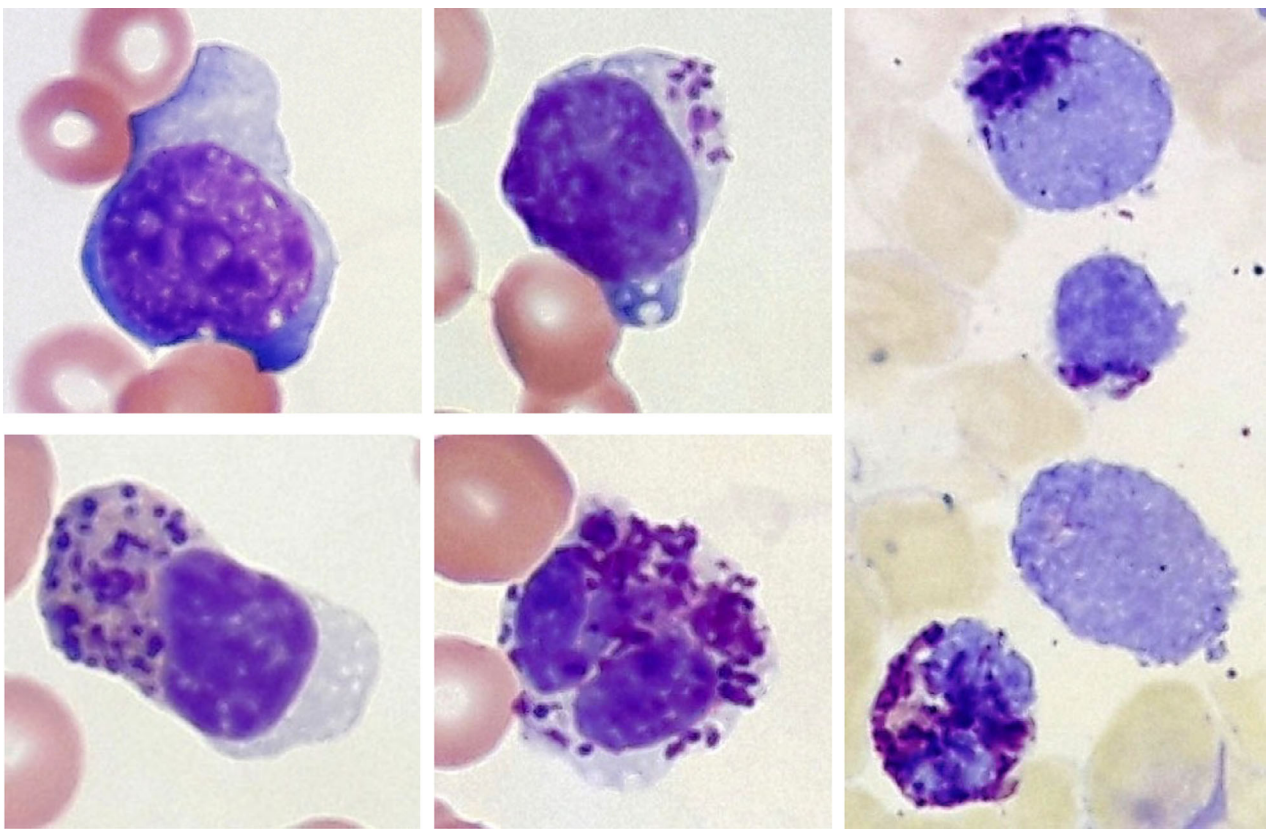
Acute basophilic leukaemia secondary to MDS with isolated del(5q)



A 63-year-old woman was admitted with asthenia. She had a two-year history of myelodysplastic syndrome (MDS) with isolated del(5)(q12-q33) and had never needed transfusion or other treatment. Physical examination was normal. Her full blood count showed haemoglobin concentration 64 g/l, platelets 5 × 10^9/l and white blood cells 6·1 × 10^9/l. A blood film showed 15% basophils, 9% agranular blast cells and 21% blast cells with basophilic granules. Serum tryptase was raised, at 24·8 μg/l, and histamine was 2000 pg/ml (normal range: <1100 pg/ml) without related symptoms. A bone marrow aspirate showed hypercellularity and marked dysplasia with 20% blast cells and 18% basophils. The blasts were medium-sized with a round nucleus and moderately basophilic cytoplasm containing a variable number of coarse basophilic granules with a polar distribution; there were also confluent vacuoles merged with the cell membrane, suggestive of degranulation (left and centre). The granules stained metachromatically with toluidine blue (right). Flow cytometric immunophenotyping demonstrated immature blast cells, which were CD34+, CD33+, CD13+, CD56+, CD59+ and CD117+, and blasts showing differentiation to basophils, which were CD33+, CD13+, CD203c+, CD123+, CD38+ and CD11b+. Myeloperoxidase was negative in both populations. Conventional cytogenetic analysis and fluorescence *in situ* hybridisation showed del(5)(q12-q33) only. Molecular studies were negative for *BCR-ABL1* and for *NPM1, FLT3, IDH1* and *KIT* mutations.

Patients with MDS with isolated del(5q) infrequently progress to acute leukaemia. Acute basophilic leukaemia is rare. It accounts for <1% of cases of acute myeloid leukaemia and its development after MDS with isolated del(5q) is particularly rare.

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