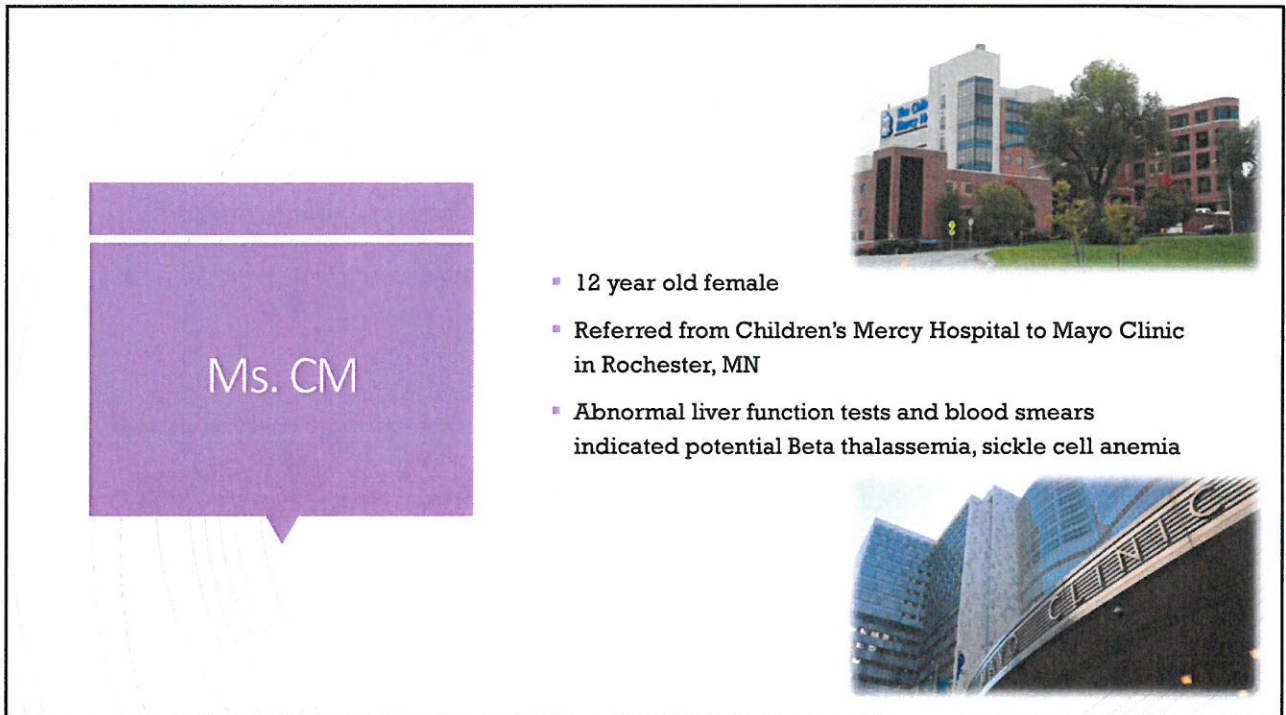


# Fighting the Good Fight: A Case Study

Mya Masterson

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Ms. CM

- 12 year old female
- Referred from Children's Mercy Hospital to Mayo Clinic in Rochester, MN
- Abnormal liver function tests and blood smears indicated potential Beta thalassemia, sickle cell anemia



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## Ms. CM Previous History

Enlarged spleen and liver (hepatosplenomegaly)

Liver

Spleen

#ADAM

- Birth weight: 4 lbs 2 oz, 3 weeks premature
- Born with anemia, jaundice and hepatosplenomegaly
- Received an exchange transfusion as a newborn to help eliminate iron deficient blood and placed on an iron supplement to help with anemia.

Normal amount of red blood cells

Anemic amount of red blood cells

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## Ms. CM Presentation at time of Diagnosis

- Hepatosplenomegaly
- Abnormal liver function tests
- Anisopoikilocytosis
  - Teardrop cells, Spherocytes, Schistocytes, Target cells, Elliptocytes
  - Basophilic Stippling, Howell Jolly bodies, Pappenheimer bodies
- Hemoglobin: 9.6 g/dL (normal female range 12-15 g/dL)
- Mean Cell Volume: 105 fl (normocytic 80-100 fl)
- Reticulocyte Count: 2.3% (normal female range 0.5-1.6%)
- Elevated iron levels

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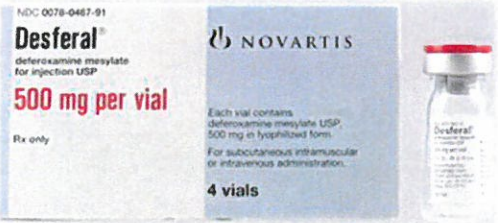
Diagnosis

Her presentation correlated with Congenital Dyserythropoietic Anemia, Type I

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Deferoxamine Administration for Elevated Iron Levels

- Deferoxamine is an iron chelation agent
- 500 mg was administered – chelating 2,781 ug/kg
- This incredibly elevated level was conclusive for Idiopathic Hemochromatosis



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## Iron Studies



Completed at time of diagnosis  
 \*Normal range from Saint Luke's Health System  
 \*\*Table used from *Mayo Clin Proc* 57:322-325, 1982

Iron Studies	Patient	Normal
Serum Iron (ug/mL)	200	60-180
Total Iron Binding Capacity (ug/mL)	282	240-450
Saturation (%)	71	18-50
Ferritin (ng/mL)	>2,000	20-200*
Deferoxamine Chelatable Iron (ug/kg)	2,781	274 +/- 18.2
Liver Iron (mg/g of dry tissue)	19	<1.0

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## Treatment

- Ms. CM underwent weekly phlebotomies for 6 weeks
  - Worsened her condition – decreased her Hgb, liver function tests continued to increase and progressed enlargement of liver
- Experimental subcutaneous deferoxamine treatment
  - Ms. CM was placed on a pump that continuously administered the chelating agent 12 hours a day, 5-7 days a week, for 14 months
  - Added a daily dose of vitamin C to help with iron excretion

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## Subcutaneous Deferoxamine Treatment

- Over the 14 months, Ms. CM's ferritin decreased to 292 ng/mL
  - Another liver biopsy showed her dry tissue iron content decreased to 1.2 mg/g
- Upon ceasing treatment, her ferritin levels increased over time – not unexpected
- Ms. CM was off and on the subcutaneous deferoxamine pump for the following several years
  - Until tests showed she could discontinue this treatment and return to phlebotomies (~1 every 3 months)

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## Rarity of Congenital Dyserythropoietic Anemia

- About 150 cases of Congenital Dyserythropoietic Anemia (types I, II, III) at time of diagnosis
- 4<sup>th</sup> known case in the world to have the diagnosis of CDA Type I, with Hemochromatosis
  - Studies showed, most others with this combination of disorders don't survive long due to complications with the hemochromatosis.



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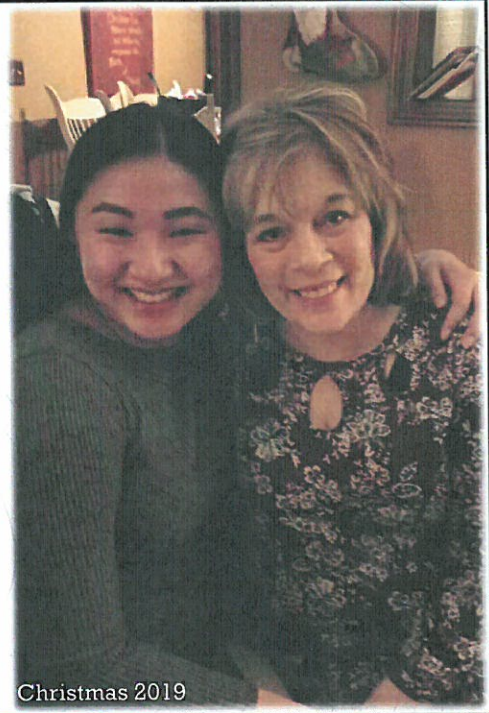
## Life Post-Diagnosis

- 1981: Cholecystostomy (due to stones caused by hemochromatosis) and Splenectomy (due to progression of enlargement)
- 1990: Contraction of virus caused enlargement of heart
- 1994: Miscarriage and placed on bedrest due to early and severe preeclampsia
- 2001: Liver Transplant (from cirrhosis caused by Hep C she contracted at birth from the transfusion)
- 2004: Diagnosis of post-transplant diabetes
- 2009: Diagnosed with an inoperable hepatic portal vein clot
- 2013: Life-flighted to Mayo Clinic for bleeding esophageal varices
- 2018: Diagnosed with Stage 4 diffuse large B-Cell lymphoma that metastasized from her small intestine to her lungs and pelvic bone

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## Where is she now?

As a retired pediatric nurse, she is now happily filling her time with scrapbooking, home improvements and keeping her family in line – all while continuing to work diligently at maintaining her health.



Christmas 2019

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A purple speech bubble graphic with a white outline, positioned on the left side of the slide. The text "Sources" is written in white, sans-serif font inside the bubble.

- <https://pubmed.ncbi.nlm.nih.gov/7078263-use-of-subcutaneous-deferoxamine-in-a-child-with-hemochromatosis-associated-with-congenital-dyserythropoietic-anemia-type-i/>
- Personal Interview with patient featured in case study
- <https://www.bizjournals.com/bizwomen/news/latest-news/2020/03/mayo-clinic-amazon-join-project-to-fight-covid-19.html>
- <https://www.missouribusinessalert.com/industries/60475/2015/06/15/health-care-checkup-childrens-mercy-adds-ranking-st-anthonys-outsources/>
- <https://www.newkidscenter.com/How-to-Get-Rid-of-Jaundice-in-Newborns-at-Home.html>
- <https://medlineplus.gov/ency/imagepages/17215.htm>
- <https://www.daviddarling.info/encyclopedia/A/anemia.html>
- <http://atlas.gechem.org/en/component/k2/item/1036-congenital-dyserythropoietic-anaemia-type-i>
- <https://mms.mckesson.com/product/436532/Novartis-00078046791>
- <https://www.vectorstock.com/royalty-free-vector/world-map-globe-cartoon-vector-23302808>

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