

WHITE SPOTTING (Congenital Absence of Melanocytes)

White spotting (sometimes previously referred to as piebaldism) is defined as the congenital absence of viable melanocytes from areas of the body or from the entire body (10, Baxter, et al., 2004). This is not albinism, in which pigmentation is reduced because of defects in melanogenesis, but the melanocytes are viable. White spotting results from a defect during the development of melanocytes in the latter part of embryonic development. Normally, the melanoblast arises as a neural crest derivative which rapidly replicates. The descendants migrate laterally through the dermis and epidermis to eventually reside in their proper locations where they become mature melanocytes and begin to produce pigment just after birth. Clearly, many intra- and inter-cellular processes are required for the successful completion of such a complex set of tasks that include:

1. Original determination of the lineage;
2. Replication of the melanoblasts;
3. Interaction of the migrating melanoblasts with the skin cells through which they are migrating, including supporting messages from the skin;
4. Colonization of the hair follicles, where most of them reside, or other locations such as the iris or choroids of the eye or the inner ear;
5. Differentiation and survival upon arrival.

The white spots on the mouse represents areas where one of the above processes has failed, with the resulting failure of melanocyte survival. The problem can reside within the melanoblast/melanocyte or in the surrounding cellular environment (usually this is the skin). Mice with white spotting permit us to identify genes that are necessary for the success of these tasks.

A third class of genes is not yet well defined, but is known to include several gene loci that do not cause melanoblast/melanocyte death, but do impact the location on the body where the pigment cells will be absent, for example, the white spot will be more anterior or more posterior on the trunk of the mouse, or larger or smaller. Because of these modifying genes, the phenotypes produced by a given genotype can vary widely with mouse genetic background, showing the importance of congenesis for any critical evaluation of spotting pattern.

References

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