BRIEF YALE REPORTS

MOTHER AND DAUGHTER WITH BILATERAL CONGENITAL AMASTIA. By H. Goldenring and E. S. Crelin, Department of Anatomy, Yale University School of Medicine.

Only three publications were found in which complete bilateral congenital absence of the mammary glands was reported to have occurred in otherwise normal individuals. Wylie[3] described a 21-year-old affected mother of an unaffected son. Williams[2] published additional information, supplied by Wylie, concerning this woman. Fraser[1] described three generations of a family in which bilateral amastia showed a dominant inheritance. The male was the affected individual in the first generation. Three of his four daughters were affected and his two sons were unaffected. One of these affected females had three affected daughters and two unaffected sons. The father of these latter children was unaffected. A three generation study of his family, similar to that made of his affected wife's family, revealed no occurrence of amastia.

The individuals with amastia of the present study are a 25-year-old woman and her 3 ½-month-old daughter (Figs. 1 and 2). Physical examination of the mother revealed, in addition to the apparent amastia, sparse axillary and pubic hair, a high arched palate, and depressed nasal bridge suggesting saddle nose. The teeth are apparently within normal limits with reference to number and configuration. The finger nails, although traumatized by nail biting, show no dystrophic changes, nor do the toe nails. The bony thorax, pectoralis major muscles, and all other findings were within normal limits. Biopsy in the axillary area revealed some hairs, numerous active eccrine sweat glands but no apocrine glands whatever.

The infant's hair and nails appeared normal although absolute evaluation at age 3 ½ months is not possible. The child did show marked hypertelorism, high arched palate, and saddle nose as well as the absence of areolae and nipples. The areas where, normally, the breasts would be on the daughter have the same surface configurations of the breast areas on an unaffected child of a comparable sex, age, and body build. However, the breast areas on the mother are similar to those on a male of a comparable age and body build because each pectoralis major muscle produces a visible elevation of the skin overlying it and there is no excessive accumulation of superficial fat. The mother also has a 3-year-old unaffected daughter. During her two pregnancies no change occurred in the subcutaneous tissue overlying the pectoralis major muscles which indicates a complete absence of mammary gland parenchyma.

The mother has American Indian, French Canadian, Irish, and Scotch ancestry. The 26-year-old father of the affected child is unaffected and has Scotch ancestry. The history of the family of both the mother and the father back to the great grandparents of the infant, data supplied by the

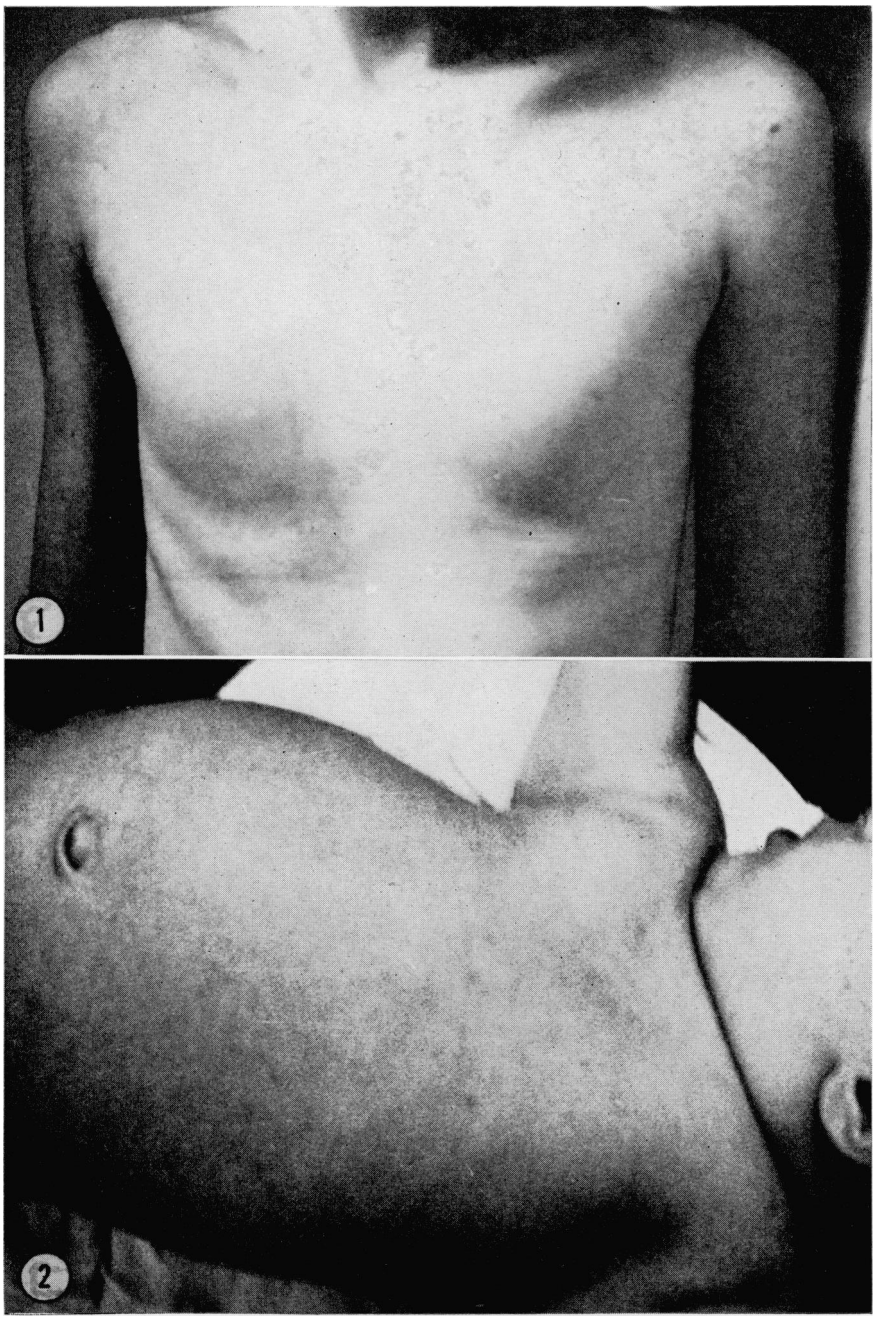

FIG. 1. Anterior view of the thorax of a 25-year-old female with bilateral amastia.

FIG.. 2. Anterior view of the thorax and abdomen of a 3½-month-old female with bilateral amastia. The child's mother is shown in Figure 1.

mother, did not reveal an occurrence of amastia. Since the history was largely second-hand information and incomplete, it is not possible to ascertain whether the amastia was transmitted as a hereditary trait to the affected mother or whether she is a mutant.

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