



# Bohring-Opitz Syndrome

Foundation, Inc.

[www.bos-foundation.org](http://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 250 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. Bianaca Russell and Dr. Wen-Hann Tan, medical advisors to the BOS Foundation.

### Overall Features of BOS

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

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### 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)

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**For more information contact the Bohring-Opitz Syndrome Foundation at:**

[info@bos-foundation.org](mailto:info@bos-foundation.org)

**Join the ASXL Chromatin Registry by emailing:**

[ASXL-CHROMATIN-REGISTRY@mednet.ucla.edu](mailto:ASXL-CHROMATIN-REGISTRY@mednet.ucla.edu)



