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## VI. CASE REPORTS

# Syndrome Identification

What's in a name? This question is often asked of a genetic counselor when a syndrome is newly delineated. The brief case reports that follow demonstrate the importance of establishing precise diagnoses. They also emphasize that many of these syndromes are recognizable only after careful physical examination of the proband and family members,

consultation with other subspecialties (for example, neurology, radiology, orthopedics, dentistry, pathology), and a review of the medical literature.

While it is true that there usually is no treatment for the basic condition, complications can often be anticipated and serious consequences averted. Thus, the diagnosis of Pierre Robin anomalad forewarns

the physician of the risk of airway obstruction in the prone position and of feeding difficulties in early infancy. Knowledge of cervical spine anomalies in chondrodystrophy alerts the neurologist to central nervous system signs or symptoms that may result from basilar brain compression.

A second benefit of syndrome identification is that a better prognosis regarding such important factors as longevity, morbidity, and ultimate adult height and mentality may be made. The dysmorphic infant with Saethre-Chotzen syndrome is not at substantial risk for mental retardation. Recognition of the earlier signs and dominant nature of the familial form of amyotrophic lateral sclerosis enables better counseling regarding longevity and morbidity.

Knowledge of the mode of inheritance provides risk figures for recurrence so that family planning can be practiced. It is not sufficient to state that the patient with a dominant condition has a 50% risk that each offspring will inherit the disorder; as demonstrated in the families with Pierre Robin anomalad and Saethre-Chotzen syndrome, some persons have

minimal manifestations, thus reducing the significance of the existence of the condition. In the case of sex-linked diseases, as in the family with chondrodystrophy, identification of female carriers can establish a 50% risk that male offspring will inherit the disorder. Further studies in the family with ring 9 chromosome abnormality should establish whether the proband, her mother, or other family members are at risk for bearing anomalous offspring.

Often patients seen in our clinic have been evaluated elsewhere for one or more components (for example, short stature, dysmorphic features, mental retardation, congenital malformations) of their disorder, but a precise diagnosis has not been made. Identifying the syndrome provides a diagnosis and permits a proper focusing on the disease process. While evaluation is often time-consuming, the reward is a family gratified by the establishment of a diagnosis that will allow a better understanding of the condition.

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## A Report of Familial Ring (9) Chromosome

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Ring chromosomes originate in the simultaneous occurrence of two breaks at opposite ends of the

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chromosome and the subsequent reuniting of the free ends to form a ring. They may be compatible with normal life, as only a fractional loss of genetic material has occurred, or they may lead to spontaneous abortion or to an offspring with severe physical and mental handicap attributable to significant genetic alterations.

The dearth of published case reports underscores