Bohring-Opitz Syndrome Foundation, Inc.

www.bos-foundation.org

Clinical Features of Bohring-Opitz Syndrome

What is BOS?

Bohring-Opitz Syndrome is an ultra-rare genetic condition with less than 85 reported cases in the world.

It is caused by a spontaneous mutation on the ASXL1 gene and affects the development of many parts of the body.

This list was created by Dr. Bianca Russell (Bianca.Russell@cchmc.org) and Dr. Wen-Hann Tan (Wen-Han.Tan@childrens.harvard.edu), medical advisors to the BOS Foundation.

Physical Features

- Proptosis (bulging eyes) with severe myopia
- Upslanting palpebral fissures (opening between eyelids)
- Variable microcephaly (smallness of the head)
- Micro- or retrognathia (small or recessed jaw)
- Depressed and wide nasal bridge
- Low-set, posteriorly angulated ears
- High palate with prominent palatine ridges
- Hypertrichosis (excessive hair growth)

Overall Features of BOS

- **Nevus flammeus over the glabella, which may become less obvious with time (birthmark on forehead)**
- **Abnormal neurodevelopment**
- **Postnatal growth retardation**
- IUGR (intrauterine growth retardation)
- Severe to profound intellectual disabilities
- Failure to thrive with significant feeding difficulties
- BOS posture (fixed contractures at the elbows)
- Hypotonia (low muscle tone)

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If you believe you know someone with BOS, contact the BOS Foundation at info@bos-foundation.org to get in contact with our medical advisors or to get a list of genetic testing options.