



3-M Syndrome Clinical Phenotype is Still the Only Mean for Prenatal and Postnatal Diagnosis

E. Lapi¹, A. Cecconi¹, M.L. Giovannucci Uzielli¹, N. Salfi², L. Guarino¹

¹Human Genetics Service, Department of Paediatrics, University of Florence, Italy;

²Institute of Anatomy and Histology, S. Orsola Hospital, University of Bologna; Italy

In 1975, Miller, McKusick, Malvaux et al. reported a new form of low-birth weight dwarfism with normal intelligence, later called 3-M syndrome (after the initials of the first three authors). As of 1994 about 30 cases have been reported from different ethnic groups. The recurrence of the syndrome in sibs, with normal parents, provides evidence for an autosomal recessive disorder (MIM *273750). Diagnostic criteria are: prenatal onset dwarfism, dolicocephaly, joint hypermobility, slenderness of the shafts of the long bones and ribs and foreshortening of lumbar vertebral bodies, abnormal small pelvis, short femoral necks and vertical talus.

We report the occurrence of 3-M syndrome in two new unrelated families; the long follow-up offers important subjects for discussion.

In the first family the proposita at birth showed a Larsen-like phenotype with flat face, very short stature and generalized joint hypermobility. The clinical aspects greatly changed in the years and we were able to give the diagnosis of 3-M syndrome when she was 15 years old.

In the second family the propositus was a boy, showing obvious features since his birth: mild joint hypermobility, omphalocele, bilateral inguinal hernia and macrocephaly. At the age of 7 years the phenotype is still highly significant.

Two subsequent fetuses in the same sibship have been demonstrated affected by 3-M syndrome. The diagnosis was made after the 20th gestation week by means of ecography, and was confirmed after the termination, by both clinical and X-ray examination. Fetal parameters were not significant in the earlier stages of gestation. Of special interest are some-radiological aspects in both parents, whose stature is in the low normal range.

A search in the church registry, going back to 1704, revealed no parental consanguinity, but the same family name is present in both paternal and maternal pedigrees.

Correspondence: E. Lapi, Human Genetics Service, Department of Paediatrics, University of Florence, 50123 Florence, Italy.