INTRODUCTION

Analploid small supernumerary marker chromosomes (asSMC) are a rare subclass of C-band-negative sSMC that are devoid of alpha-satellite DNA. These marker chromosomes cannot be identified unambiguously by conventional banding techniques alone, being necessary to apply molecular cytogenetic methods in favour of a detailed characterization [1]. Approximately 135 asSMC have been described until now involving 20 of the 22 autosomes and both sex chromosomes, with about 50% resulting from chromosomes 15, 13 and 8 [2]. Only three cases are sSMC(7) and just two of them have been reported with a sparse details [2].

In terms of clinical findings, there are three major groups to be considered: no clinical consequences (~5%), moderate to severe clinical consequences (~75%) and the largest group with the most severe clinical consequences. If an analploid sSMC does not cause an imbalance and/or the sSMC causing a large imbalance is present only in mosaic, clinical findings may be absent or mild [2].

We report on a child with several dysmorphic features and severe development delay presenting a de novo analploid sSMC in mosaic, which by molecular cytogenetics and microarray analysis was shown to be originated from the terminal arm of chromosome 7 and to harbour an invdup rearrangement of 7p35-qter region.

PATIENT CLINICAL INFORMATION

The patient was a newborn male, the first child of an healthy and non consanguineous couple. He was born at 28th week of gestation with normal somatometric parameters (weight 1167g corresponding to a PS0). In the neonatal period he had respiratory distress syndrome and feeding difficulties. He was diagnosed with bronchopulmonary dysplasia (psp-prematurity?). At physical examination he revealed open and wide anterointerior, bifrontal narrowing, bilateral eyelid edema, low insertion ears, short nose with wide and depressed root, small mouth, full lips, short neck, fingers and toes nail hypoplasia. He had bilateral hip dysplasia, cryptorchidism and bilateral inguinal hernia. He was submitted to a tracheotomy and a gastrostomy.

Currently, he is 10 years-old and has severe development delay without language, bruism and auto-aggressive behaviour (started at age of 8 years-old). He has an unstable, assisted broad based walk. He still depends on enteral feeding and has a tracheostomy tube for breathing. The physical exam reveals an accentuation of the dysmorphic features with straight eyebrows with synophrys, short palpebral fissures, opened eye sockets, and a high palate.

RESULTS & DISCUSSION

Cyto genetic analysis revealed a mosaic karyotype with the presence of an sSMC, de novo, in 20% of the lymphocytes and 73% of the fibroblast cells (Fig. 1A). FISH analysis with alpha-satellite probes for all chromosomes indicated that the sSMC was an analploid marker (Fig1B), while the presence of euchromatin material was revealed with WCP(7) (Fig 1C). Hybridization with RP11-298A10 and TelVision 7q probes, allowed establishing that the asSMC results of an invdup involving the terminal region of chromosome 7.

GTL-banded metaphase chromosomes were prepared using standard techniques and FISH analysis was performed according to standard procedures on fixed cells from the cytogenetic analysis.