Proposal for Epidemiologic and

Genetic Study of Congenital Malformations

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I. <u>Objectives</u>

The objective of the study if fivefold:

1) To study the epidemiology of all congenital malformations which occurred in children of Collaborative Study mothers at birth and at one year of age.

2) To test the hypothesis of quasi-continuous distribution as an inheritance mechanism for most malformations.

3) To assess the genetic load due to congenital malformation utilizing the information on inbreeding.

4) To test the etiologic significance of several medical, genetic and socioeconomic factors in the occurrence of selected malformations.

5) To explore more fully previous findings suggesting a relationship of diabetes in the mother with the occurrence of congenital heart disease and possibly other malformations in the offspring.

II. Background

Numerous epidemiologic studies of congenital malformations exist in the literature, some containing information on racial and regional variation (Gittelsohn and Milham, 1965, Hewitt, 1965) but the majority of these were