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Hypoparathyroidism, retarded growth and development, and dysmorphism or Sanjad-Sakati syndrome: an Arab disease reminiscent of Kenny-Caffey syndrome

EDITOR—In the December 1998 issue of *Journal of Medical Genetics*, four letters discussed whether or not Kenny-Caffey syndrome (KCS) is a part of CATCH 22.[1–4] These were based on a report of four Bedouin sibs with an unusual form of KCS that includes the additional features of marked IUGR, severe psychomotor retardation, and microcephaly. Two of the affected sibs and their phenotypically normal mother were found to have microdeletion 22q11.[5] The same authors subsequently suggested that this entity represents the Arab variant of KCS and because of some clinical resemblance to DiGeorge syndrome (DGS) they suggested that the phenotype is the result of 22q11 microdeletion or some abnormality of chromosome 10p where a second locus for DGS lies.[6]

This entity was originally described by Sanjad *et al*[7 8] in 1988 and 1991 as a new syndrome consisting of congenital hypoparathyroidism, seizures, growth and developmental retardation, and dysmorphic features in a group of Arab children of consanguineous parents. Several other reports followed.[9–14] All were Arabs, particularly Bedouin, with several sets of multiple affected sibs. The children have recognisable identical facies with deep set eyes, depressed nasal bridge with a beaked nose, long philtrum, thin upper lip, micrognathia, and large, floppy ear lobes. Medullary stenosis and other skeletal defects were found in most of them. This, together with the hypocalcaemia, hyperphosphataemia, and low concentration of immunoreactive parathyroid hormone in some of them, makes the phenotype similar to KCS. Recently the syndrome was localised to 1q42-43 by three independent groups.[15–17] Although they have a similar phenotype, different locus names have been given, namely *HRD* for hypoparathyroidism, retarded growth and development, and dysmorphism,[14] *KCS* for autosomal recessive KCS,[15] and *SSS* for Sanjad-Sakati syndrome.[17] The name of the syndrome in OMIM is hypoparathyroidism-retardation-dysmorphism and is given the number 241410 with autosomal recessive inheritance considered certain. The acronym Sanjad-Sakati syndrome is shown as an alternative. It is also listed among the Arab diseases in our text book *Genetic disorders among Arab populations*.[18]

With regard to the family with 22q11 microdeletion, the association is probably fortuitous or as a remote possibility it might be the cause in a subset of patients with this Arab disease. This issue should be easily resolved at the molecular level.

I believe that it is important for us (practitioners and scientists) to decide on one term for this disorder. For convenience and in order to credit the original authors, I suggest the acronym "Sanjad-Sakati" for the syndrome and *HRD* for the locus, an abbreviation which refers to the components of the syndrome.

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