Putting A Spotlight On...

Lenz Microphthalmia Syndrome

What is Lenz Microphthalmia Syndrome?

- Lenz microphthalmia syndrome is an extremely rare inherited disorder characterized by small eyes and/or droopy eyelid, resulting in visual impairment. Most affected children also exhibit developmental delays and intellectual disability.

What are the signs/symptoms?

- Signs/Symptoms include:
  - small eyes (one or both)
  - mild to severe delays in certain developmental milestones
  - mild to severe intellectual disability

What is Lenz Microphthalmia Syndrome a result of?

- Lenz microphthalmia syndrome is caused by pathogenic variants, or changes, in the BCOR gene and is inherited in an X-linked pattern. This condition is fully expressed in males only. However, females who carry a variant BCOR gene may exhibit some symptoms, such as microcephaly, short stature and/or anomalies of the fingers and/or toes.

What are the complications of Lenz Microphthalmia Syndrome?

- Abnormalities of the ears, teeth, hands, skeleton, and urinary system
- Heart defects (less common)
- Delayed development
- Intellectual disability

References:

- https://rarediseases.org/rare-diseases/lenz-microphthalmia-syndrome/#:~:text=Lenz%20microphthalmia%20syndrome%20is%20an,eyes%20(anophthalmos%20or%20anophthalmia).
- https://rarediseases.org/rare-diseases/lenz-microphthalmia-syndrome/#disease-overview-main

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