Hereditary Congenital Deafness in Uniovular Twins

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A deaf-mute boy of 14 years, who was examined owing to his poor progress in the special school for the deaf-mute, proved to be one of a twin. Both boys were born 6 weeks before the normal term by breach presentation at the end of an uncomplicated pregnancy. There was one placenta. The mother had noticed A's deafness when he was 11 weeks old, as he did not react to street noises as his brother did. The twin brother B is deaf in the right ear; he attends the secondary school and his performance is average.

Heredity

The father's hearing is normal; his audiogram presents only a slight dip ($\pm$20 db.) at 4000 cps; this is probably of traumatic origin. In his family no hard-of-hearing members are known.

The mother has slightly diminished hearing of a mixed type in both ears. There is a gradually increasing loss from 1000-5000 cps. On the left side a slight conductive deafnes is also discernable. The mother's sister and brother attended the school for the deafmutes. The mother's sister has upon audiological examination a severe perception-deafness on both sides; the « recruitment » is strong. She does not hear speech; lipreading is reasonably good. Her husband has a normal hearing. The son of this sister has a serious perceptions-deafness in the left ear, whereas there exists only a slight disorder in the right ear; his ability to understand speech is good. The daughter of the sister was too young for audiometric examination; she seems to hear well.

The audiogram of the mother's brother could not be taken. The hearing acuity of both ears is bad; his wife has normal hearing, their two children as well.

The parents of the mother, that is the grandparents of A en B, had normal hearing acuity but a sister and a brother of the mother's father were deaf-mutes; they did not marry. They died at a young age, as did the hearing brothers and sisters of the father, without offspring. The mother's grandfather was deaf in his old age. He had besides one hearing brother and two hearing sisters two deaf brothers who died rather young. The hearing sisters had no children, the brother's children and grandchildren have normal hearing.

In the family of the mother's mother no hearing-disorders are known.

The family tree is given in Fig. 1.

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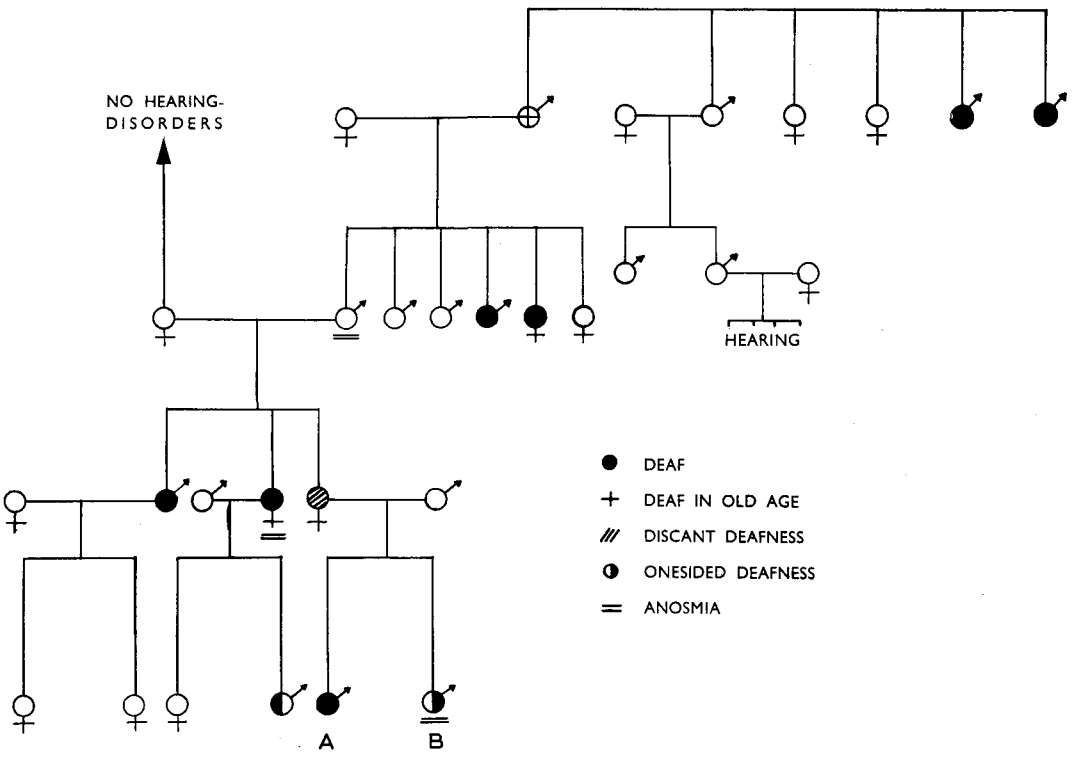


Fig. 1

ANTHROPOLOGICAL EXAMINATION (with the collaboration of J. van den Bosch, M.D.)

The boys were so much alike that even the mother had difficulties in distinguishing between them in the first half year and it is still hard for anybody who does not happen to know them well.

Tab. 1 - Anthropological examination, October 1955

	A (deaf in both ears)	B (deaf in right ear)
Length	1.565 m.	1.565 m.
Weight	47 kg	47 kg
Greatest circumference of the skull	53.5 cm	53.5 cm
Length of the head	17.6 cm	17.2 cm
Breadth of the head	14.2 cm	14.5 cm
Cephalic index	80.1	84.7
Distance of inner eye corners	3 cm	3 cm
Distance of outer eye corners	10 cm	10 cm
Length of ear	5.3 cm	5.2 cm
Length of hand	17.1 cm	16.8 cm

The colour and the implantation of the hair and the vertex are identical.

The iris of both is white-blue, lacunar and identical.

The visual acuity of both is L=R=1.

The colour discrimination of both is unimpaired.

Both boys show a slight epicanthus.

The shape and alignment of the teeth are identical. B. who eats more sweets has slightly more caries. The dental arches are similar, so that B. could use A.'s brace. Both show a marked overbite.

The tonsils are identical as well as their position between the fauces. The uvula of both boys shows a slight deviation to the left. The form of the mouth and of the lips is identical in both boys, the left angle of the mouth being somewhat lower than the right one.

The form of the ears and the profile of the nose are identic.

Both A. and B. have an extra lobule of the tragus.

Both are bathrocephalic with a slight impression under the lambda-point. The small front has the same configuration in both twins.

Both A. and B. react to wounds with depigmentation; leukodermic patches therefore are dissimilar, A. who has a more uninhibited primitive motorium manifesting a greater number of these.

Pigmented nevi are very dissimilar. Dactyloscopic examination shows a similarity such as known to occur between brothers and sisters but no identity; this is the general rule for uniovular twins.

The feet are identical with the same thumb-like form of the halluces. They have the beginning of hammer toes in the second, third and fourth toes.

The skin of the palms of the hand is dry, a symptom of heredity from the mother's family; the mother's father as well shows this symptom.

B. shows anosmia; this symptom as well is hereditary in the mother's family.

Both manifested a beginning of pubes in October 1955.

B. had pregnyl injections and underwent rightsided orchidopexia for retentio testis; his prepuce is shorter than that of A.

B. has a better muscular development than A, but he used to do more sports and gymnastics.

Internal and neurological examination showed no disorders; neither did ophthalmologic examination (Dr. R. P. Mesker); in particular no retinitis pigmentosa was present.

The electro-encephalogram of both boys is normal.

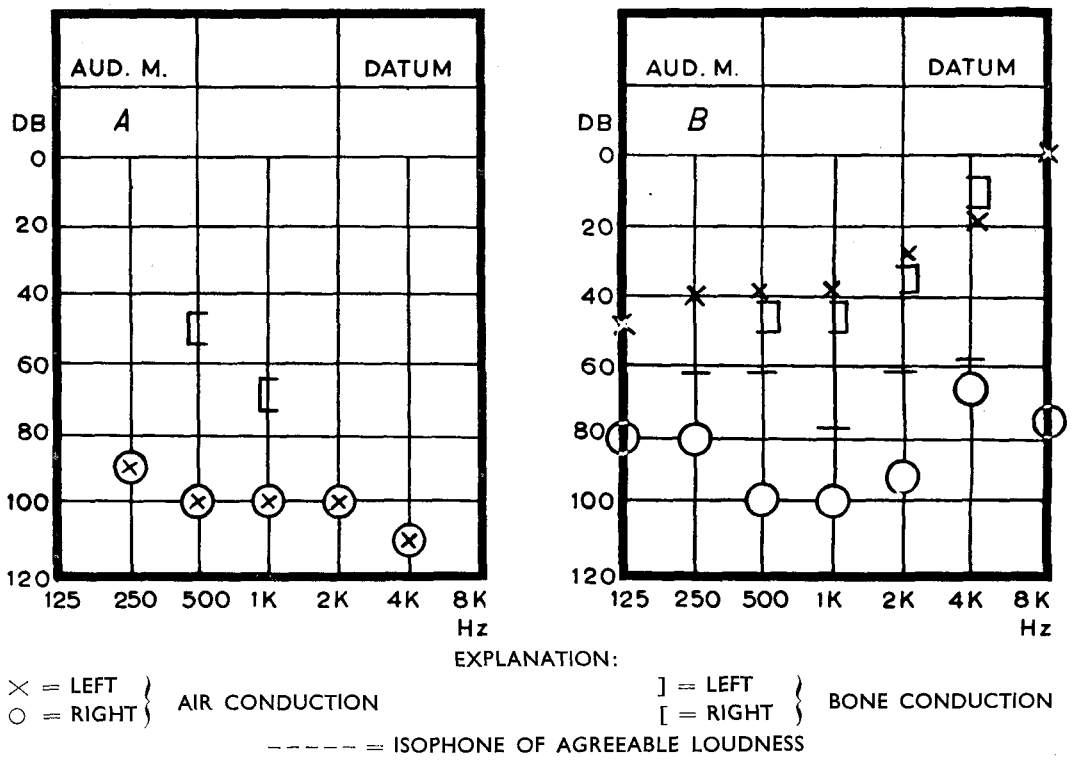
On X-ray examination the planigraphic (tomographic) examination of the cochlea of both boys included, no abnormalities were detected (Dr. B. G. Ziedses des Plantes).

Audiological Findings (Dr. J. van Ebbenhorst Tengbergen, otologist, and Dr. J.A.J. Klijn, physicist-audiologist).

The extra knob on the tragus excepted, no abnormalities of the outer ear were detected. The external meati of B are normal, A.'s right meatus being more tortuous.

The tympanic membranes of both boys are somewhat dull but of normal form. The passage of the Eustachian tube is normal with both.

The audiograms (Fig. 2 and 3) show the hearing loss of both boys. A. has a double-sided perception-deafness, probably there is no hearing at all; probably vibration simulates some hearing in the bone audiogram. B.'s audiogram shows a moderate

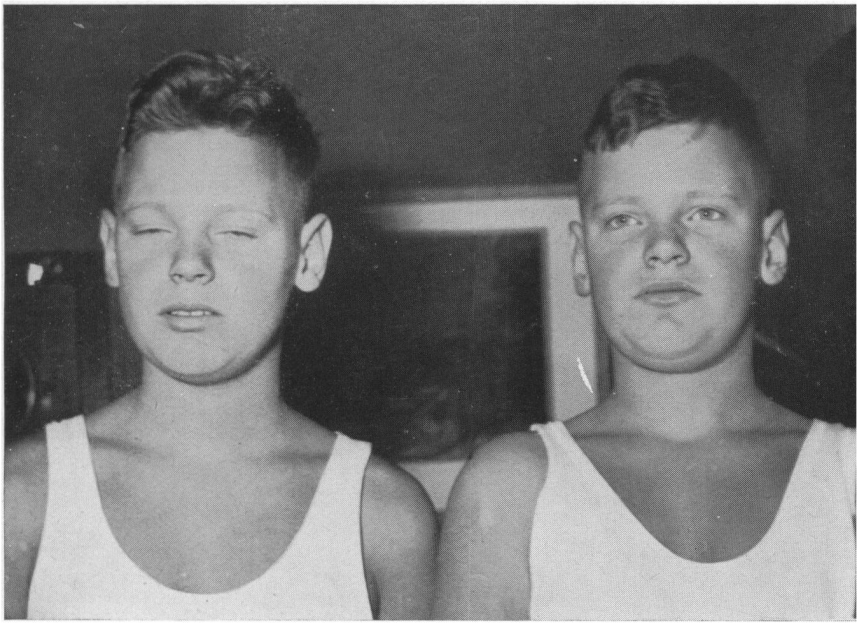


Figg. 2, 3

perception-deafness at the left side. It could not be determined whether B. had residual hearing of the right ear indeed, or whether this was a « shadow-audiogram ». Owing to recruitment hearing of speech with the left ear is high and acoustic reinforcements give no practical amelioration.

According to the mother A. may react to noises like the barking of a dog in the neighbourhood, clapping of the hands and sometimes clicking of the knife on the bread board. B. hears many noises but has difficulties in following conversation, wireless and telephone.

A Fig. 2 B



A Fig. 3 B

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Development in the first years

As babies, both A. and B. lalled, but B.'s lalling was more differentiated than that of A who in the course of the months grew mute. A. was late in sitting, standing and walking, whereas in B. this was all normal. A. only started to speak in the last months (14 years old), B. started at 1½ or 2 years.

Both twins were clean, and could dress and undress at a normal age.

Schooling. A. visited the school for the deaf since the age of 4 years, but has made insufficient progress. The reading technique is moderate, but his understanding of what he reads is poor. Arithmetic develops slowly but the level is low. Arithmetical thinking-problems are too difficult for him. The mastery of language, in spoken as well as in written form, and sentence formation is poor. If excited by an experience he writes mostly isolated words, illustrating the missing links with drawings. He likes geography, zoology and drawing most; he is able to make good pictures of many animals.

B. finished the Montessori-School and attends a secondary school. He is an average pupil, not only owing to his hearing defect but to carelessness and an uncritical attitude as well; in written work he may write anything, and as a result his notes for geometry and algebra are low. In oral lessons he is better. It seems that listening during the lessons causes a fatigue owing to which he is unable to concentrate during written work. He is able to hear the masters, sitting in the front row of his classroom.

Psychological examination

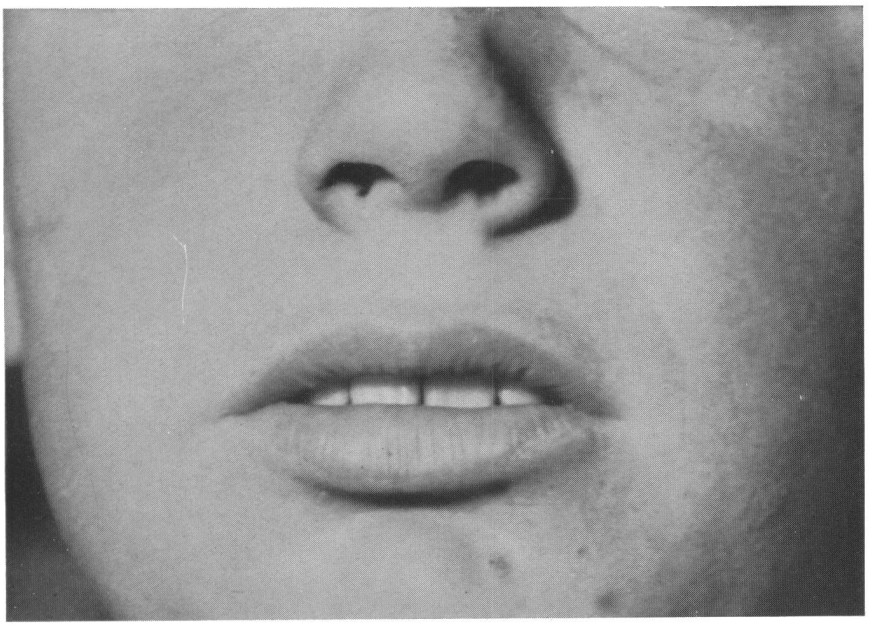
Both boys are sociable. Their behaviour is similar in many respects, but A.'s gait is more sluggish, a phenomenon that according to our experience is rather frequent in severely deaf children. B. has a better contact with other people, A. keeps more to himself, and has a different attitude towards the outer world, and is as it were more introverted.

A.'s motoricity is more restless; he makes many superfluous movements, such as tapping his cheek or ear, throwing his arms upwards, after which he remains seated in that position, with his face turned aside, making a sound like « prrr » or « trrr » especially if he is pleased. During the night he rolls in his bed with a hand under his back; B. used to do this as an infant.

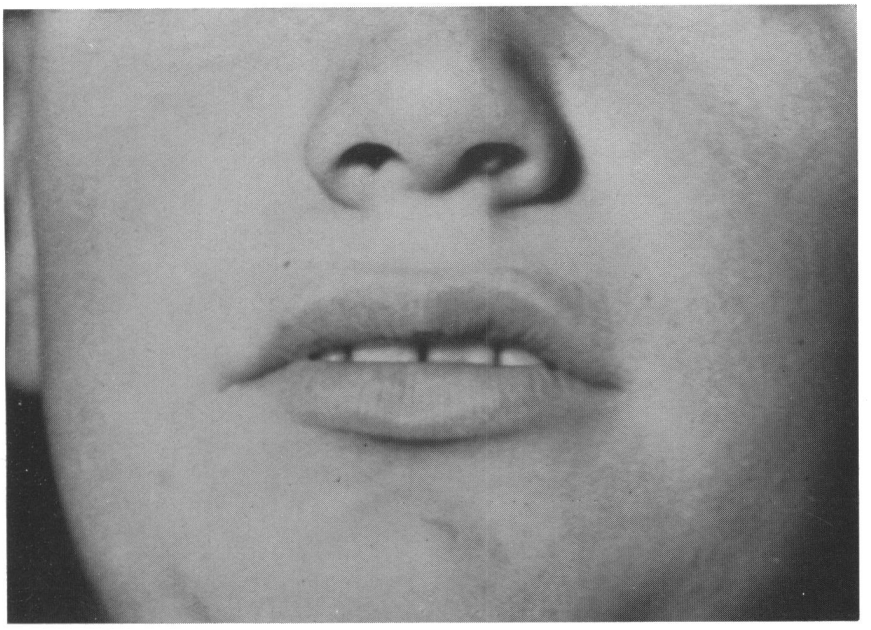
His restlessness disturbs A.'s attention, and influences his school work unfavourably. He is incapable of a long concentration. Especially when he is absorbed with a problem and is unable to find a way of abreaction, he fails in lipreading. If he is quiet on the other hand, he may take up much of the material of a lesson.

B. is much more tranquil than his brother, though he is not a quiet boy.

A. with his poor control of his emotions is loved by the children of his form who defend him; however he seldom plays with the children. He may be quick tempered but outbursts seldom occur. B. is a more mature personality and better adapted to the outer world, less infantile than his brother, but clearly handicapped as



A



B

Fig. 4. Form of the mouth

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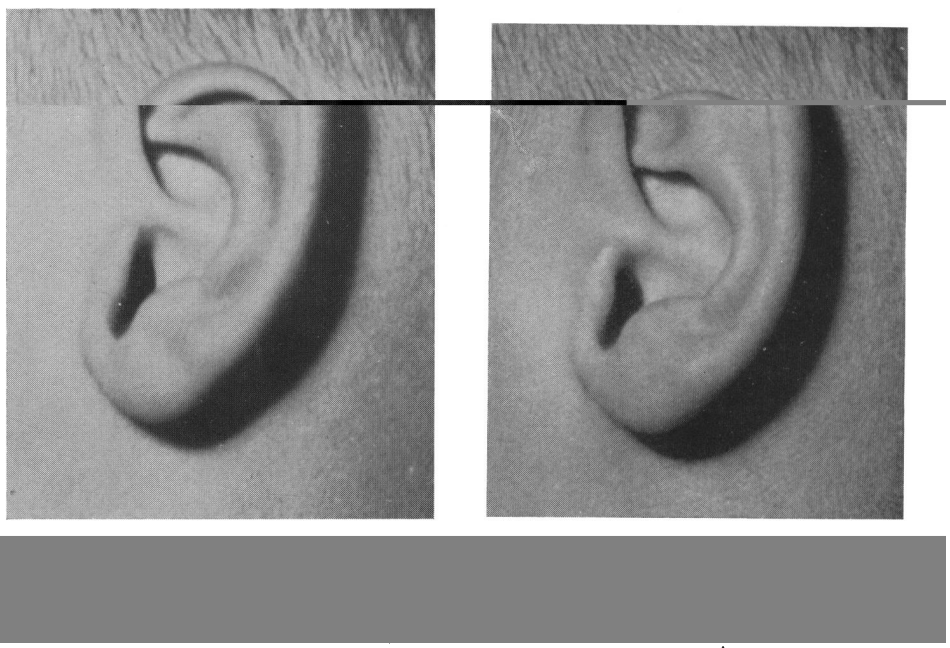
A Fig. 5 B



B Fig. 6 A

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compared with normal children; he is anxious and uncertain, agressive tensions are not fully discharged, and his urge for contact is not sufficiently satisfied.

A. has the characteristic monotonous speech of the deaf-mute, his speech-motoricity, however, showing the same lack of control as the total motoricity. His speech is bad, impulsive and clumsy, though he is able to articulate most sounds well. He does not like spontaneous speech and may be easy-going. He expresses his experiences and emotions mostly in gestures, often adding a single word like « bah », « dirty », « fine weather » etc.

B. is able to follow language well and in contrast to his brother he is a good listener, for example when attending to the instructions of the mental tests. His speech and use of language are normal.

In a story, e.g. in the Children's Apperception Test B.'s adequate and well elaborated story shows a good style and he uses abstractions; A. uses a string of disjointed words, sometimes resembling short sentences, full of repetitions, without a general plan.

Corporeal adolescence has started with A but mentally he is still infantile; B.'s adolescence follows a normal course. A. is preoccupied with death problems, B. manifests sexual problems.

A. is conservative, clinging to ritual; he imitates B. even in the choice of sweets, clothes etc. The boys are not jealous of each other. B. is unconsciously hindered by the fact that his brother is different. B. experiences their disturbed identity probably as an asymmetry, A. their likeness as a symmetry.

Intelligence

The intelligence was measured with Snijder-Oomen's revised intelligence scale for the deaf. Table 2 gives the raw scores and their value according to the standard scores of the sub-tests, the principle of the standard scores being a medium of 25 with a standard deviation of 5.

Tab. 2

	Raw scores		Standard scores		Differences in standard scores between A & B
	A	B	A	B	
Mosaic	33	45	25	31	6
Retention for pictures	11	18	19	32	13
Combination	17	25	23	33	10
Drawing	10	15	23	31	8
Knox-blocks	9	9	20	20	0
Similarities	14	16	26	30	4
Completion	17	19	24	28	4
Sorting out	13	15	26	31	5
Total			186	236	50
I.Q.			91	121	

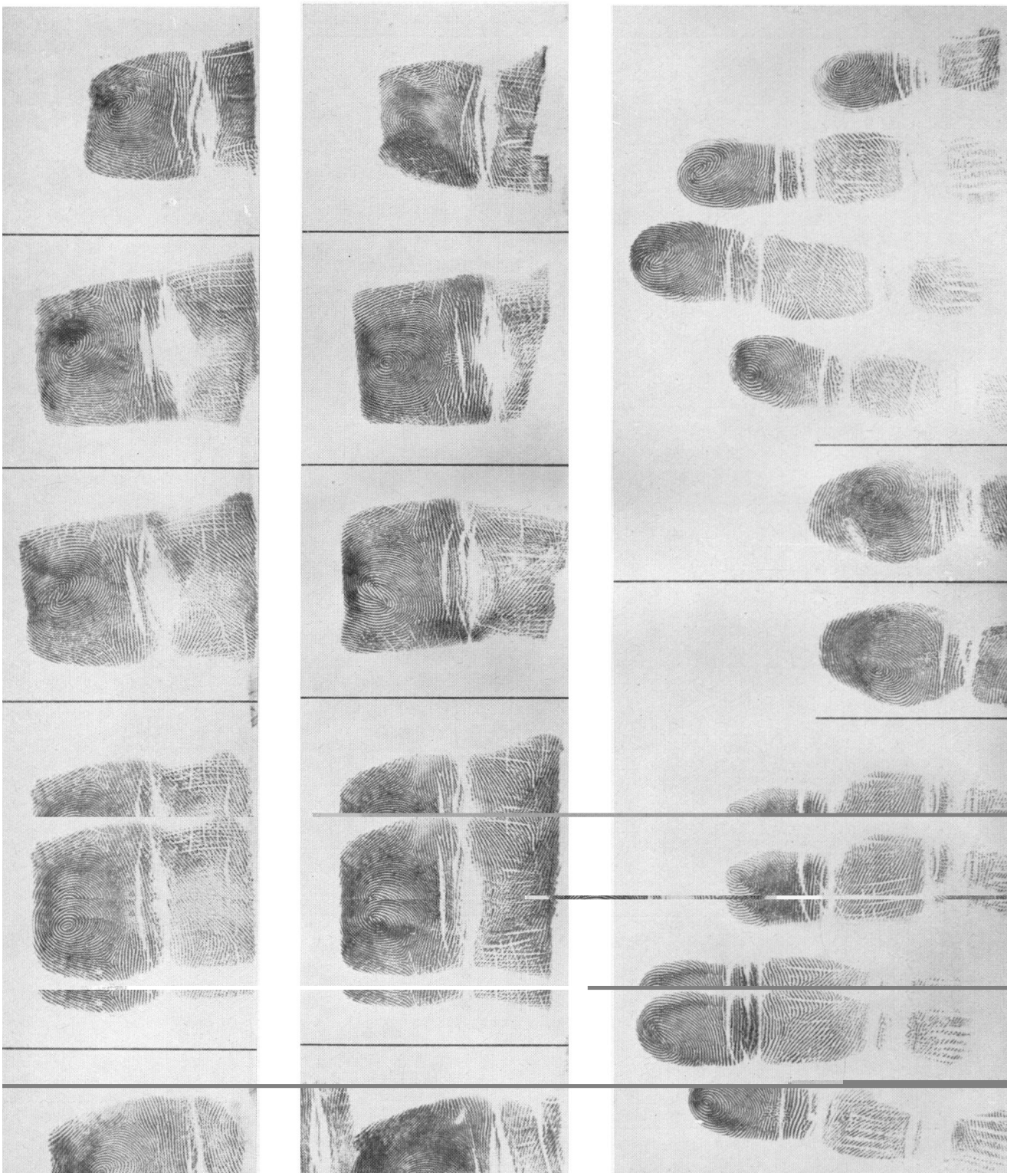

Fig. 7. A's dactylogram

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The Knox-blocks excepted A and B show a manifest divergence; the Knox-blocks is the subtest with the lowest correlation with the total score.

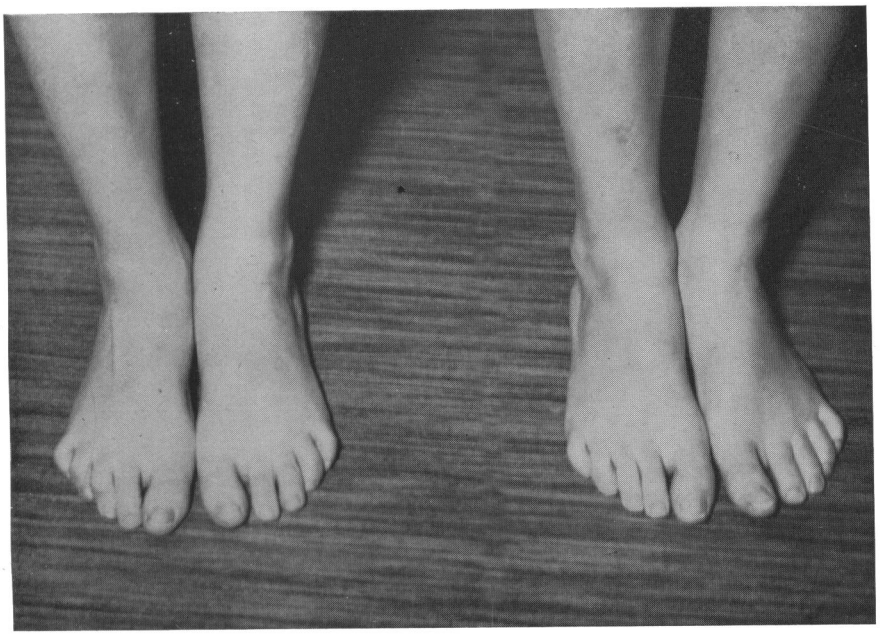
It is unnecessary to comment upon these results in this article. The reliability of these I.Q.'s is corroborated by the outcome of different intelligence tests performed under different conditions.

The revised S.O.-test gives the norms for the deaf and the hearing; these two norms showing a small difference for the higher intelligence levels. In B.'s case calculation according to both norms gives a difference in I.Q. of only 1 point; therefore it is possible to compare A. en B. following the norms for the deaf mute.

Test-examination

The extensive research carried out by means of tests will be mentioned superficially.

	A	B
Pintner-Cunningham	± 45 pt., i.e. ± 8¼ year	
Raven	III - (11, 10, 8, 7, 2)	III + (11, 11, 9, 9, 4)
Childr. Apperc. Test.	Primitive, concrete; no line; iterations, restless	Good abstractions, good story, good elaboration, language and style.
Wartegg	Primitive, experience-prompted; impulsive, explosive	Good level, playful fantasy, problems of the twinship.
Alexander Performance	15,7 year (the norms of this test are too low)	
Figure Reasoning Test (Daniels)		Score 102
Szondi	Severe tensions are discharged	Tensions are not discharged
Bourdon-Wiersma stipple Test (double task)	Worse than B.	Slightly unstable curve; too many omissions a. faults. Concentration is insufficient
Drawing		B.'s copying from memory is notably better than A.'s
Drawing a tree	Scheme of small children, with a small top	Good scheme for his age with manifest adolescence problems.



B Fig. 9 A

Summary of the findings

1. These monoplacentar, uniovular twins show a manifest identity.
2. A. is deaf in both ears, B. in the right ear with a slight hearing loss in the left ear.
3. The deafness in this family is hereditary.
4. The anthropological identity is very high, confirming the fact of the uniovular origin.
5. Besides psychological similarities there are manifest differences.
   a) A's intelligence is much lower over all performances (all along the line) than that of B. This is not only valid when their intellectual performances are compared as such, but also when in A.'s case the norms for the deaf are used and in B.'s case those for the hearing are used.
   b) A. is more restless than B. and over-lively.
   c) A. is hyperemotive, more infantilistic and immature than B.
6. a) A. can smell, B. can not.
   b) B. had a retentio testis at the right side.

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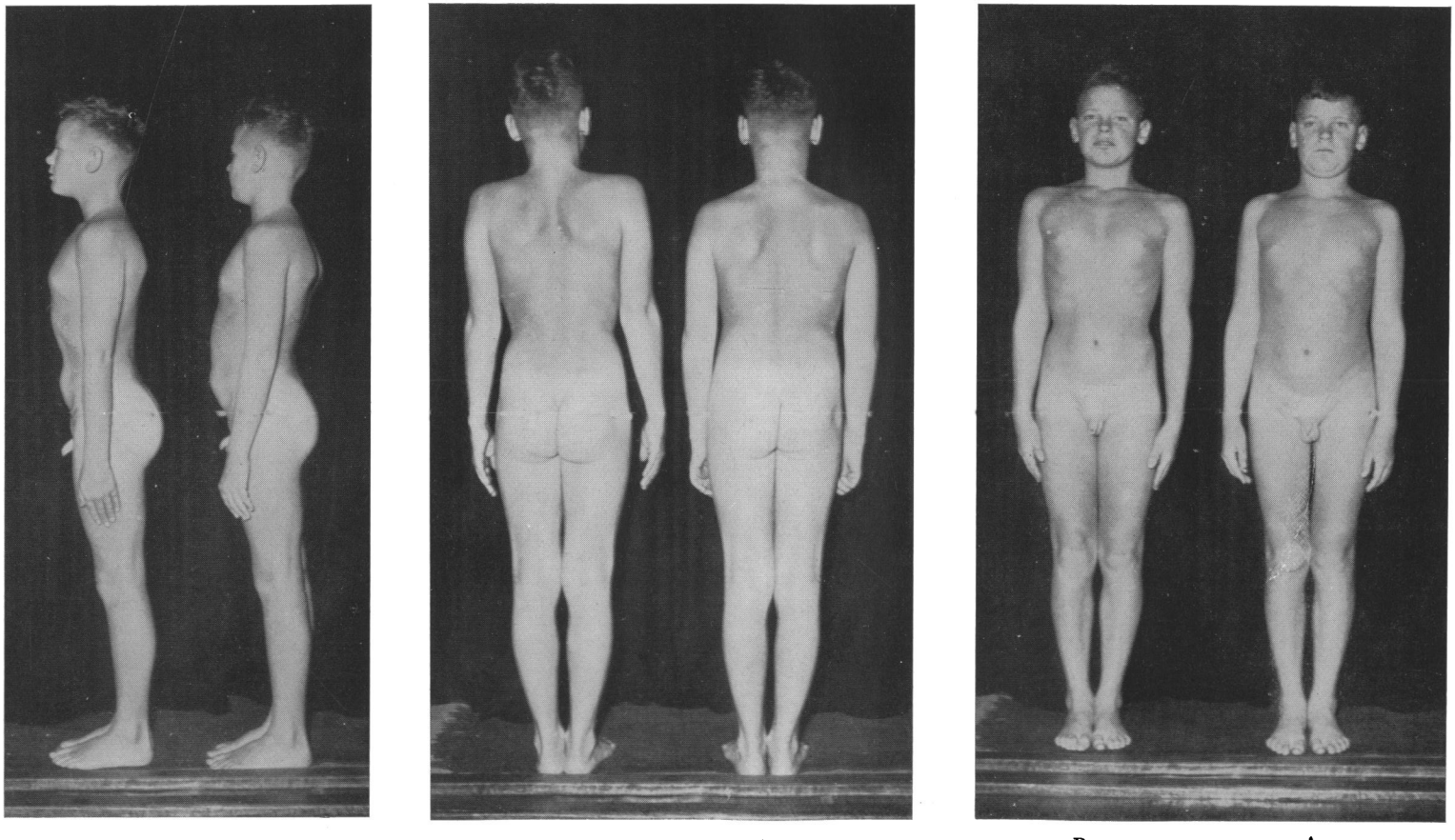


B A B A B A

Fig. 10

Considerations

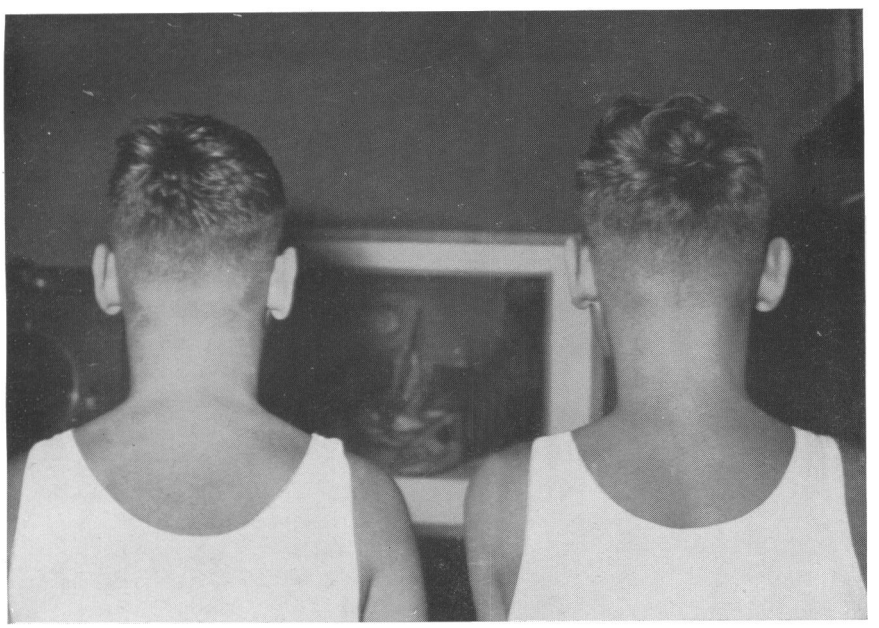
Accepting the fact that these boys are uniovular twins many problems arise.

I. How is the difference of the hearing disorder to be explained. It confronts us with a remarkable difference in detail in this case. One would expect both boys to have the same hearing disorder in both ears. What are the additional factors, superimposed upon the hereditary disposition causing the difference between the left ears of A. and B., and between the left and the right ear of B?

It is a well known fact that in many hereditary malformations like e.g. harelip, exogenic factors such as a vitamin deficiency in the mother, are active besides the genotypic factor. But why is the difference only noticeable in one of B.'s ears? At any rate it means that a hereditary (genotypic) disposition to deafness need not lead infallibly to deafness. In this connection it is noteworthy that a cousin of A. and B. is deaf in one ear (the left ear in this case), with a slight perception-loss in the right ear. These facts in the case of B. and the cousin refute entirely Langenbeck's hypothesis according to which in hereditary congenital deafness the hearing loss in both ears must be identical.

This conception had already been repudiated by De Kleyn, Van Gilse a.o.

II. A remarkable difference between the twins is B.'s anosmia, a phenomenon



A Fig. 11 B

hereditary in the family of the mother. We did not find a correlation with the deafness, but it was impossible to pursue our investigations further in this direction.

III. B.'s right-sided retentio testis could not be brought into relation with other phenomena.

IV. Neither could we find a relation between the deafness and the hyperkeratosis that is hereditary in the family of the mother. However, one cannot deny a relation a priori, because in families with hereditary deafness metabolic disorders are found to occur in the deaf as in the hearing members of the family, e.g. hypercholesterinemia and hyperpyruvemia. The absence of material for comparison and technical reasons prevented us to extend our research in this direction.

V. The psychological differences between A. and B., in intelligence as well as in other mental traits cannot be explained by A.'s severe deafness alone. Theoretically A. and B. should have the same intelligence. In the Revised Snijders-Oomen Test for the deaf the difference of the scores of the deaf and the hearing are only 4 I.Q.-points. The difference of 30 points between A. and B. is therefore not explained by the difference in hearing.

One must conclude that either A.'s deafness had a much stronger repercussion upon his mental and intellectual development than is the rule, or that besides deafness other organic differences between A and B exist, possibly as a result of the same external factor explaining the difference in deafness. We did not find any starting point for such a supposition, neither in the neurological, nor in the electro-encephalographic examination.

We may end with the conclusion that a detailed examination of uniovular twins with comparison of the differences to be found is of high scientific value.

Literature

ALTAMANN, F: (1950) Arch. Otolaryng. (Chicago), 51. 882.

GILSE, P. H. G., VAN HINNEN A. B. and NIEUWENHUYSE A. C. (1942). Bibliographia Genetica, 13. 301, 364, 412.

KLEYN, A. DE: (1941) Proc. Kon. Nederl. Akad. Wetensch., 44.90.

LANGENBECK, B.: (1940) Monatschr. Ohrenheilk., 74.562.

SNIJDERS, J. TH. and SNIJDERS-OOMEN N.: Niet-verbaal intelligentieonderzoek van horenden en doofstommen. Nijmegen 1958.

RIASSUNTO

Descrizione di due gemelli uniovulari; essi presentano una grande eguaglianza antropologica. Ciononostante uno di essi (soggetto A) è sordo al livello delle due orecchie, mentre il soggetto B è sordo soltanto dall'orecchio destro e soffre di una leggera perdita di udito all'orecchio sinistro. La sordità è ereditaria nella famiglia. Dal punto di vista psicologico i due ragazzi dimostrano una certa concordanza, ma presentano anche delle importanti differenze. L'intelligenza di A è in tutti i campi molto inferiore a quella di suo fratello. Se vengono applicati i nuovi test di Snijders-Oomen per i sordi, A raggiunge il livello normale dei sordomuti, mentre B raggiunge il livello normale delle persone che hanno l'udito normale; inoltre A è inquieto, cangiante e presenta ipermotilità, instabilità e infantilismo.

Nella famiglia esiste l'anosmia; A percepisce bene dal punto di vista dell'olfatto mentre B non percepisce. B presenta criptorchidia a destra la quale è assente presso A. Si pone il problema del come si possa spiegare la sordità unilaterale presso B, bilaterale presso A. Per influenza di fattori esogeni la disposizione si manifesta nel caso di B. da un lato solo come è il caso presso un cugino. L'idea sostenuta da Langenbeck che una sordità congenita ereditaria si manifesti in modo identico nelle due orecchie viene contraddetta da questa osservazione come del resto da altre (Kleyn, van Gilse ecc.). Dal punto di vista teorico, l'intelligenza di A e quella di B dovrebbero essere quasi uguali; il test di Snijders-Oomen permette di introdurre una correzione tenendo conto della sordità di A. La grande differenza nella intelligenza dei due gemelli non si spiega peraltro nè mediante l'esame neurologico, nè mediante l'elettroencefalogramma.

RÉSUMÉ

Description de deux jumeaux uniovulaires; ils sont d'une grande conformité anthropologique. Pourtant l'un d'eux, A, est sourd des deux oreilles, tandis que B est sourd de l'oreille droite et souffre d'une légère perte de l'ouie de l'oreille gauche. La surdité est héréditaire dans la famille. Du point de vue psychologique les deux garçons montrent une certaine conformité mais présentent d'importantes différences. L'intelligence de A est dans tous les domaines très intérieure à celle de son frère. même si — en appliquant les nouveaux tests de Snijders-Oomen pour les sourds — on admet pour A la norme des sourds-muets, pour B la norme des personnes ayant l'ouie normale, l'échelonnage étant faite pour ces deux groupes.

En outre A est inquiet, remuant, présente une hypermotilité, instabilité et infantilisme.

Dans la famille on rencontre l'anosmie; A sent bien, B ne sent pas. B présentait une retention du testicule droit que A ne présentait pas.

La question se pose comment expliquer la surdité unilatérale chez B, bilatérale chez A. Sous l'influence de facteurs exogènes la disposition ne se manifeste dans le cas de B d'un côté seulement, tout comme c'est le cas chez un de leurs cousins. L'idée soutenue par Langenbeck qu'une surdité congénitale héréditaire se manifeste identiquement des deux oreilles est contredite par cette observation, comme du reste d'autres (de Kleyn, van Gilse e.a.) l'avaient déjà contestée. Du point de vue théorique l'intelligence de A et de B devraient être à peu près égaux; le test de Snijders-Oomen permet d'introduire une correction en tenant compte de la surdité de A. La grande différence dans l'intelligence des deux frères ne s'explique ni par l'examen neurologique ni par l'électrencéphalogramme.

ZUSAMMENFASSUNG

Ein eineiiger Zwilling mit hochgradiger anthropologischer übereinstimmung wovon aber A taub ist an beiden Ohren, B am rechten Ohr mit gereingem Hörverlust am linken Ohr. Die Taubheit in der Familie ist erblich. Psychologisch sind die beiden Jungen seht verschieden, trotz bestimmter Übereinstimmung. A 's Intelligenz ist auf der ganzen Linie bedeutend niedriger als die Intelligenz des Bruders; dies gilt auch noch wenn man an Hand der neuen Snijders-Oomen-tests für Taube und Schwerhörige für A de Taubstümmennorm, für B den Norm für Hörende gelten lässt. Ausserdem ist A unruhig, zappelig und über-beweglich, hyperemotionell, infantilistisch und unreifer als B.

In der Familie kommt auch Anosmie vor; A kann riechen, B nicht; ausserdem hatte B eine rechtsseitige Retentio testis die bei A nicht bestand. Es erhebt sich die Frage wie die einseitige Taubheit bei B und die doppelseitige bei A zu erklären zei. Bei gleicher genetischer Anlage ist unter Einfluss exogener Faktoren die Taubheit bei B nicht beidseitig, wie das auch bei einem Vetter von A und B der Fall ist. Es wird hiermit ausserdem Engenbechk's Annahme dass eine erblich-kongenitale Taubheit sich an beiden Ohrenidentisch manifestiert, widerlegt, wie dies übrigens vorher schon von de Kleyn, van Gilse u.a., angefochten sourde.

Theoretisch sollte A 's Intelligenz die gleiche sein wie die B's, wenn man die Taubheit in Ammerkung nommt. Wir fanden keine Erklärung für die erhobene Differenz, weder in der neurologischen Untersuching, noch elektro-enzepahlografisch.

Es wird gefölgert dass die genaue Untersuchung eineiiger Zwillinge unter Betrachtung der Verschiedenheiten von grosser wissenschaftlicher Bedeutung ist.