A patient with Ellis-van Creveld (EVC) syndrome

- 4 year-old Hispanic male with EVC syndrome; his sister 13 months older with similar features. Pathological findings are from autopsy and bone marrow (pre-mortem).
- EVC [1-5] is a rare autosomal-recessive disorder characterized classically by the tetrad:
  1. Chondrodysplasia: short-limb dwarfism, length < 5th percentile
  2. Bilateral post-axial polydactyly
  3. Ectodermal dysplasia: hypoplastic spoon-shaped nails, multiple frenula with gum abnormalities, supernumerary upper teeth including one conical
  4. Cardiac malformation: cor triatratum, ostium primum and secundum atrial septal defects

HEMATOLOGIC FINDINGS

Peripheral blood examination
- Normochromic microcytic RBCs
- Hgb 13.9 g/dl
- Hct 40.8%
- Anisopoikilocytosis
- Many nucleated RBCs
- Leukocytosis (47,900/mm3) with left shift
- 54% neutrophils
- 14% band forms
- 10% lymphocytes
- 1% monocytes
- 18% metamyelocytes
- 3% myelocytes
- Granulocytes without dysplastic changes
- Decreased platelets (73,000/mm3) with a few large forms

Bone marrow aspirate and biopsy
- Erythroid hyperplasia (M:E ratio 0.9:1) with numerous dysplastic forms
- Normal granulopoiesis and megakaryopoiesis
- Decreased iron stores
- Erythroid hyperplasia
- Marked dyserythropoiesis
- Abnormal mitosis
- Decreased RBCs
- Basophilic stippling
- Decrease in megakaryocytes
- 0.9:1 M:E ratio
- Increased band forms
- 54% neutrophils
- 10% lymphocytes
- 5% monocytes
- 3% myelocytes
- 18% metamyelocytes
- 15% myeloblasts
- 5% granulocytes
- 1% lymphocytes
- 1% monocytes
- 10% bands
- 5% myelocytes
- 15% metamyelocytes
- 5% band forms
- 15% neutrophils

DISCUSSION

Myelodysplastic syndromes (MDS) are clonal hematopoietic disorders associated with dysplasia in one or more myeloid cell lines [7]. MDS may develop as primary disorders with no known etiology or as therapy-related secondary disorders. Secondary MDS have been associated with chemotherapeutic agents and radiotherapy. The patient presented had no known exposure to such agents. MDS typically affect older adults (median age: 70 years) and are uncommonly encountered in the pediatric population.

However, MDS in pediatric patients with predisposing genetic abnormalities such as Down’s syndrome is a well-recognized association [8].

The dyserythropoiesis identified in this child with EVC syndrome may signify a unilineage myelodysplastic change or a primary myelodysplastic syndrome. In either event, the dyserythropoiesis may be a coincidental occurrence or may represent an unusual EVC syndrome association. Patients with EVC syndrome that present with unexplained hematologic findings may warrant evaluation for possible myelodysplastic change or MDS.

REPORTED INSTANCES OF HEMATOLOGIC ABNORMALITIES IN EVC SYNDROME ARE EXTREMELY RARE

A single case report from 1969 describes a neonate with a possible variant of EVC syndrome with acute myeloblastic leukemia [6].

REFERENCES