soho2023
ELEVENTH ANNUAL MEETING

Case Presentation

1- **25-year-old** male, presented with **disseminated HPV**, **bicytopenia** and bone-marrow **hypocellularity**. History of grade V vesicoureteral reflux.

2- Physical exam shows no lymphadenopathies or splenomegaly. No bleeding. **Multiple warts** on hands and some in genitalia.

3- Peripheral blood WBC 2.4 x 109/L, ANC 732, **monocytopenia**. Platelets 152 x 109/L. Bone-marrow biopsy hypocellular, megakaryocyte **dysplasia,** no blasts. Flow cytometry: loss of monocytes, dendritic cells, precursor/mature B-cells.

4- **GATA2-deficiency** (Sanger sequencing), with a **germline** mutation p.Arg361Cys. Normal 46,XY.

5- Myeloid NGS panel: acquired ASXL1 and STAG2, concerning for evolving **hypoplastic-MDS**.

6- Started on preventive Azithromycin 100 mg, and a donor search for **pre-emptive** HSCT.

Myeloid neoplasia is a late presentation in AYA with GATA2 deficiency syndrome

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AYA with GATA2 Deficiency Syndrome with Evolving Myeloid Neoplasia

GATA2 deficiency is a **germline** myeloid predisposition syndrome. It affects the **hematopoietic** and **immunologic** systems.

Inheritance is autosomal dominant with more than 150 discovered pathogenic variants. Hematologic manifestations start with **cytopenia** and acquisition of somatic mutations (ASXL1, STAG2) and/or monosomy 7, trisomy 8, for **clonal evolution**. Most cases are diagnosed in the setting of established myeloid neoplasm.

Extra-hematologic manifestations, like recalcitrant warts/ disseminated **HPV, genito-urinary** malformations, lymphedema, etc, should also prompt **screening**, by germline assessment.

Allogeneic HSCT offers cure to GATA2 deficiency syndrome. Related donors should be screened and not be used if positive for a germline variant.