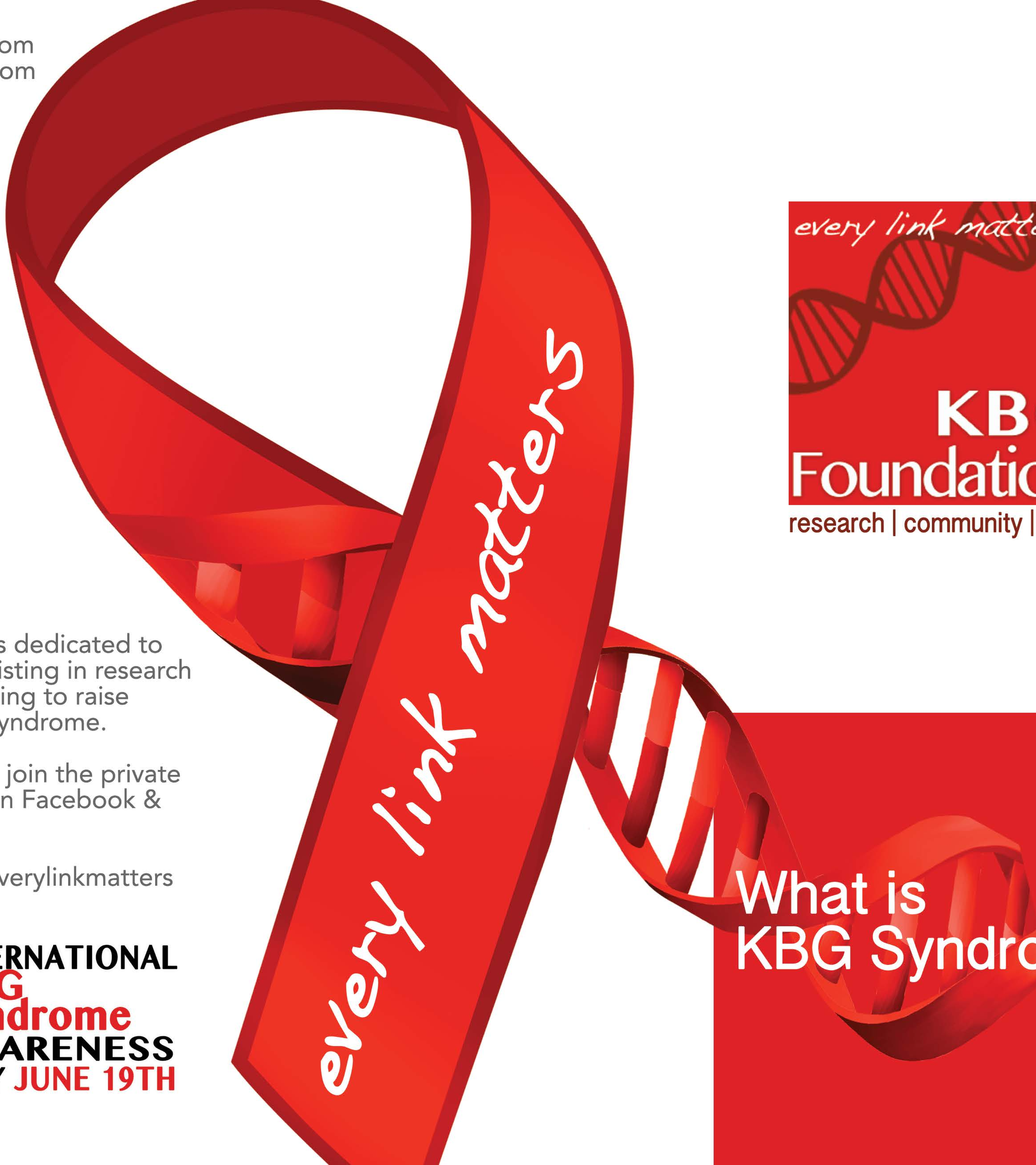
www.kbgfoundation.com
info@kbgfoundation.com
(801) 566-5949



every link matters

KBG Foundation

research | community | cure

The KBG Foundation is dedicated to providing support, assisting in research programs and advocating to raise awareness about the syndrome.

To stay up to date and join the private community follow us on Facebook & Twitter.

@KBGFdn #kbgfdn #everylinkmatters

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INTERNATIONAL KBG Syndrome AWARENESS DAY JUNE 19TH

What is KBG Syndrome?

What is KBG Syndrome?

KBG Syndrome is caused by a mutation in the ANKRD11 gene at location 16q24.3 (maybe more) which creates a shortening of a scaffolding protein in that region.

This short protein may be completely ineffectual or may be somewhat functioning which could contribute to the variety and severity of the symptoms.

Appearance

Nearly all patients present with large upper front teeth, bushy eyebrows and triangular faces. The 'KBG appearance' is quite distinctive and is usually the first noticeable trait along with developmental delay and other bone anomalies.

Incidence

New research suggests that less than 200 indivuduals worldwide have been diagnosed with KBG Syndrome. For no known reason, males seem to be more affected than females.

Prognosis

KBG has an excellent prognosis with no known impact on longevity.

TREATMENT

There is no singular treatment for KBG Syndrome but is determined on a case by case basis as symptoms arise.

Common Traits/Symptoms

Facial dysmorphisms
Autistic Characteristics
Abnormal hair implantation
(low in front or back of skull)
Brachy-clinodactylous 5th finger
(short/curved pinky)
Macrodontia (large teeth)with oligodontia
(absence of more than 6 teeth)
Abnormal EEG with or without seizures
Cognitive deficits/psychomotor delay
Anteverted nostrils (upturned nose)
Short femoral necks/hip dysplasia
Cutaneous syndactyly, toes II/III
Palatal defects (including uvula)
Webbed/short neck
Mild synophrys
Brachycephaly/turricephaly
Sternum abnormalities
Cryptorchidism
Abnormal ribs/vertebrae
Epicanthal folds
Delayed bone age
Abnormal spine curvature
Ptosis
Prominent/high nasal bridge
Long philtrum
Short hand tubular bones
Hearing loss
Wide eyebrows
Wormian bones in skull
Thin upper lip
Prominent/anteverted ears
Strabismus
Congenital heart defects

1975

The KBG Phenotype is defined by Herrmann,Pallister, Tiddy, Opitz
PMID: 1218237

2004

Genetic inheritanceconfirmed
PMID: 21782149

2011

ANKRD11 genetic mutation identified
PMID: 21782149

2012

16q24.3 Gene location reported
PMID: 22307766

2014

C - terminus leads to KBG mutation
PMID: 25413698

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2015