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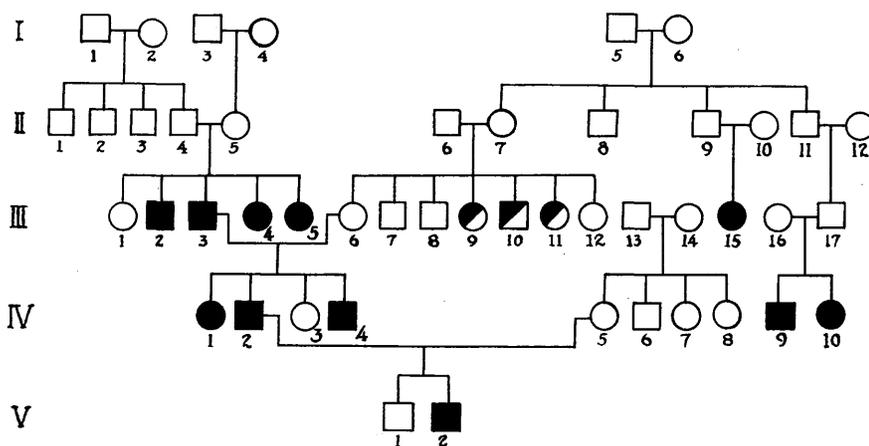
STUDIES IN HUMAN INHERITANCE

XXII. THE INHERITANCE OF CONGENITAL PYLORIC STENOSIS

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In a recent issue of this Journal (Vol. XXXIV, No. 1, Jan., 1943), Cockayne and Penrose discussed the genetics of congenital pyloric stenosis. They concluded that the condition is a recessive diathesis, that males are more susceptible than females, and that the first-born child is more susceptible than children born afterwards in the same sibship. In the recorded cases Cockayne and Penrose find a low familial incidence and relatively few pedigrees containing more than one affected child.



Pedigree of congenital pyloric stenosis:

III—2, Died of pyloric stenosis at 5 months. 3, Recovered from pyloric stenosis. 4, Died of pyloric stenosis at 5 months. 5, Died of pyloric stenosis at one day. 9, Died at two months, cause undetermined. 10, Died at three days, cause undetermined. 11, Born dead. 15, Died in infancy of pyloric stenosis.

IV—1, Died of pyloric stenosis at 8 days. 2, Recovered from pyloric stenosis; father of propositus. 4, Died of pyloric stenosis at 9 months. 9, Died of pyloric stenosis in infancy. 10, Recovered from pyloric stenosis.

V—2, Propositus; operated on for pyloric stenosis; recovered.

There has recently come to my attention a pedigree of congenital pyloric stenosis which provides interesting additional data (figure 1). The diagnosis of the propositus (V, 2) was confirmed at operation. The diagnoses of all other affected members were made on the basis of similarity of symptoms, which are very striking (projectile vomiting, constipation, gastric peristalsis, etc.).

Cockayne and Penrose recorded from their data a male to female ratio of six to one among affected cases. In the pedigree presented here there were six affected males and five affected females: no significant departure from equality. Moreover, Cockayne and Penrose state that on the basis of their data, a female who is not first-born stands very little chance of being affected. In the family under discussion here, however, only two of the five affected females were first-born, one was second-born, one was third-born and one was fourth-born.

Primogeniture has been suggested more than once as a contributing cause to congenital pyloric stenosis. The critical evidence necessary to establish this, namely, that children born after the patient are almost always normal, has never been produced. The pedigree herewith presented contains strong evidence against the causal nature of primogeniture. The birth orders of the affected children in figure 1 are as follows:

First-born: three, one of whom was an only child, one of whom was followed by one affected sib, and one of whom was followed by an affected sib, then by a normal sib, and finally by another affected sib.

Second-born: four, in two of which cases the first-born also was affected.

Third-born: one, in which case the first-born was normal, the second-born affected.

Fourth-born: two, in one of which cases the second-born and third-born also were affected, and in the other of which the first-born and third-born were similarly affected.

Fifth-born: one, in which case the second-, third- and fourth-born also were affected.

Cockayne and Penrose recorded an increased tendency to hare-lip and cleft palate in the families of patients. In the family presented here, there were two cases of cleft palate, namely II, 8 and a third cousin of IV, 4 not shown in the chart. No consanguineous marriages occurred in the pedigree shown in figure 1.

The data presented here are consistent with the hypothesis that congenital pyloric stenosis is due to an autosomal recessive gene substitution, but these data do not support the contention that primogeniture is a contributing cause, nor do they support the conclusion that males are more susceptible than females.

LITERATURE CITED

- Cockayne, E. A., and L. S. Penrose. 1942. The Genetics of Congenital Pyloric Stenosis. Ohio Journal of Science, 43: 1-16.
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