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A GENETIC STUDY OF
CLEFT LIP AND CLEFT PALATE
AUCKLAND 1960-1976

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A thesis submitted in fulfilment of
the requirements for the degree of
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To those of our children thus afflicted

'cripplings visited upon us by
nature in her madcap moods;
games of blindman's buff among
the genes, all up and down the
double spiral staircase.'

Frank Sargeson

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ABSTRACT

A study of cleft lip and cleft palate was carried out in order to determine whether or not any differences in incidence between Europeans and Polynesians were accompanied by differences in recurrence risks, and to test the genetic hypotheses currently favoured as explanations of familial aggregation of these disorders.

An incidence study was undertaken on all live births in the Auckland urban area for the years 1960 to 1976. Family information was obtained from these probands and from other affected persons or their close relatives, by interview at the cleft palate clinic at Middlemore Hospital.

The ascertainment probability for cleft lip and cleft palate probands was about 95% and was not correlated with any of the demographic characteristics measured on the probands. After correction for ascertainment, the incidence of cleft palate in Maoris was estimated to be 1.867/1000 live births. For Europeans the estimate was 0.643/1000. The corresponding figures for cleft lip with or without cleft palate were 0.397/1000 and 1.195/1000. The sex ratio for cleft palate was 0.485 with heterogeneity between the races. For cleft lip the sex ratio was 0.649 overall. There were no secular or seasonal trends in the incidence of facial clefts and no significant effects of maternal age, or paternal age. The mean birth rank for probands with cleft lip with or without cleft palate was higher than expected. For probands with cleft palate, mean birth rank was not significantly elevated. The pattern of additional malformations in these probands was similar to those

reported in similar studies from other centres.

The recurrence risk for cleft palate was 1.8% overall. Although it was slightly higher in Polynesian families than in European families, the difference was nowhere near statistical significance. For cleft lip the recurrence risk was 2.6% overall, with the risk being slightly higher in Polynesian families, but again not significantly higher than in European families. Using current analytical techniques, no discrimination was possible between a generalized single autosomal locus model and a multifactorial threshold model. A consideration of the parameter estimates for both models suggests that the multifactorial threshold model is the more appropriate one to use for the calculation of recurrence risks in complicated family situations.

It is concluded that further family studies of this nature would no longer be warranted unless hypotheses can realistically be tested on the samples available. However, incidence studies in special populations will remain important for hypothesis testing. Following on the work using animal models, a study of face shape within and among races in New Zealand may provide clues to the aetiology of facial clefts, particularly isolated cleft palate. It will be important to follow changes in incidence over time and discover what effects intermarriage and cultural changes might have on the incidence of facial clefts.

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