
Holly Tavianini RN, BSN, MSHSA, CNRN  
*Lehigh Valley Health Network*, Holly.Tavianini@lvhn.org

Amanda Yerkes RN, BSN, CMSRN, CNRN  
*Lehigh Valley Health Network*, Amanda.Yerkes@lvhn.org

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ABSTRACT: The American Stroke Association reports more than 795,000 strokes/year; of these 3.7 – 5.5% occur in people under 45 years of age. Stroke costs an estimated $34 billion annually including the cost of healthcare, medications, and missed days of work. The etiology of stroke in the young is based more on genetic markers and resulting blood dyscrasias. Sickle cell disease is associated with 26% mortality from stroke. Fibromuscular dysplasia increases risk for aneurysm and arterial wall irregularities which causes stenosis, dissection and stroke. Fabry’s disease results in abnormal deposits of fatty substances in blood vessel walls, significantly increasing risk for ischemic stroke, subarachnoid or intracranial hemorrhage. This presentation will use 2 case studies highlighting genetically-linked disorders that hide in the blood and increase stroke risk in this unsuspecting patient population. Learners will become familiar with unusual stroke presentations and will gain insight into predispositions leading to stroke in the young.

INTRODUCTION

In US, acute ischemic stroke hospitalizations increased by almost 44% in adults ages 25-44 during 2000-2010

Stroke and transient ischemic attacks in the young compared to elderly

- Profound and long-lasting impact
- Greater economic impact due to inability to work during productive years

African and Hispanic Americans

- Suffer stroke at earlier age
- African Americans - twice as likely to die from a stroke compared to Caucasians
- Hispanic American Mortality
  - 1 in 4 male and 1 in 3 female strokes result in death

Obesity and lipid abnormalities are rising in younger populations

- Increase risk for cardiovascular disease

Genetic disorders may contribute to cerebrovascular disease

- Sickle Cell Disease
  - Stroke common in pediatric and young adults
  - Estimated 25% of patients stroke by age 45
  - Vascular disease of the brain associated with large vessel vasculopathy
  - Middle and anterior cerebral arteries and internal carotid artery
  - Other underlying disorders undiagnosed until after suffering a stroke

OBJECTIVES

- Discuss stroke statistics for multiple age groups and outline the emotional, physical, and financial impact.
- Review pathophysiology of genetic mutations within cerebral vasculature that increase predispositions for stroke in patients less than 45 years of age.
- Using case studies of 2 young stroke patients, illustrate the devastating effects of unsuspected blood dyscrasias or genetic abnormalities.

CASE STUDY 1

Fabry’s Disease:
- Inherited disorder of lipid metabolism resulting from deficient activity of enzyme alpha-galactosidase (a-Gal A)
- Affects 1 in 40,000 to 60,000 males
- Leads to progressive kidney damage, heart attack and stroke
- Consideration in cryptogenic stroke in young patients
- Clinical presentation: hemiparesis, vertigo, diplopia, dysarthria, and memory loss
- Treatment: enzyme replacement and antiplatelet medications

Fibromuscular Dysplasia:
- Abnormal growth of fibrous tissue in the walls of arteries that can cause the vessels to narrow and bulge resulting in aneurysms, stenosis and dissections
- Carotid (25-30%) or renal (60-75%) arteries; most patients have evidence of FMD in more than one artery
- 7-11% of cases inherited
- Other causes: abnormal development of arteries that supply the vessel wall → inadequate oxygen supply and anatomic position of the artery within the body
- Predominantly seen in young women of childbearing years
- No cure
- Treatment: antplatelets, antihypertensives, tobacco cessation, and pain management

Presentation:

- CT Scan Head - confirmed subarachnoid hemorrhage with a Hunt and Hess Grade 5
- Outcome:
  - CT Scan Head - confirmed subarachnoid hemorrhage with a Hunt and Hess Grade 5
  - Transplantation, and intravenous tissue plasminogen activator (tPA) therapy
  - Multiple days of ischemic infarcts and painful crises
  - Transplantation, and intravenous tissue plasminogen activator (tPA) therapy

Imaging:

- CT Scan Head - multiple small foci of encephalomalacia in the right basal ganglia and centrum semiovale
- MRI Brain - Greater than 40 foci of acute infarction in bilateral frontal and parietal lobes, right occipital lobe and lateral aspects of the temporal lobes
- CT Angiography - thrombectomy on M1 segment of right middle cerebral artery (MCA)

Intervention:
- Thrombectomy - right MCA m1 thrombectomy with stent deployment

Outcome:
- Transferred to rehabilitation facility

CONCLUSION

In the presence of stroke in the young, consideration of genetically-linked disorders of the blood should be considered for cause.