to report a case in which cutis verticis gyrata and mental deficiency were combined with thyroid aplasia. Fiocco described a case of cutis verticis gyrata, cretinism and mental deficiency in 1913, but as far as I can see, he is the only other author who has reported a case of thyroid deficiency together with the other two disorders.

Case report

The patient was a male, normally delivered after an uncomplicated pregnancy, weighing 3,400 gm at birth, severely retarded from birth. He never learned to talk, did not begin to walk until he was 7, and was incontinent of urine and feces. He was admitted to a home for severely retarded when he was 10. When he was 2 myxedema was observed and he was given thyroid compounds up to the age of 10, without any noticeable improvment resulting.

At 23 he broke his right femur. The fracture healed poorly, and he was bedridden from then on.

I examined him when he was 32. Observations of note were: height 142 cm; arrested physical development like that in cretinism; severe cutis verticis gyrata

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over the vertex; head circumference 56.5 cm; maximum frontal and sagittal measures 15.0 and 18.5 cm respectively; myxedematous skin; hernia throught the linea alba; normal external genitals; normal distribution of hair; heart and lungs normal to physical examination; normal reflexes; no great toe reflex on either side, both hands held in abnormal ulnarward position.

The patient was bedridden, could not talk and had to be fed and tended in every respect. He did not recognize any of the staff, and had no contact with the world around him. He was too backward for ordinary intelligence testing; his IQ was assumed to be between 10 and 20.

Roentgen examination of his skull showed; normal shape, structure and vascularization; no abnormal calcification; enlarged sella turcica measuring 1,600 mm^3 as against a normal maximum of 1,092 mm^3. Standard examination of blood and urine showed nothing of note. Chromatographic urine analysis gave normal results. Total gonadotropins measured $<$ 6 mouse units per 24 hours (low value) and fractional estrogen analysis (estrone, estradiol and estriol) gave normal values.

One day later on in the year the patient suddenly began to have difficulty in breathing and 24 hours later he was dead. Autopsy at the Institute of Pathology at the University of Lund revealed; aplasia of the thyroid gland, generalized arteriosclerosis, purulent tracheo-bronchitis, bilateral lobar pneumonia, signs of heart failure including pulmonary edema and congested internal organs. The brain showed no gross lesions. Microscopic study of skin from the area of cutis verticis gyrata showed a substantially normal scalp (see Figs. 1 and 2).

Genetic analysis

Study of the family data revealed 4 more severely retarded males (Fig. 3). That they were severely retarded is certain, several of their relatives testifying convincingly to this, but it was impossible to obtain any more clinical data about them, for instance, whether or not they showed signs of cutis verticis gyrata or cretinism. Besides these 4 deceased relatives the proband had a cousin dead in infancy who was probably also affected.

It is to late now to determine whether these four relatives had the same disease as the proband. The heaping up of severe mental deficiencies in close relatives, however, points to the operation of the same genetic mechanism in all the affected members.

When a disease is transmitted by X-linked recessive inheritance with full penetrance, 1 out of every 4 children of the female carriers can be expected to be affected, and as the disease only develops in males, one may expect 1 out of every 2 sons to be affected.

The present sibships with affected members contained 5 affected sons, 5 healthy sons, and 11 healthy daughters. Thus 50 per cent of the sons and 24 per cent of the children were affected, which agrees well with the 50 and 25 per cent one would expect with X-linked recessive inheritance.

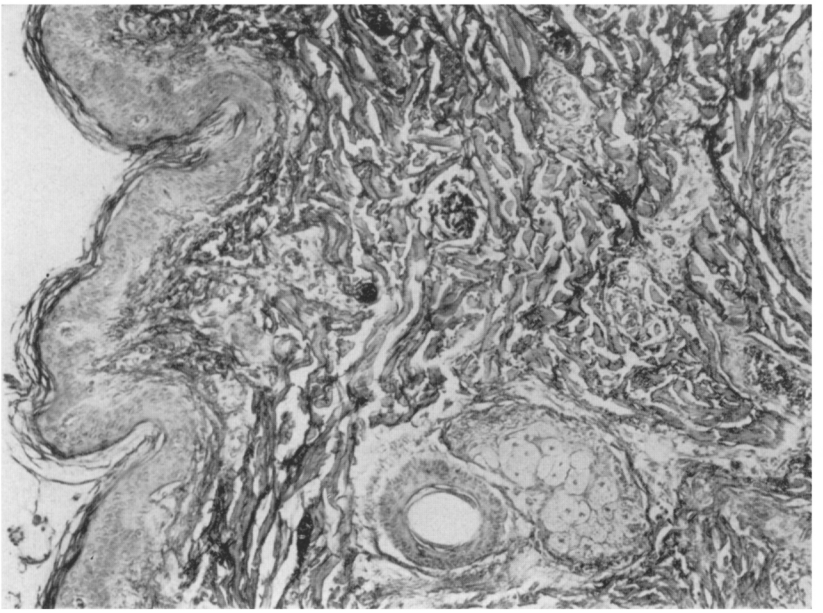

Fig. 2. Skin from gyrus of scalp. Essentially normal histologic picture. Elastin stain. x 100

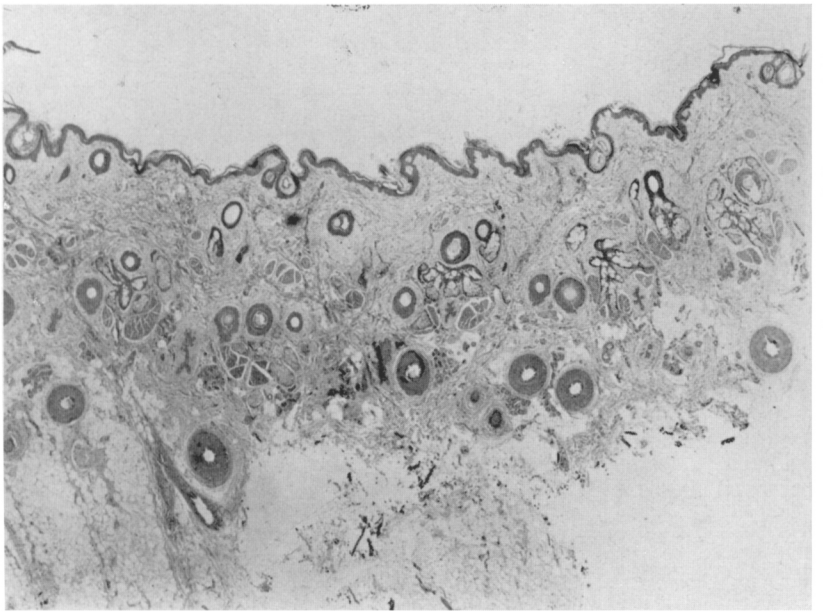

Fig. 1. Skin from gyrus of scalp. Essentially normal histologic appearance. Hematoxylin-eosin stain. x 16

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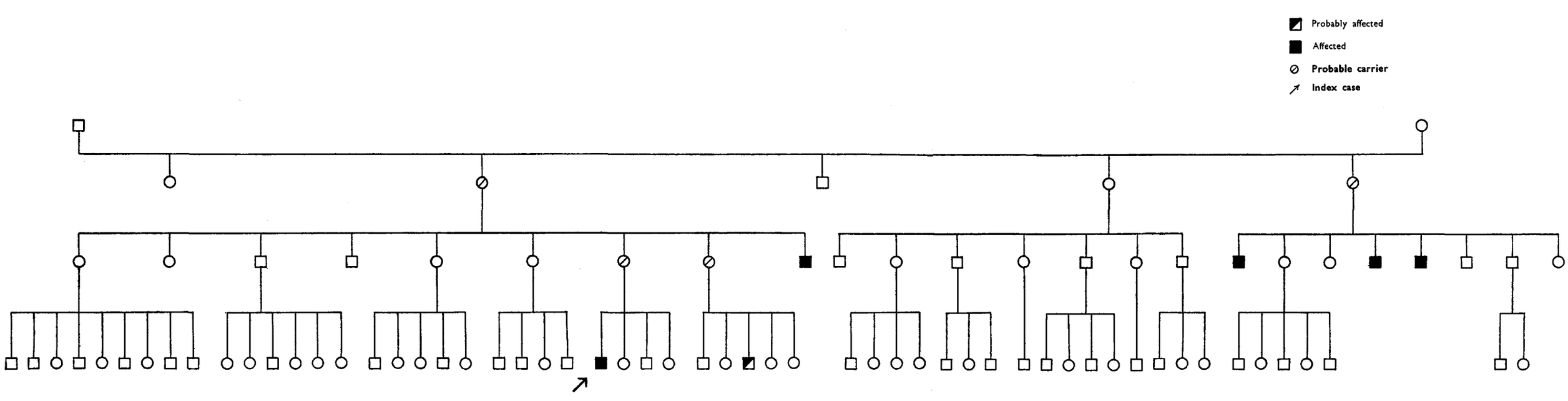

Fig. 3. Pedigree of the investigated family

Thus, there is much evidence in favour of the disease being inherited through an X-linked recessive gene. The cases here are too few, however, to permit any definite conclusions. Sex-limitation cannot be excluded. It should be pointed out that, as a rule, syndromes including a combination of cutis verticis gyrata and mental deficiency are probably not inherited through sex-linked genes. (Åkesson, 1965).

Summary

A man of 32 is described suffering from a combination of three disorders: cutis verticis gyrata, severe mental deficiency and aplasia of the thyroid. Four of his close relatives were also severely retarded. The possibility is discussed of the syndrome being inherited through an X-linked recessive gene.

Acknowledgement

Dr. Joakim Mark of the Institute of Pathology at the University of Lund, who did the autopsy in this case, kindly allowed me to examine the autopsy report and the histologic sections. Miss Ing-Britt Augrell of the same institute photographed the histologic material. I am deeply indebted to them both for their help.

References

1. Åkesson H. O.: Cutis verticis gyrata and mental deficiency in Sweden I. Epidemiologic and clinical aspects. Acta med. scand., 1964, 175: 115.
2. Åkesson H. O.: Cutis verticis gyrata and mental deficiency in Sweden II. Genetic aspects. Acta med. scand., 1965, in press.
3. McDowall, T. W.: Case of abnormal development of the scalp J. ment. Sci., 1893, 39: 62.
4. Fiocco: cit. Spinz O.: Cutis verticis gyrata. Arch. Derm. Syph. (Wien) 1921, 132: 281.

RIASSUNTO

Viene riportato il caso di un uomo di 32 anni affetto da una combinazione di tre malattie: cutis verticis gyrata, grave deficienza mentale ed aplasia della tiroide. Anche quattro dei suoi parenti stretti presentavano grave ritardo mentale. Viene discussa la possibilità che la sindrome venga trasmessa mediante un gene recessivo legato al cromosoma X.

RÉSUMÉ

L'Auteur décrit le cas d'un homme de 32 ans atteint d'une combinaison de trois maladies: cutis verticis gyrata, grave déficience mentale et aplasie de la thyroïde. Aussi quatre de ces parents étroits présentaient un grave retard. La possibilité d'une transmission recessive X-linkée de ce syndrome est discutée.

ZUSAMMENFASSUNG

Es wird der Fall eines 32jähr. Mannes vorgetragen, der an einer Kombinierung von drei Krankheiten leidet: Cutis verticis gyrata, schwerer Schwachsinn und Schilddrüsenaplasie. Auch vier seiner engen Verwandten sind geistig stark zurückgeblieben. Es wird die Möglichkeit erörtert, daß das Syndrom durch ein rezessives, an das X - Chromosom gebundenes Gen übertragen wird.