Putting A Spotlight On...
Wiedemann-Steiner Syndrome

What is Wiedemann-Steiner Syndrome?

- Wiedemann-Steiner Syndrome is characterized by developmental delay, intellectual disability, and characteristic facial features, with or without additional congenital anomalies.

What are the signs/symptoms?

- Signs/Symptoms include:
  - Delayed speech and language development
  - Thick eyebrows
  - Long eyelashes
  - Vertically narrow palpebral fissures
  - Widely spaced eyes
  - Wide nasal bridge with broad or bulbous tip
  - Lateral (or other) flare to the eyebrow
  - Downslanted palpebral fissures
  - Blepharoptosis
  - Exaggerated Cupid's bow
  - Thin vermilion border to the upper lip
  - Posteriorly rotated ears

What is Wiedemann-Steiner Syndrome a result of?

- Wiedemann-Steiner syndrome is a genetic disease, which means that it is caused by one or more genes not working correctly. Disease-causing variants, or differences, in the following gene(s) are known to cause this disease: KMT2A.

What are the complications of Wiedemann-Steiner Syndrome?

- Difficulty feeding
- Behavior problems
- Seizures
- Excessive hair on the elbows, arms, and back

References:

- https://www.ncbi.nlm.nih.gov/books/NBK580718/#:~:text=Clinical%20Description,or%20without%20additional%20congenital%20anomalies.
- https://rarediseases.info.nih.gov/diseases/5565/wiedemann-steiner-syndrome

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