Piebaldism with a Variant Gene

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Piebaldism is patchy areas of depigmentation on the skin, most frequently on the forehead (producing a white forelock), ventral trunk, elbows, and knees. It is a rare autosomal dominant condition, caused by mutations in the cell-surface receptor tyrosine kinase gene (KIT). Piebaldism must be differentiated from other pigmentation disorders, such as vitiligo, nevus depigmentosus, and Waardenburg syndrome.

We present a preterm baby boy born at 32^{+} weeks due to antepartum hemorrhage. The birth weight was 1.85 kg. The baby was found to have a white forelock (Piebaldism) and gray eyebrows and eyelashes. His neonatal course was complicated by mild respiratory distress syndrome which was managed by one-day intubation and assisted ventilation (on PTV mode). The gene panel revealed that the baby carries in exon 16 of KIT the variant of uncertain significance c.2318C>T p.(Ser773Phe) in heterozygous state.

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