Molecular diagnosis reveals genetic heterogeneity for the overlapping MKKS and BBS phenotypes

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Résumé en anglais	Hydrometrocolpos and polydactyly diagnosed in the prenatal period or early childhood may raise diagnostic dilemmas especially in distinguishing McKusick-Kaufman syndrome (MKKS) and the Bardet-Biedl syndrome (BBS). These two conditions can initially overlap. With time, the additional features of BBS appearing in childhood, such as retinitis pigmentosa, obesity, learning disabilities and progressive renal dysfunction allow clear differentiation between BBS and MKKS. Genotype overlap also exists, as mutations in the MKKS-BBS6 gene are found in both syndromes. We report 7 patients diagnosed in the neonatal period with hydrometrocolpos and polydactyly who carry mutations in various BBS genes (BBS6, BBS2, BBS10, BBS8 and BBS12), stressing the importance of wide BBS genotyping in patients with this clinical association for diagnosis, prognosis and genetic counselling.
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