

Bohring-Opitz-Syndrome



Symptoms (all participated children)

Child 1	Child 2	Child 3	Child 4	Child 5	Child 17	Child 18	Child 22	Child 25	Child 27	Child 29
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Clinical Symptoms

Feeding Difficulties		1	1	1	1	first year	1	mild at first	1	1	1
Failure to thrive			1	1	1	1		1		1	1
Intrauterine growth restriction (IUGR)	1	1				1		1		1	1
Severe/profound learning difficulties	1	1	1	1	1	1	1	1	1	Presumed	1
Recurrent infections				1		1	1		1	1	
Absences		1				1			1		
Seizures						1	1	1	1		
Arrhythmias (Irregular heartbeat)										1	
Apneas (suspension of external breathing)		1		1	1		1	1	1		
Bradycardia (slow heartrate)				1							

Craniofacial Symptoms

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Microcephaly (Abnormally small head)			1			1		1			
Trigonocephaly (premature fusion of the metopic suture)		1				1	1		1	1	
Micro/retrognathia (abnormal posterior positioning of the maxilla or mandible)				1	1	1			1		
Flammeaus nevus (Birth mark)		1		1	1	1	1	1	1	1	1
Prominent eyes	1	1	1	1	1	1	1		1	1	1
Abnormal palate	1	1	1	1	1	1	1	1	1	1	1
Hypertelorism (abnormally large distance between the eyes) (Proptosis is a condition resulting in forward displacement and entrapment of the eye from behind by the eyelids)	1		1			1			1		1

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Upslanting palpebral fissures (the elliptic space between the medial and lateral canthi of the two open eye lids)		1		1			1			1	
Epicanthal folds (The skin of the upper eyelid, from the nose to the inner side of the eyebrow, that covers the inner corner (canthus) of the eye)							1				
Broad alveolar ridge (one of the two jaw ridges either on the roof of the mouth between the upper teeth and the hard palate or on the bottom of the mouth behind the lower teeth)											
Cleft/notch lip	1								1		
Cleft palate	1					within the limits		high pallette	1		
Buccal frenulae (skin connecting the cheek to the gums)		1	1	1		1	1		1		
Depressed nasal bridge	1		1	1			1				
Anterverted nares (Nasal openings are tipped upwards)	1			1							
Low-set posteriorly rotated ears	1	1	1	1	1	1	1		1	1	1
Retrognathia (the jaw is set back)			1	1	1				1	1	
Short philtrum (Space between upper lip and tip of nose)						1					

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Ophthalmic – Eye symptoms

Strabismus (squint, cross eyed)	1	1					1	1	1		
Anterior chamber abnormalities (the fluid-filled space inside the eye between the iris and the cornea's innermost surface)											
Myopia (short-sighted)		1		1	1	1	1	1			1
Retinal/optic nerve abnormalities	1						1		1		
Absence of tears	1	1		1	1		1			1	1
Glaucoma (a group of eye conditions in which the optic nerve is damaged due to changes in eye pressure)									1		
High intraocular pressures initially (causes damage to the optic nerve, which can lead to glaucoma)									1		
Thick Cornea (corneal thickness is an important factor in accurately diagnosing eye pressure)									1		
Excavatie									1		

Hair/Skin

Low hairline		1	1	1	1	1			1		1
Hypertrichosis (abnormal hair density and length)	1	1	1	1	1	1	1		1		1
Hirsutisme dorsal (hairy back)	1	1			1	1			1	1	
Synophrys (abundant hair between the eyebrows)	1							1			1

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Neurological/Skeletal

BOS Posture	1	1	1	1	1	1		1	1	1
Scoliosis (Curvature of the spine)		1				1		1		
Fixed constrictures of the upper limbs					1		1			1
Absence of distale interphalangeale crease from fingers								1		1
Deviation from fingers (a hand deformity in which the swelling of the metacarpophalangeal joints (the big knuckles at the base of the fingers)	1							1		1
Dropping/clenched hands (BOS posture)	1	1	1	1	1			1	1	1
Rhizomele humerus (shortening of the humerus and femur)					1			1		
Bilateral hip dysplasia/luxation (dislocation of the hips)					only luxation			1		
Thoracic kyphosis (also called roundback or Kelso's hunchback), is a condition of over-curvedness of the thoracic vertebrae (upper back)					1		1	1		
Platyspondilie thoracal (Narrowing of the intervertebral disc spaces in the upper lumbar and lower thoracic spine and clefting of the posterior end plates at L2-4)								1		

Congenital dislocations

Hypertonia (a condition of excessive tone of the skeletal muscles; increased resistance of muscle to passive stretching)	1	1							1	1
Hypotonia (low muscle tone (the amount of tension or resistance to stretch in a muscle), often involving reduced muscle strength)		1	1	1	1	1	1	1	1	1

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Brain

Brain abnormalities		1	1	1				1	1	1
Cavum septum pellucidum (this is a problem with the 'soft spot' - as far as I can translate from the medical terminology)								1		
Hypoplasie partiele agenesie posteriore corpus callosum (The Corpus Callosum is the central region of the human brain consists of nerve tissue which convey "messages" between the two hemispheres. This part is only partially formed or not formed at all)	Partial	Partial	1	1				1		
Spina bifida occulta (The mildest form of spina bifida, spina bifida occulta describes a group of neural tube birth defects that can affect the spinal column.)								1		

Urology

Recurrent Urinary tract infections				1				1		
Thickened bladder wall				1				1		
Infravesical obstruction (bladder outlet obstruction)								1		
residue								1		
Gastroenterology										
Emesis (vomiting)	1	1	1	1	1			1		1
Fundoplication (a surgical procedure to treat gastroesophageal reflux disease)	1						1			
Gastrostomy	1	1		1	1		1	1		
NG Tube	1	1	1				1	1		1
Constipation		1		1	1	1	1	1		
Eating solid food		1			pureed	1	1			
Gallstones								1		

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Other

Genital abnormalities						1			1	
Renal abnormalities						1			1	
Cardiac abnormalities									1	1
Wilms Tumours										
Short stature		1	1							1
Winds hands and straightens legs when showing emotion	1	1		1	1	1				1
Tonsilectomy						1				
Able to Communicate Verbally (please put in age they started)										
Able to walk - (please put in age they started)										
Hearing Impairment		Slight						poor		
Thalassaemia (a form of anaemia)										
Polythelia (the presence of supernumerary nipples)	1							1		
Inguinal hernia								1		
Precocious puberty							Bone age of 13.9 at age 10			
Turned in ankles										Slight
Quick growing finger and toe nails				1	1					1
Silent aspiration (Silent aspiration occurs in people that have swallowing problems, which is known as dysphagia. Aspiration is when food, liquids, or stomach contents are swallowed poorly, and go into the lungs.)			1		1					1

outcome	1 year	1 year	1 year	3 years	2 years	6 years	7 years	10 years	5 years, 8	20 days	14 months
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