Bohring-Opitz Syndrome

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What is BOS?

- Bohring-Opitz Syndrome (BOS) is a very rare congenital disorder
- Fewer than 300 children in the world have been diagnosed with this syndrome
- It is a mutation on ASXL1 gene

History

- In 1999 Dr. Bohring presented four patients who had similar features to two other patients who had been reported on in 1975 and 1995
- He suggested these six children either represented the severe end of the clinical spectrum of 'Opitz trigonocephaly C syndrome' or comprised a separate entity.
- Since then there have been further reports of individual patients with similar findings.
  - The terms 'Bohring', 'Bohring-Opitz' and 'C-like syndrome' have all been used to describe this condition (Greenhalgh et al. 2003).
  - Because Oberklaid et al. (1975) reported another case there is also the name 'Oberklaid-Danks-Syndrome' for 'Bohring-Opitz-Syndrome' (Hasting et al. 2011).
- In 2011 genetic testing became available to move the diagnosis from just clinical findings to a genetic DNA test.

Overall Features

Overall Features

- 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)

Physical Features

Physical Features

- Glabellar nevus simplex (birthmark between eyebrows and above nose)
- Proptosis (bulging eyes) with severe myopia
- Upslanting palpebral fissures (opening between the eyelids)
- Variable microcephaly (smallness of 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)

Diagnosis

- Clinical diagnosis is based on doctor observations using the BOS key features
- Genetic diagnosis is the Whole Exome Sequencing Test or Whole Genome Decoding looking for mutations to ASXL1 gene

Diagnosis Barriers

- BOS Awareness
- Sequencing/Decoding cost and insurance reimbursement
- Availability of laboratory with capabilities

Prognosis and Treatment

- There is currently no cure for BOS
- BOS has a higher childhood mortality
- There is little known about the clinical management of children with BOS
- Treatment is geared toward minimizing symptoms and preventing complications

Rosemary Ross, age 31, is one of the oldest living patients with BOS.

Therapy

- Many BOS children attend various therapies to help improve symptoms
  - Vision Therapy
  - Occupational Therapy
  - Physical Therapy
  - Aqua Therapy
  - Speech Therapy
  - Equestrian Therapy

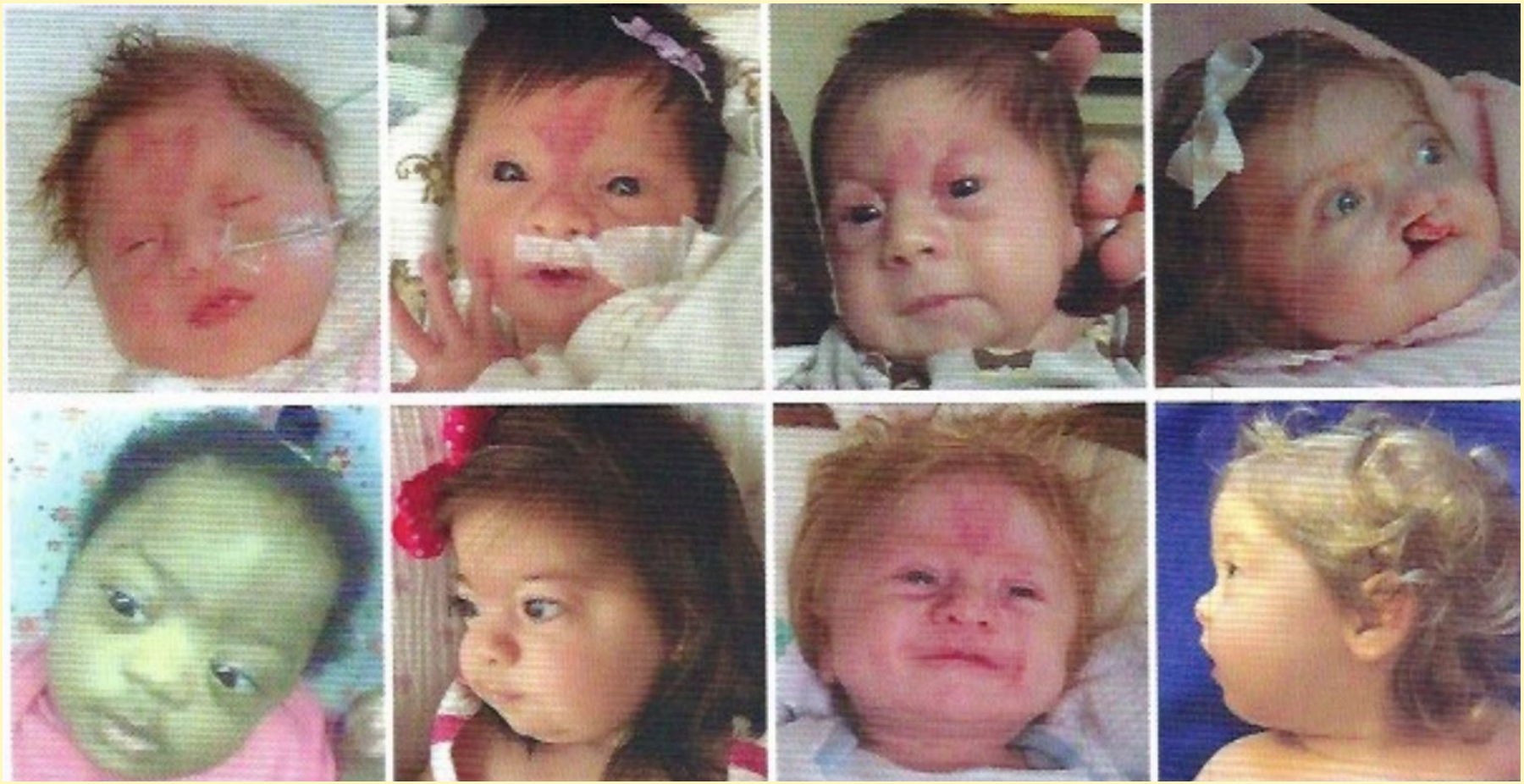
BOS Foundation

The BOS Foundation was founded in 2015 by parents of children with BOS. It is a 501c3 nonprofit organization dedicated to improving the lives of people affected by BOS.

We exist to:

- Help families and patients affected by BOS through family assistance programs
- Support research that increases the medical community's knowledge about BOS and its best treatments
- Teach and share knowledge about BOS, and its best practices and treatments
- Increase a sense of community surrounding BOS

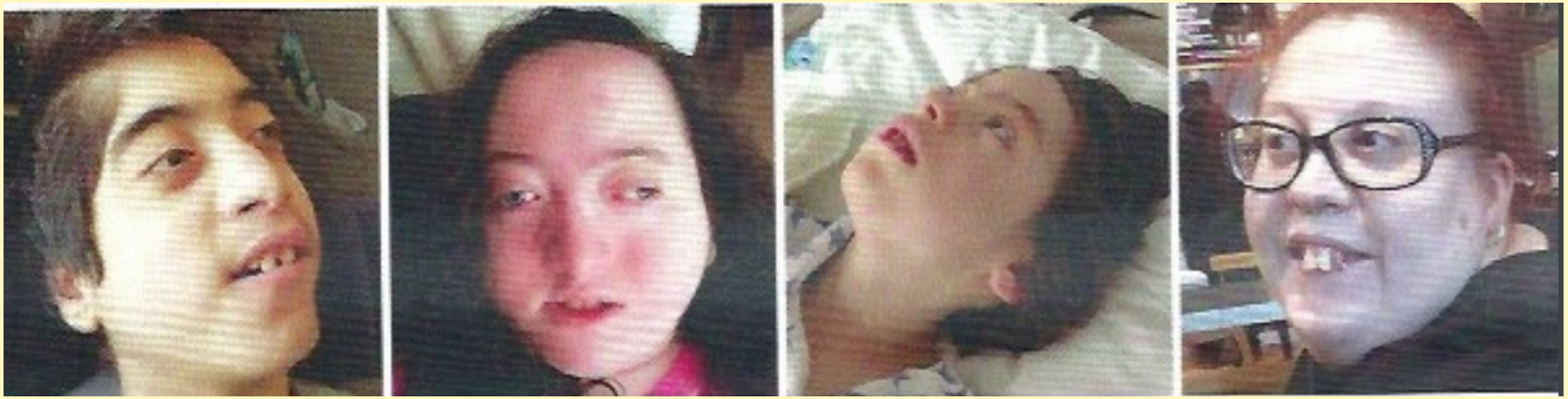
BOS Patients



BOS Patients

Get Involved & Learn More

- For more information on BOS and how you can get involved please visit: www.bos-foundation.org
- Other resources please visit: www.bohring-opitz.org and www.arrefoundation.org



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