cases the serum vitamin A was subnormal while that of carotene was within normal limits. The role of vitamin A in tylosis and various other skin conditions and its possible relationship to squamous metaplasia and carcinoma are discussed. The metabolism of vitamin A and its functions in maintaining the integrity of epithelial structures is still poorly understood. Some of the possible genetic mechanisms underlying this association between low serum vitamin A and familial skin diseases are outlined.

In addition to the propositus, two other living members had sustained a myocardial infarction. Hypercholesterolaemia and hyperlipaemia were noted in several members, but were unrelated to tylosis, clinodactyly, or serum vitamin A.

Tylosis is transmitted via a monomeric autosomal dominant gene with a high degree of penetrance. The expressivity of the condition is governed to a large extent by environmental modifiers. Clinodactyly is inherited as a non-sex-linked Mendelian dominant with low penetrance. The explanation for the coexistence of two genetically determined entities is considered, and in the case of tylosis and clinodactyly we feel that the association is due either to distant linkage or to the genes being on different chromosome pairs.

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HEREDITARY BRANCHIAL DEFECTS IN A HAMPSHIRE FAMILY

BY

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This paper records the occurrence of multiple congenital branchial defects in several members of a family covering four generations. A number of similar inherited anomalies have been described previously, but in this particular group multiple lateral cervical sinuses were associated with maxillary malformations and ear-pits, a combination which does not appear to have been reported before.

The grandfather of the propositus was a Wiltshire man. His daughter married a Hampshire farm labourer, moved to a small village near Winchester, and with her increasing progeny has remained in this area ever since. The family group comprised 21 members; 15 were blood relatives, and eight of these possessed abnormalities. The affected persons were the grandfather himself, two of his daughters and one of his sons, and three grandsons and one granddaughter. I examined four normal and three affected relatives. The remaining 14 members, including five affected persons, were either dead or untraceable at the time of inquiry. The descriptions of these are thus unfortunately only second-hand, being provided from the memories of the relatives personally examined; but, since these tallied though supplied without prior collusion, they are probably reliable. These people were uncertain about the presence of ear-pits in the other affected relations, but this is not surprising, as these small malformations are quite unobtrusive.

The ear-pits when present were situated in the upper anterior end of the helix, being about 1 mm. wide and 1 mm. deep. They were observed only in the propositus and his brother. There were two maxillary abnormalities, always coexistent. The upper front teeth projected forward at an angle of some 20 to 30 degrees from the vertical, giving a slightly simian cast to the facies. The hard palate was high and arched, equally on both sides. The lateral cervical sinuses were typical of their kind. Their openings in the skin were 2 to 3 mm. in diameter and were always located in the anterior line of either sternomastoid muscle at various levels. Their depth was not probed. They usually caused little trouble, but when a collar was worn were inconvenient

because of their tendency to exude small quantities of clear fluid. The brother of the propositus was the only person in the group to be ever more than merely irritated by his defects. His lower left sinus had become infected several times, with on one occasion a cellulitis which spread down over the chest and brought him near to death. The sinuses were bilateral in every case, though often at different levels on each side, and in some they were multiple.

The three affected members personally examined were the propositus, his brother (III 5), and his mother (II 2). The propositus had the maxillary malformations, a left ear-pit, and a pair of lateral cervical sinuses. The right-sided sinus lay at the level of the inferior margin of the thyroid cartilage, while that on the left was situated a little higher, nearly lateral to the junction of the superior horn with the main thyroid cartilage. The brother, III 5, had a similar ear-pit and maxillary anomalies. He had besides, however, no fewer than four of these sinuses. Two were at the same level—one about 1 in. (2.5 cm.) above the anterior clavicular insertion of each sternomastoid muscle; another was just below and behind the left mandibular angle; and a fourth was a little inferior and anterior to the lobe of the right ear. The mother, II 2, had no ear-pit, but the maxillary defects were well developed. Her two sinuses were situated at the same level, one immediately in front of the clavicular insertion of each sternomastoid. The father, paternal uncle, wife, and daughter of the propositus were personally examined and found to be normal in all respects.

The five affected members not examined were I 1 and II 1, who were both dead; and II 4, III 1 and 6, whose whereabouts were unknown. They all were said to have had at least two similar sinuses in their necks, high palates, and upper front teeth which "stuck out." Two maternal uncles, II 3 and 5, and three cousins, III 2, 7, and 8, were not examined, but were said to be unaffected. Three spouses not examined—the two husbands of II 1 and the wife of II 4—were also said to be normal, and, so far as these people knew, were at least not closely related. Neither the father nor the paternal uncle of the propositus knew of any relationship between themselves and these other spouses, or between the spouses individually "as far back as they could remember."

Family tree. ■ = Affected male. ● = Affected female. PPU = Paternal uncle of propositus.

Discussion

Neither congenital lateral cervical sinuses nor ear-pits are uncommon isolated anomalies. Ewing (1946) observed ear-pits in 0.9% of 3,500 British Service men, and Stannus (1914) found them in 4.5% of 6,491 Bantus in Nyasaland. But they are unobtrusive, usually trouble-free, and thus may pass unnoticed for years by their owner even if he is medically qualified (Fourman and Fourman, 1955).

Aird (1958) states that careful search will reveal a family history of fistula or accessory auricle in some 25% of cases of lateral cervical ("branchial") fistula.

Previous reports of the occurrence of such malformations in several members of a family include those by Paget (1877), Kindred (1921), Přecechtěl (1927), Whitney (1939), Edmonds and Keeler (1940), Quelpud (1940), Kinder (1954), Fourman and Fourman (1955), and Wilson (1955). None of these mentioned an associated high arched palate as appears in this family. The only instance of dominant inheritance of such a defect given by Gates (1946) is that integral in the well-known condition of arachnodactyly, but none of the members of the above family showed any other stigmata of that syndrome.

When ear-pits occur alone or with lateral cervical sinuses as the only other associated anomaly, the apparently dominant inheritance shows incomplete or irregular penetration (Kindred, 1921; Edmonds and Keeler, 1940; Royk-Lewinski, cited by Quelpud, 1940), sometimes "skipping" of generations (Whitney, 1939), or may appear in only one of uniovular twins (Quelpud, 1940). However, when a further concurrent abnormality is present, such as deafness (Přecechtěl, 1927; Fourman and Fourman, 1955), or a high palate as in this family, the dominance seems to be enhanced, so that the penetrance of the combination as an entity becomes more complete. This family report further confirms the evidence of previous accounts in demonstrating that either sex may manifest and transmit hereditary branchial developmental abnormalities.

For what such small numbers are worth in deducing gene characteristics, the proportion of 8 to 15 presenting here suggests a simple dominant mode of inheritance, and the pattern of appearance of the defects throughout the pedigree strongly supports this form of transmission. Interest focuses naturally upon the consanguineous marriage between the propositus and his cousin. Although this woman came from an affected mother, II 1, neither she, III 3, nor her daughter, IV 1, by the propositus showed any of these defects. This normalcy of the wife and daughter of the propositus admittedly could mean that the woman II 1 was homozygous recessive and, with her second husband either normal or a heterozygous recessive carrier, that III 3 and her daughter were therefore heterozygous carriers. But were this gene or character complex recessive, the pattern of transmission from the other matings could be explained only by heterozygous possession of an identical recessive factor by no fewer than four different unrelated spouses—the wife of I 1, the husband of II 1, the husband of II 2, and the wife of II 4. This is unlikely, whereas simple dominance explains every manifestation of this defect pattern.

It would appear, then, that the linkage between the character for lateral cervical sinuses and that for ear-pits is ordinarily weak; so that the pair may separate during nuclear division, cross over, and "recess" among normal genes. In this reported family it seems that the advent of yet another abnormal character, for maxillary defect, has consolidated the linkage between all three to produce a complex which, when transmitted, behaves as a full dominant.

Summary

A family of fifteen persons is described wherein eight members in three generations exhibited closely linked congenital anomalies comprising multiple branchial malformations and maxillary defects. The complex is

revealed to have been inherited as a simple dominant, uninfluenced by sex in either transmission or penetration. The literature is reviewed and the heredity discussed.

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Medical Memoranda

A Case of Artificial Abdominal Urethra

Early in 1959 I was fortunate enough to see a demonstration by Lapides at Michigan University Hospital, Ann Arbor, of a boy who, as a result of a fracture of the pelvis, developed a urethral stricture, and in whom the urethral canal could not be reopened. The child was passing his urine through an abdominal urethra, was perfectly continent, and had complete control over urination.

Back in Mauritius, I was confronted with a similar case.

CASE REPORT

An Indian schoolboy, aged 11 years, was admitted into hospital on July 16, 1958, suffering from a fracture of both rami of the right os pubis and passing blood per urethram, as the result of a street accident. The surgeon who attended him found a complete rupture of the urethra, with extravasation of blood into the cave of Retzius and paravesical spaces. Subsequently several attempts at external urethrotomy failed for various reasons, and when I saw the boy in April, 1959, he had a de Pezzer catheter draining the bladder through a suprapubic cystostomy wound.

Bougienage and urethroplasty having failed, an attempt was made on April 8, 1960, to fashion an artificial abdominal urethra, as described by Lapides.

Under a general anaesthetic, a median incision extending from the umbilicus to the symphysis pubis was made and the peritoneal cavity opened. The bladder was detached from the abdominal wall and the cystostomy wound closed with No. 2 chromic catgut. An area of the anterior bladder wall, measuring four centimetres square, was mapped out. Three sides of the square were cut out to form a flap of bladder wall with the base attached to the dome of the viscus. The flap was converted into a tube, the sides being stitched in two layers with No. 2 chromic catgut, the first layer to approximate the sides and the second layer to invaginate. The defect in the bladder was similarly closed by two layers of chromic No. 2 catgut. The open end of the tube was passed through the linea alba and skin, away

from the abdominal recti. The mucosa of the mouth of the bladder tube was stitched to the skin. The abdominal wound was closed in layers and the bladder was drained by a rubber catheter passed through the artificial urethra thus formed. The patient was placed on antibiotics.

After 14 days the catheter was removed. It was then found that the boy could hold his water for half an hour, and when he micturated there was a small stream and a lot of dribbling down his abdominal wall. Within a week he was able to hold his water for a period of two hours and could get a good jet, as shown by the accompanying photograph.

When he was discharged from the hospital on June 1 he was perfectly continent. When seen six months after the operation he was found to be completely continent, and got up only once a night to empty his bladder.

In Lapides's cases there was no residual urine, but in the present one there was a residual urine of about 5 ml. Cystography after micturition showed a small pool at the natural neck of the bladder. It is doubtful if this residue, situated right at the bottom of the bladder, can ever be got rid of.

My thanks are due to Dr. E. Losonczy, Director of Medical Services, Mauritius, for his permission to publish this report.

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The MINISTER OF LABOUR, Mr. John Hare, has used his powers under the Factories Act, 1959, to reappoint the Joint Standing Committee on Health, Safety and Welfare in the Drop Forging Industry. The function of the committee, which has been appointed for three years, is to advise the Minister on matters affecting the health, safety, and welfare of persons employed in the drop forging industry.