

Lethal neonatal bone marrow failure syndrome with multiple congenital abnormalities, including limb defects, due to a constitutional deletion of 3' MECOM

Lars T. van der Veken,¹ Merel C. Maiburg,¹ Floris Groenendaal,² Mariëlle E. van Gijn,¹ Andries C. Bloem,³ Claudia Erpelinck,⁴ Stefan Gröschel,^{4#} Mathijs A. Sanders,⁴ Ruud Delwel,⁴ Marc B. Bierings⁵ and Arjan Buijs¹

¹Department of Genetics, ²Department of Neonatology, ³Department of Immunology, University Medical Center Utrecht, Wilhelmina Children's Hospital, Utrecht University; ⁴Department of Hematology, Erasmus University Medical Center, Rotterdam and ⁵Department of Pediatric Hematology and stem cell transplantation, University Medical Center Utrecht, Wilhelmina Children's Hospital, Utrecht University, the Netherlands

[#]Present affiliation: German Cancer Research Center [DKFZ], Molecular Leukemogenesis Group and Internal Medicine V, University Hospital Heidelberg and German Cancer Consortium [DKTK], Heidelberg, Germany

Correspondence: a.buijs@umcutrecht.nl
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Supplementary Table. Characteristics of neonate with syndromic BMF associated with chromosome 3q26.2 interstitial deletion affecting the 3' region of *MECOM*.

Prenatal ultrasound at 20 weeks	club feet polyhydramnios normal growth
Birth	38+5 weeks gestation weight 3730 g (50 th -84 th centile) length 50 cm (50 th centile) head circumference 38 cm (>97 th centile) Apgar scores at 1', 5' and 10' were 2, 4 and 6, respectively
Hematology	anemia (2.1 mmol/l) thrombocytopenia ($52 \times 10^9/l$) leucopenia ($1.3 \times 10^9/l$) absence of granulocytes none but T-lymphocytes hypocellular bone marrow
Neurology at day 1	reduced gyrfication wide peripheral cerebrospinal fluid spaces and fissures deep sulci large cavum of the septum pellucidum small vermis convulsions
Cardiopulmonary	respiratory failure structurally normal heart
Skeletal	bilateral club feet short mid phalanx fifth finger (right) ulnar deviation (right)
Dysmorphic features at day 2	macrocephaly minor facial dysmorphisms (prominent tragus of the ear, small chin) short neck deep palmar creases
Skin	edematous cutis marmorata