The Genomic Era: The Future of Genetics in Medicine - Spot the difference answer

The karyotype in picture 1 is abnormal showing an interstitial deletion of the short arm of chromosome 5. This would be written on a report as 46,XY,del(5)(p13p15).

This deletion on chromosome 5 would be associated with Cri du Chat syndrome. Cri du Chat syndrome is more commonly associated with a terminal deletion of the short arm of chromosome 5 and is therefore also known as 5p- syndrome. The condition is characterised by a moderate to severe intellectual disability, small head size (microcephaly), weak muscle tone (hypotonia) in infancy and some children are born with a heart defect. Affected individuals also have characteristic facial features, including widely set eyes (hypertelorism), low-set ears and a small jaw (see image below). The condition is called cri du chat because young babies can have a distinctive cry that is said to resemble that of a cat.

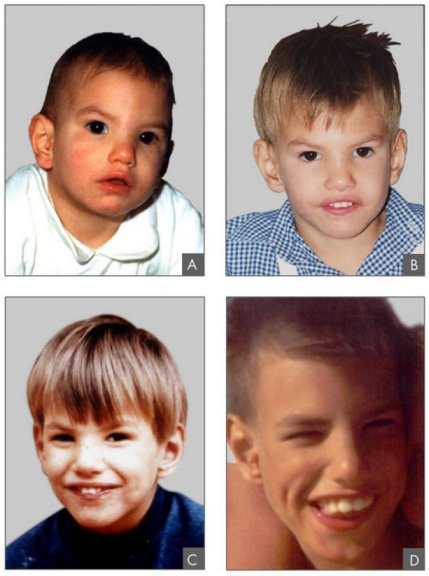

Figure: Clinical features of a patient with Cri du Chat syndrome at age of 8 months (A), 2 years (B), 4 years (C) and 9 years 6/12 (D)