



Nicolaides-Baraitser Syndrome

Synonym: NCBRS

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Created: October 15, 2015.

Summary

Clinical characteristics

Nicolaides-Baraitser syndrome (NCBRS) is characterized by sparse scalp hair, prominence of the interphalangeal joints and distal phalanges due to decreased subcutaneous fat, characteristic coarse facial features, microcephaly, seizures, and developmental delay / intellectual disability. Seizures are of various types and can be difficult to manage. Developmental delay / intellectual disability (ID) is severe in nearly a half, moderate in a third, and mild in the remainder. Nearly a third never develop speech or language skills.

Diagnosis/testing

The diagnosis of NCBRS is established in a proband with suggestive clinical findings and the identification of a heterozygous *SMARCA2* pathogenic variant by molecular genetic testing.

Management

Treatment of manifestations: Anti-seizure medication (ASM) for seizures under the care of a neurologist or epileptologist; occupational, physical, and/or speech therapy; routine management of refractive errors and hearing loss.

Surveillance: At least yearly evaluation by a neurologist to assess for and/or manage seizures; yearly evaluation by a developmental pediatrician to assess developmental progress and therapeutic and educational interventions; regular follow up of ophthalmologic and/or audiologic abnormalities.

Genetic counseling

NCBRS is inherited in an autosomal dominant manner. All affected individuals reported to date have NCBRS as the result of a *de novo* *SMARCA2* pathogenic variant. Although no affected sibs have been reported, the risk to sibs is presumed to be greater than in the general population because of the possibility of germline mosaicism in a parent. If the *SMARCA2* pathogenic variant has been identified in an affected family member, prenatal testing for pregnancies at theoretic increased risk is possible.

Diagnosis

Consensus clinical diagnostic criteria for Nicolaides-Baraitser syndrome (NCBRS) have not been established.

Suggestive Findings

Nicolaides-Baraitser syndrome (NCBRS) **should be suspected** in individuals with the following clinical and radiographic findings.

Clinical findings (from most common to least common)

- Developmental delay / intellectual disability: 50% severe, 30% moderate, 20% mild
- Sparse scalp hair
- Prominence of the inter-phalangeal joints and distal phalanges secondary to poor subcutaneous fat distribution (See Figure 1 and Figure 2.)
- Characteristic facial features (anteverted nares, long philtrum, wide mouth, thin upper lip vermilion and thick lower lip vermilion). The facial features can be subtle in the newborn period and early childhood, but coarsening of the face and increased skin wrinkling occurs over time (see Figure 3).
- Microcephaly
- Seizures

Radiographic findings

- The hands may show cone-shaped epiphyses, metaphyseal flaring of the phalanges, and shortening of the phalanges, metacarpals, and/or metatarsals (especially of the 4th and 5th rays).
- Bone age can be variable: 16 children with delayed bone age and two with advanced bone age have been reported.
- Platyspondyly, flat intervertebral discs, small pelvis, pubic bone hypoplasia, small femoral heads, and short femoral necks have been reported.

Establishing the Diagnosis

The diagnosis of Nicolaides-Baraitser syndrome **is established** in a proband with suggestive clinical findings and the identification of a heterozygous *SMARCA2* pathogenic variant by molecular genetic testing (see Table 1).

Molecular testing approaches can include **single-gene testing**, use of a **multigene panel**, and **more comprehensive genomic testing**.

- **Single-gene testing.** Sequence analysis of *SMARCA2* is performed first, followed by gene-targeted deletion/duplication analysis if no pathogenic variant is found.
- **A multigene panel** that includes *SMARCA2* and other genes of interest (see Differential Diagnosis) may also be considered. Note: (1) The genes included in the panel and the diagnostic sensitivity of the testing used for each gene vary by laboratory and are likely to change over time. (2) Some multigene panels may include genes not associated with the condition discussed in this *GeneReview*; thus, clinicians need to determine which multigene panel is most likely to identify the genetic cause of the condition while limiting identification of variants of uncertain significance and pathogenic variants in genes that do not explain the underlying phenotype. (3) In some laboratories, panel options may include a custom laboratory-designed panel and/or custom phenotype-focused exome analysis that includes genes specified by the clinician. (4) Methods used in a panel may include sequence analysis, deletion/duplication analysis, and/or other non-sequencing-based tests.

For an introduction to multigene panels click [here](#). More detailed information for clinicians ordering genetic tests can be found [here](#).



Figure 1. Prominent interphalangeal joints with reduced fat deposition in the digits



Figure 2. Moderately prominent interphalangeal joints

- **More comprehensive genomic testing** may be considered if single-gene testing (and/or use of a multigene panel that includes *SMARCA2*) has not confirmed a diagnosis in an individual with features of Nicolaidis-Baraitser syndrome. Such testing may include **exome sequencing** and **genome sequencing**.

For an introduction to comprehensive genomic testing click [here](#). More detailed information for clinicians ordering genomic testing can be found [here](#).



Figure 3. Coarse facies with sparse scalp hair, thin upper lip vermilion, and thick lower lip vermilion

Table 1. Molecular Genetic Testing Used in Nicolaides-Baraitser Syndrome

Gene ¹	Method	Proportion of Proband with a Pathogenic Variant ² Detectable by Method
SMARCA2	Sequence analysis ³	36/44 ⁴
	Gene-targeted deletion/duplication analysis ⁵	2/61 ⁶
Unknown ⁷	NA	Unknown

1. See Table A. Genes and Databases for chromosome locus and protein.

2. See Molecular Genetics for information on allelic variants detected in this gene.

3. Sequence analysis detects variants that are benign, likely benign, of uncertain significance, likely pathogenic, or pathogenic. Variants may include small intragenic deletions/insertions and missense, nonsense, and splice site variants; typically, exon or whole-gene deletions/duplications are not detected. For issues to consider in interpretation of sequence analysis results, click [here](#).

4. Van Houdt et al [2012]

5. Gene-targeted deletion/duplication analysis detects intragenic deletions or duplications. Methods used may include a range of techniques such as quantitative PCR, long-range PCR, multiplex ligation-dependent probe amplification (MLPA), and a gene-targeted microarray designed to detect single-exon deletions or duplications.

6. Tsurusaki et al [2012], Wolff et al [2012]

7. Locus heterogeneity involving other genes within the SWI/SNF complex may be causative in individuals who have findings suggestive of NCBRS but no detectable SMARCA2 pathogenic variant (see Molecular Genetics). Of the 44 individuals evaluated by Van Houdt et al [2012], the clinical diagnosis of NCBRS was only felt to be certain in 37. Thirty-four of the 37 were found to have a SMARCA2 pathogenic variant; three of the 37 were found to have an ARID1B pathogenic variant and were reclassified as having Coffin-Siris syndrome (see Differential Diagnosis).

Clinical Characteristics

Clinical Description

The most common findings in 61 individuals (35 male and 26 female) with Nicolaidis-Baraitser syndrome (NCBRS) and a *SMARCA2* pathogenic variant are summarized in Table 2 [Sousa et al 2014].

Table 2. Summary of the Most Common Clinical Findings in 61 Individuals with Nicolaidis-Baraitser Syndrome

Finding	% of Affected Individuals
Intellectual disability	100%
Sparse hair	97%
Prominent interphalangeal joints	85%
Coarse facies	77%
Microcephaly	65%
Seizures	64%

From Sousa et al [2014]

Ectoderm. Scalp hair is usually sparse at birth and becomes increasingly so with age, particularly in the second decade of life. In some, the sparseness improves with time.

Skin pigmentation appears to be reduced, although affected individuals do not exhibit true cutaneous albinism.

Poor subcutaneous fat distribution leads to prominent veins; interphalangeal joints are prominent. The distal phalanges widen with age, becoming oval shaped and broad. Increasing space between the first and second toes can occur over time.

Delayed tooth eruption is common, often requiring surgical extraction of primary dentition to allow secondary dentition to migrate into place.

Seizures can be difficult to manage, requiring multiple anti-seizure medications to achieve reasonable control. The mean and median age of onset of seizures is 24 months and 18 months, respectively. The range is birth to 14 years. Various seizure types have been reported. Regression or lack of developmental progress has been noted with the onset of seizures.

Developmental delay / intellectual disability. Nearly half of affected individuals experience severe developmental delay / intellectual disability (ID) with particular delays in speech and language development. A third exhibit moderate intellectual disability and the remainder have mild ID.

Nearly a third never develop speech or language skills.

The mean age for walking independently is 21 months (range: 10 months - 5 years).

Although psychomotor regression is not typical, the high incidence of seizures that progressively worsen has been associated with loss of speech.

Behavior problems have been reported in at least 19 individuals with some displaying autistic-like features (such as perseveration and hyperacusis). However, a formal clinical diagnosis of autism has not been made in any affected individual reported in the literature.

Growth. About half of affected individuals (24/46) have low birth weight and the same proportion experience short stature. None was above the 50th centile for height.

Microcephaly tends to be acquired, being noted in almost one third of infants at birth (7/30) and in two thirds (34/52) at follow up.

Other findings

- Cryptorchidism is seen in most males.
- Hearing loss has been noted in 4/59 affected individuals. The nature of the hearing loss and age of onset have not been reported.
- Visual deficits including myopia and astigmatism have been seen in ten and four affected individuals, respectively.
- Congenital heart defects (e.g., atrial septal defect, stenosis of the pulmonary artery, coarctation, patent ductus arteriosus, and double aortic arch) have been reported in six affected persons.

Genotype-Phenotype Correlations

No clear genotype-phenotype correlations have been noted; however, all individuals with a pathogenic variant within the C-terminal helicase region of the ATPase domain have severe intellectual disability and epilepsy – a frequency higher than that in individuals with pathogenic variants in other parts of the gene.

Penetrance

Data are insufficient to determine penetrance. All 61 affected individuals published to date had a *de novo* pathogenic variant, suggesting that penetrance is likely complete [Sousa et al 2014].

Nomenclature

Morin et al [2003] proposed the name Nicolaides-Baraitser syndrome after the authors of the 1993 article in which the earliest known person with NCBRS was described.

Prevalence

The prevalence of NCBRS is not known, but is estimated to be extremely low. Fewer than 100 affected individuals have been described in the literature.

Genetically Related (Allelic) Disorders

No phenotypes other than those discussed in this *GeneReview* are known to be associated with pathogenic variants in *SMARCA2*.

Sporadic tumors occurring as single tumors in the absence of any other findings of this syndrome frequently harbor somatic variants in *SMARCA2* that are **not** present in the germline; thus, predisposition to these tumors is not heritable.

Differential Diagnosis

Coffin-Siris syndrome (CSS) is characterized by coarse facial features, hypertrichosis, absent or hypoplastic fifth nails (fingers and/or toes), agenesis of the corpus callosum, and developmental delay / intellectual disability. Although the facial features can be similar to those of Nicolaides-Baraitser syndrome (NCBRS), the digital findings are particularly helpful in differentiating the two disorders, as individuals with NCBRS uniquely have prominent interphalangeal joints and do not have hypoplasia of the fifth digits. Additionally, individuals with NCBRS have sparse scalp hair rather than the hypertrichosis seen in CSS.

CSS is caused by a heterozygous pathogenic variant in one of the following genes: *ARID1A*, *ARID1B* (see [ARID1B-Related Disorder](#)), *SMARCA4*, *SMARCB1*, or *SMARCE1*. In the majority of affected individuals the pathogenic variant is *de novo*.

The phenotypic overlap between CSS and NCBRS is likely due to the fact that both conditions are caused by pathogenic variants in genes involved in the SWI/SNF complex.

Of note, Tsurusaki et al [2012] reported an individual with a clinical diagnosis of Coffin-Siris syndrome (CSS) who had a 55-kb interstitial deletion of *SMARCA2* and whose clinical findings were more consistent with a diagnosis of NCBRS (prominent interphalangeal joints) than CSS (fifth fingernails were not absent).

Williams syndrome (WS) is characterized by typical facial features (long philtrum, thick vermilion of the upper and lower lips, and full cheeks), supraaortic stenosis, joint laxity, growth deficiency, hypercalcemia, and intellectual disability. However, their cognitive profile shows a relative strength in language. The facial features, cardiac lesions, and cognitive profile are sufficient to differentiate WS from NCBRS. WS is caused by microdeletion of the Williams-Beuren syndrome critical region (WBSCR). The majority of affected individuals have a *de novo* microdeletion.

Cornelia de Lange syndrome (CdLS) is characterized by typical facial features (long philtrum, arched eyebrows, synophrys, and short nose), growth deficiency, and limb anomalies. The limb findings primarily include hypoplasia of the digits and can be severe. The facial features and limb changes are sufficient to differentiate CdLS from NCBRS. CdLS is caused by mutation of one of the following genes: *NIPBL*, *HDAC8*, *RAD21*, *SMC1A*, or *SMC3*. In the majority of affected individuals the pathogenic variant is *de novo*.

2q37 microdeletion syndrome is characterized by typical facial features (round face, frontal bossing, arched eyebrows, upslanted palpebral fissures, epicanthal folds, underdeveloped nasal alae, and thin vermilion of the upper lip), brachymetaphalangy of digits 3-4, short stature, hypotonia, and intellectual disability. Seizures have been reported in individuals with the 2q37 deletion syndrome, but are not usually difficult to manage. In most affected individuals microdeletion is *de novo*.

Biotinidase deficiency is primarily associated with seizures, hypotonia, and cutaneous abnormalities including hair loss. Facial features are typically normal. Biotinidase deficiency is caused by biallelic pathogenic variants in *BTD* and is inherited in an autosomal recessive manner.

Management

Evaluations Following Initial Diagnosis

To establish the extent of disease and needs in an individual diagnosed with Nicolaidis-Baraitser syndrome (NCBRS), the following evaluations are recommended:

- EEG to determine if seizures are present, with consideration of referral to an epileptologist for seizure management
- Neurologic and/or developmental examination to assess developmental milestones and identify neurologic findings or deficits
- Evaluation for occupational, speech, or physical therapy as needed
- Dental evaluation if dental eruption is delayed
- Audiology evaluation with auditory brain stem response testing and otoacoustic emission testing to assess for hearing loss
- Ophthalmologic examination, including visual acuity and dilated fundus examination
- Evaluation for cryptorchidism in males
- Consultation with a clinical geneticist and/or genetic counselor

Treatment of Manifestations

The following are appropriate:

- Anti-seizure medication (ASM) for seizures under the care of a neurologist or epileptologist
- Occupational, physical, and/or speech therapy to optimize developmental outcomes
- Spectacles as needed to correct refractive errors
- Hearing aids as needed
- Orchiopexy as needed

Surveillance

Surveillance includes the following:

- At least yearly evaluation by a neurologist to assess for and/or manage seizures
- Yearly evaluation by a developmental pediatrician to assess developmental progress and therapeutic and educational interventions
- Regular follow up of ophthalmologic and/or audiologic abnormalities

Evaluation of Relatives at Risk

See Genetic Counseling for issues related to testing of at-risk relatives for genetic counseling purposes.

Therapies Under Investigation

Search [ClinicalTrials.gov](https://clinicaltrials.gov) in the US and [EU Clinical Trials Register](https://clinicaltrialsregister.eu) in Europe for access to information on clinical studies for a wide range of diseases and conditions. Note: There may not be clinical trials for this disorder.

Genetic Counseling

Genetic counseling is the process of providing individuals and families with information on the nature, mode(s) of inheritance, and implications of genetic disorders to help them make informed medical and personal decisions. The following section deals with genetic risk assessment and the use of family history and genetic testing to clarify genetic status for family members; it is not meant to address all personal, cultural, or ethical issues that may arise or to substitute for consultation with a genetics professional. —ED.

Mode of Inheritance

Nicolaides-Baraitser syndrome (NCBRS) is inherited in an autosomal dominant manner. All affected individuals reported to date have NCBRS as the result of a *de novo* pathogenic variant.

Risk to Family Members

Parents of a proband

- All probands with NCBRS reported to date have the disorder as a result of a *de novo* *SMARCA2* pathogenic variant.
- Recommendations for the evaluation of parents of a proband with an apparent *de novo* pathogenic variant include testing of the parents for the *SMARCA2* pathogenic variant identified in the proband. However, the high penetrance of the disorder suggests that it would be highly unlikely to discover a pathogenic variant in an apparently asymptomatic individual.

Sibs of a proband. To date, all affected individuals have had a *de novo* *SMARCA2* pathogenic variant, suggesting a low risk to sibs. However, because of the theoretic possibility of parental germline mosaicism, the empiric recurrence risk to sibs is approximately 1%.

Offspring of a proband. Each child of an individual with NCBRS has a 50% chance of inheriting the *SMARCA2* pathogenic variant. To date, individuals with NCBRS have not been known to reproduce.

Other family members. The risk to other family members appears to be low given that all probands with NCBRS reported to date have the disorder as a result of a *de novo* *SMARCA2* pathogenic variant.

Related Genetic Counseling Issues

Family planning

- The optimal time for determination of genetic risk and discussion of the availability of prenatal/preimplantation genetic testing is before pregnancy.
- It is appropriate to offer genetic counseling (including discussion of potential risks to offspring and reproductive options) to parents of affected individuals.

DNA banking. Because it is likely that testing methodology and our understanding of genes, pathogenic mechanisms, and diseases will improve in the future, consideration should be given to banking DNA from probands in whom a molecular diagnosis has not been confirmed (i.e., the causative genetic mechanism is unknown).

Prenatal Testing and Preimplantation Genetic Testing

Once the *SMARCA2* pathogenic variant has been identified in an affected family member, prenatal testing for a pregnancy at increased risk and preimplantation genetic testing for NCBRS are possible.

Resources

GeneReviews staff has selected the following disease-specific and/or umbrella support organizations and/or registries for the benefit of individuals with this disorder and their families. GeneReviews is not responsible for the information provided by other organizations. For information on selection criteria, click [here](#).

- **Genetic and Rare Diseases Information Center (GARD)**
[Nicolaidis-Baraitser syndrome](#)
- **NCBRS Worldwide Foundation**
Email: contactus@ncbrs.com
ncbrs-worldwide-foundation.weebly.com

Molecular Genetics

Information in the Molecular Genetics and OMIM tables may differ from that elsewhere in the GeneReview: tables may contain more recent information. —ED.

Table A. Nicolaidis-Baraitser Syndrome: Genes and Databases

Gene	Chromosome Locus	Protein	Locus-Specific Databases	HGMD	ClinVar

Table A. continued from previous page.

SMARCA2	9p24.3	Probable global transcription activator SNF2L2	SMARCA2 @ LOVD	SMARCA2	SMARCA2
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Data are compiled from the following standard references: gene from [HGNC](#); chromosome locus from [OMIM](#); protein from [UniProt](#). For a description of databases (Locus Specific, HGMD, ClinVar) to which links are provided, click [here](#).

Table B. OMIM Entries for Nicolaides-Baraitser Syndrome ([View All in OMIM](#))

600014	SWI/SNF-RELATED, MATRIX-ASSOCIATED, ACTIN-DEPENDENT REGULATOR OF CHROMATIN, SUBFAMILY A, MEMBER 2; SMARCA2
601358	NICOLAIDES-BARAITSER SYNDROME; NCBRS

Molecular Pathogenesis

The SWI/SNF family of ATPase-dependent chromatin remodelers is essential for the regulation of gene expression, differentiation, and development. *SMARCA2* encodes SMARCA2 which is within the family of helicase-related proteins that share an ATPase domain critical for the coupling of ATP hydrolysis with DNA binding, which in turn results in chromatin remodeling. SMARCA2 is a subunit of the BRG1-associated factors (BAF) complex, the human analog of the SWI/SNF complex.

Mutation of other genes within the SWI/SNF complex has been suggested as the cause in individuals who have findings suggestive of Nicolaides-Baraitser syndrome (NCBRS) but no detectable *SMARCA2* pathogenic variant [Van Houdt et al 2012].

Gene structure. *SMARCA2* comprises 34 exons and makes a transcript of 5879 base pairs. Alternatively spliced transcript variants encoding different isoforms have been found for this gene, which contains a CAG trinucleotide repeat length polymorphism. For a detailed summary of gene and protein information, see Table A, **Gene**.

Pathogenic variants. In a review of 61 individuals with NCBRS, 59 had pathogenic missense variants and two had in-frame deletions [Sousa et al 2014]. All pathogenic variants clustered within the ATPase *SMARCA2* domain (exons 15-25). Based on this finding, it is believed that pathogenic variants in *SMARCA2* most likely cause a dominant-negative or gain-of-function effect.

Normal gene product. *SMARCA2* codes for a 1,590-amino acid catalytic subunit of the SWI/SNF family of ATPase-dependent chromatin remodelers. The catalytic subunit contains a helicase-like ATPase domain that plays a role in the regulation of gene expression through DNA binding that is coupled with ATP binding and ATP hydrolysis.

Abnormal gene product. Pathogenic variants in *SMARCA2* may result in aberrant chromatin remodeling, causing downstream dysregulation of further genes and resulting in the NCBRS phenotype. It is suspected that mutated *SMARCA2* is able to incorporate into the SWI/SNF complex that then binds to downstream targets within the genome. In this scenario, remodeling of the nucleosome structure in order to effect gene expression would not be able to occur normally, due to a dominant-negative or gain-of-function effect.

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Chapter Notes

Author Notes

The author of this review studies the clinical features and molecular basis of the Nicolaidis-Baraitser syndrome and is a member of the Nicolaidis-Baraitser Syndrome International Consortium.

Acknowledgments

The author wishes to thank Sérgio B Sousa for his collaboration.

Revision History

- 15 October 2015 (me) Review posted live
- 23 July 2015 (oa) Original submission

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