A (4;21) translocation, unbalanced, 46 chromosomes

Repository identification No. GM-98

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Cell culture established from the skin biopsy of a 10-month-old male suffering from multiple congenital anomalies and slow development. Cultures of his peripheral blood and skin fibroblasts showed all his cells to have 46 chromosomes with a chromosome missing from the G group and a metacentric marker similar in size to chromosome 16. Banding techniques demonstrated a translocation of the short arm of chromosome 4 with the short arm of chromosome 21. His cells have a chromosome complement of 46, XY, der(21), t(4;21)(pll;pl2)mat. His mother and sister are both carriers of the balanced form of the translocation, 46, XX, t(4;21)(4pter- > 4pll::21pl2- > 21qter;21pter→21pl2::4pll→4qter). A brother died at age 2 weeks and is presumed to have had the same translocation on the basis of multiple congenital anomalies. A trypsin-Giemsa banded karyotype is shown in fig. 1.

Seed stock is stored in tissue-culture passage 5.

Supported by U.S. Public Health Service Contract No. N01-GM-3-2112 to the Institute for Medical Research, Camden, N.J.

Human Genetic Mutant Cell Repository

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Fig. 1. Trypsin-Giemsa banded karyotype 46, XY, der(21), t(4:

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