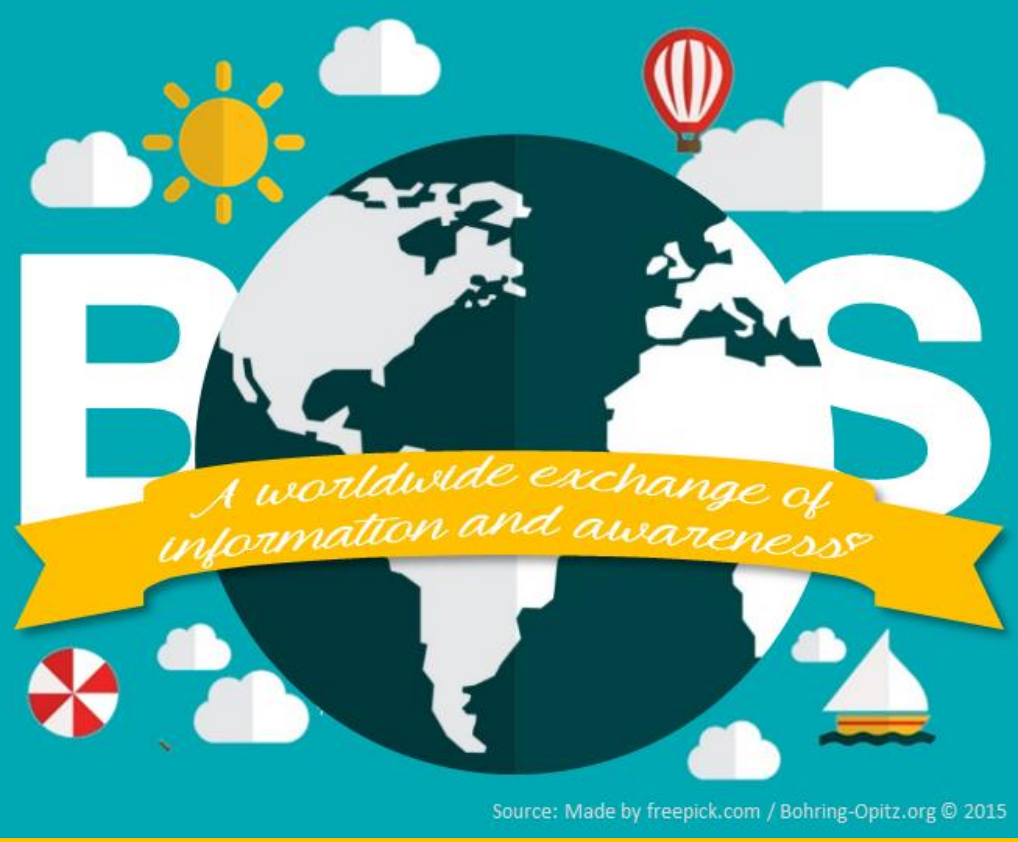

Source: Made by freepick.com / Bohring-Opitz.org © 2015

What is Bohring-Opitz Syndrome?

Bohring-Opitz Syndrome (BOS) is a life limiting rare genetic condition with a high infant mortality.

less than 60 children worldwide

presented in medical studies

1999 discovered

first by Axel Bohring

2011 heterogeneity

one **cause** is found
by Alexander Hoischen

2015 first study clinical management

by Bianca Russell

Clinical diagnostic

BOS-posture
distinctive facial features
trigonocephaly and microcephaly
birthmark on forehead
cleft lip/palate
prominent eyes
hypotonia
brain abnormalities
and more …

novo mutation in ASXL1 gene

Chromosome 20

p-arm ASXL1 q-arm

Symptoms

different in expression and seriousness
failure to thrive
severe feeding issues
recurrent respiratory infections
myopia
seizures
Wilms tumor
severe disability
and more …

Heredity

BOS occurs mostly sporadic
autosomal recessive inheritance has also been reported

What to expect?

Children with BOS are interactive, curious and happy

They need intensive and special care
good communication
for optimal development
and living conditions

as well as further research
and international standard of care

© 2015 Created by Bohring-Opitz Syndrome - A worldwide exchange of information and awareness

Please feel free to visit bohring-opitz.org for further information • Bohring-Opitz Syndrome Awareness Day April 6th

Source: Russell, B. et al. 2015: Clinical management of patients with ASXL1 mutations and Bohring-Opitz syndrome, emphasizing the need for Wilms tumor surveillance. Am J Med Genet Part A 9999A:1-10. Hoischen, A. et al. 2011: De novo nonsense mutations in ASXL1 cause Bohring-Opitz syndrome, Nature Genet. 43: 729-731. OMIM http://www.omim.org/entry/605039