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Majeed Syndrome – RETIRED CHAPTER, FOR HISTORICAL REFERENCE ONLY

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Summary

NOTE: THIS PUBLICATION HAS BEEN RETIRED. THIS ARCHIVAL VERSION IS FOR HISTORICAL REFERENCE ONLY, AND THE INFORMATION MAY BE OUT OF DATE.

Clinical characteristics

Majeed syndrome is characterized by:

- Chronic recurrent multifocal osteomyelitis (CRMO) that is of early onset with a lifelong course; and
- Congenital dyserythropoietic anemia (CDA) that presents as hypochromic, microcytic anemia during the first year of life and ranges from mild to transfusion dependent.

Some individuals also develop a transient inflammatory dermatosis, often manifesting as Sweet syndrome (neutrophilic skin infiltration).

Diagnosis/testing

The diagnosis is based on clinical findings and molecular genetic testing of *LPIN2*, the only gene in which pathogenic variants are known to cause Majeed syndrome.

Management

Treatment of manifestations: CRMO is treated with nonsteroidal anti-inflammatory drugs (NSAIDs) and physical therapy to avoid disuse atrophy of muscles and contractures. If CRMO does not respond to NSAIDs, corticosteroids can be used short term to control CRMO and skin manifestations; however, the complications of long-term use of corticosteroids limit their use in children. Two affected children had resolution of their bone inflammation when treated with an IL-1 inhibitor. Physical therapy should be employed to avoid disuse atrophy of muscles or contractures. CDA is treated with red blood cell transfusion if indicated.

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Surveillance: Routine complete blood count (CBC) to determine if red blood cell transfusion is necessary.

Agents/circumstances to avoid: Prolonged bed rest.

Genetic counseling

Majeed syndrome is inherited in an autosomal recessive manner. At conception, each sib of an affected individual has a 25% chance of being affected, a 50% chance of being an asymptomatic carrier, and a 25% chance of being unaffected and not a carrier. Carrier testing for at-risk relatives is possible if the pathogenic variants in the family are known. If the pathogenic variants in the family have been identified, prenatal testing for pregnancies at increased risk is possible through laboratories offering either testing for the gene of interest or custom testing.

Diagnosis

Clinical Diagnosis

The diagnosis of Majeed syndrome is based on the following findings [El-Shanti & Ferguson 2007, Ferguson & El-Shanti 2007]:

- **Chronic recurrent multifocal osteomyelitis (CRMO)** that is of early onset with a lifelong course
  - Skeletal radiographs show irregular osteolytic (radiolucent) lesions with surrounding sclerosis, usually in the metaphyses of long bones. Hyperostosis may be present in clavicular lesions.
  - Tc-99 or Ga-67 skeletal scan shows increased uptake at the inflammatory lesions; silent (asymptomatic) lesions may be identified.
  - MRI may be required to confirm the diagnosis, follow up lesions, or guide biopsy of an active bone lesion. Active bone lesions show increased signal intensity on T_2-weighted and STIR images and decreased signal intensity on T_1-weighted images. MRI is the imaging modality of choice for vertebral body lesions.
  - Biopsy of a bone lesion shows nonspecific inflammatory changes with granulocytic infiltration.
- **Congenital dyserythropoietic anemia (CDA)**
  - Hypochromic, microcytic anemia manifests during the first year of life and ranges from mild to transfusion dependent.
  - Bone marrow examination shows increased erythropoiesis associated with evidence of dyserythropoiesis including up to 25% of normoblasts that are binucleated and trinucleated. The Ham test is negative.
- **Inflammatory dermatosis** [Majeed et al 2000, Majeed et al 2001, Al-Mosawi et al 2007] that may be transient and is often Sweet syndrome (neutrophilic skin infiltration)
  - Biopsy of pustular skin lesions usually shows intraepidermal collection of neutrophils.

Testing

Laboratory testing is nonspecific:

- The most consistent findings are elevated erythrocyte sedimentation rate [El-Shanti & Ferguson 2007, Ferguson & El-Shanti 2007] and anemia.
- The white blood cell count may or may not be elevated.

Cultures from blood, bone biopsies, and pustular lesions are always negative.

Molecular Genetic Testing

Gene. *LPIN2* is the only gene in which pathogenic variants are known to cause Majeed syndrome.

Table 1. Molecular Genetic Testing Used in Majeed Syndrome

Gene [1]	Method	Allelic Variants Detected [2]	Variant Detection Frequency by Method [3]
LPIN2	Sequence analysis [4] / scanning for pathogenic variants [5]	Sequence variants	See footnote 6

1. See Table A. Genes and Databases for chromosome locus and protein.
2. See Molecular Genetics for information on allelic variants.
3. The ability of the test method used to detect a variant that is present in the indicated gene
4. Sequence analysis detects variants that are benign, likely benign, of uncertain significance, likely pathogenic, or pathogenic. 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.
5. Sequence analysis and scanning of the entire gene can have similar variant detection frequencies; however, variant detection rates for scanning may vary considerably between laboratories depending on the specific protocol used.
6. The variant detection frequency using sequence analysis in individuals with CRMO and microcytic CDA is 100%; the variant detection frequency using scanning is unknown.

Testing Strategy

To confirm/establish the diagnosis in a proband

- Clinical evaluation to identify the three components of Majeed syndrome (may require bone biopsy of affected osteolytic lesion, bone marrow biopsy to document dyserythropoiesis, and skin biopsy to document neutrophilic dermatosis)
- Sequence analysis / scanning of *LPIN2* for pathogenic variants in those who meet clinical diagnostic criteria

Carrier testing for at-risk relatives requires prior identification of the pathogenic variants in the family.

Note: Carriers are heterozygotes for this autosomal recessive disorder and are not at risk of developing the disorder.

Prenatal diagnosis and preimplantation genetic diagnosis (PGD) for at-risk pregnancies require prior identification of the pathogenic variants in the family.

Clinical Characteristics

Clinical Description

Majeed syndrome is characterized by chronic recurrent multifocal osteomyelitis (CRMO), congenital dyserythropoietic anemia (CDA), and inflammatory dermatosis.

Onset of CRMO ranges from age three weeks to no later than age two years. It is characterized by short remissions, one to three exacerbations per month with each remaining for a few days, and a prolonged, probably lifelong, course. Each exacerbation consists of high fever, severe pain, and the appearance of periarticular tender soft tissue swelling, mainly involving large joints and occasionally small joints. The CRMO in Majeed syndrome is often associated with delayed bone age, growth failure, and development of permanent flexion contractures.

CDA usually presents during the first year of life and varies in severity from mild to transfusion dependent.

The inflammatory dermatosis is not a consistent phenotypic component of Majeed syndrome, although this may be a result of its transient nature. Of the affected individuals reported to date, two brothers had Sweet syndrome, their father had psoriasis, and an affected member of another family had cutaneous pustulosis.

Hepatomegaly, neutropenia, and transient cholestatic jaundice may occur during the neonatal period. These findings have no clinical consequences because they are transient. In rare instances, neutropenia may predispose to infections.

Individuals with Majeed syndrome have linear growth retardation with short adult stature.

If left untreated, the quality of life is poor as a result of recurrent pain, chronic anemia, and possible complications of contractures and disuse atrophy of the muscles.

The oldest individual known to have Majeed syndrome has been lost to follow up since he was about 28 years old.

Genotype-Phenotype Correlations

Although the number of individuals reported with Majeed syndrome is too small to study genotype-phenotype correlations, the affected individuals in the family with a frameshift variant [Majeed et al 2001] appear to have a more severe course and complications than families with other classes of pathogenic variants. Conversely, another affected individual with a splice site variant [Al-Mosawi et al 2007] and two affected Turkish brothers with a frameshift variant [Herlin et al 2013], who were all diagnosed and treated early, had a less complicated course. It is unclear whether their milder clinical course is attributable to the earlier detection and treatment.

Penetrance

Penetrance is 100%.

Nomenclature

Majeed syndrome has been known by its components (i.e., CRMO, CDA, and neutrophilic dermatosis).

Prevalence

Majeed syndrome is very rare. The four affected families reported to date are from the Middle East:

- One of Jordanian/Palestinian origin, reported from Kuwait
- One Jordanian, reported from Jordan
- One from Bahrain
- One Turkish family residing in Denmark

Genetically Related (Allelic) Disorders

No phenotypes other than those discussed in this *GeneReview* are known to be associated with mutation of *LPIN2*.

Differential Diagnosis

The clinical diagnosis of Majeed syndrome is straightforward once the complete triad is established, and can be confirmed by identifying any of the various pathogenic variants in *LPIN2*. However, suspicion should be raised in the presence of any component, especially with onset in infancy and early childhood, such as unexplained congenital anemia and multifocal osteomyelitis in infancy.

Because of its recurrent febrile episodic course, Majeed syndrome should also be included in the differential diagnosis of the periodic fever syndromes.

The combination of bone and skin involvement, in particular, is shared by a variety of disorders including the following:

- Synovitis, acne, pustulosis, hyperostosis, and osteitis (SAPHO) syndrome is believed to be the CRMO of adults with skin, bone, and synovial membrane inflammation. The contribution of genetics to the etiology of SAPHO syndrome is unclear. The early onset and CDA of Majeed syndrome distinguish it from SAPHO syndrome.
- Pyogenic arthritis, pyoderma gangrenosum, and acne (PAPA) syndrome is characterized by oligoarticular, corticosteroid-responsive arthritis beginning in childhood. Pyoderma gangrenosum and severe cystic acne begin in adolescence. Pathogenic variants in *PSTPIP1* are causative; inheritance is autosomal dominant. In contrast, Majeed syndrome is associated with CDA and not associated with pyoderma gangrenosum or cystic acne, and inheritance is autosomal recessive.
- Sporadic nonsyndromic CRMO is an autoinflammatory bone disease characterized by bone pain with or without fever with an unpredictable course of exacerbation and spontaneous remissions [El-Shanti & Ferguson 2007, Ferguson & El-Shanti 2007]. It is a sporadic disease. However, some indications suggest that it is a multifactorial disease with both genetic and environmental causes, although most causes have not yet been identified. The CRMO in Majeed syndrome is distinguished from sporadic CRMO as shown in Table 2. Furthermore, CDA is not a feature of sporadic CRMO.

Table 2. Comparison of Sporadic CRMO and CRMO of Majeed Syndrome

Feature	Sporadic CRMO	CRMO of Majeed Syndrome
Age at onset	Later onset (≤55 yrs)	Early onset (1-19 mos)
Frequency	2-4/yr	1-4/mo
Duration	1-20 yrs	Lifelong
Remission	Frequent	Rare and brief
Long term	Benign course for most	Growth delay
Contractures	Infrequent	Common

CRMO = chronic recurrent multifocal osteomyelitis

Other disorders to consider:

- Chronic multifocal non-bacterial osteomyelitis in hypophosphatasia mimicking malignancy [Girschick et al 2007]
- Cherubism, characterized by painless bilateral, symmetric enlargement of the mandible and/or maxilla resulting from replacement of bone with multilocular cysts composed of fibrotic stromal cells and osteoclast-like cells. Onset is typically between ages two and five years. Other bones are usually not affected and the affected person is otherwise normal. The jaw lesions progress slowly until puberty, when they stabilize and then regress such that by age 30 years facial abnormalities are no longer apparent. *SH3BP2* is the only gene associated with cherubism. *PTPN11* pathogenic variants have also been associated with a cherubism-like phenotype in a small number of patients. Inheritance is autosomal dominant.
- Chronic infantile neurologic, cutaneous, and articular (CINCA) syndrome, a chronic congenital inflammatory disorder characterized by cutaneous rash, neurologic impairment, and arthropathy. CINCA is caused by heterozygous pathogenic variants in *CIAS1* [Aksentijevich et al 2002] and is distinguished from Majeed syndrome by the presence of neurologic involvement, joint symptoms, and a distinctive appearance (frontal bossing, protruding eyes, and limb shortening) and the absence of CDA.
- Deficiency of the interleukin-1 receptor antagonist (DIRA), a chronic inflammatory disorder that begins in the neonatal period. It presents with generalized pustulosis and osteitis. If not recognized and treated

appropriately, affected individuals can develop systemic inflammatory response syndrome (SIRS), which can be a fatal complication of the disease. DIRA is an autosomal recessive disorder caused by pathogenic variants in *IL1RN* [Aksentijevich et al 2009]. It is distinguished from Majeed syndrome by the absence of CDA and by the distinctive radiographic bone lesions.

- Congenital dyserythropoietic anemia (CDA). The CDAs are a heterogeneous group of diseases in which the anemia is predominantly caused by dyserythropoiesis and marked ineffective erythropoiesis [Wickramasinghe & Wood 2005]. Three major types (I, II, and III) have been identified, as well as a few minor types. CDA I is an autosomal recessive disorder (caused by pathogenic variants in *CDAN1* or *C15orf41*) characterized by macrocytic anemia and occasionally associated with acrodysostosis, nail hypoplasia, and scoliosis. In contrast, Majeed syndrome is associated with microcytic CDA and no bone dysplasia or deformity.

Management

Evaluations Following Initial Diagnosis

To establish the extent of disease and needs in an individual diagnosed with Majeed syndrome, the following evaluations are recommended:

- Skeletal radiographs, if not already performed
- CBC, if not already performed
- Bone marrow biopsy if significant anemia is present
- Examination of the skin
- Consultation with a clinical geneticist and/or genetic counselor

Treatment of Manifestations

Because of the rarity of Majeed syndrome, treatment is empiric.

CRMO is treated as follows:

- Nonsteroidal anti-inflammatory drugs (NSAIDs), which provide moderate improvement. If there is an inadequate response to NSAIDs, corticosteroids are useful in controlling CRMO and skin manifestations; however, their long-term use in children is limited by side effects such as growth delay and cataracts.
- Tumor necrosis factor inhibitors and bisphosphonates have been reported to improve sporadic CRMO; however, the two individuals with Majeed syndrome treated with TNF-alpha inhibitors did not improve [Herlin et al 2013]. To date, bisphosphonates have not been used to treat individuals with Majeed syndrome; thus, their efficacy in this disorder is unknown.
- The same two affected individuals reported by Herlin et al who failed to improve with TNF blockade did respond to IL-1 blockade (IL-1 receptor antagonist [anakinra] and IL-1β antibody [canakinumab]) with clinical, radiographic, and laboratory evidence of improvement [Herlin et al 2013].
- Physical therapy to avoid disuse atrophy of muscles or contractures

CDA is treated with periodic monitoring with complete blood count (CBC) and blood transfusion if indicated. One patient had a splenectomy, after which the anemia markedly improved. In the two affected individuals reported by Herlin et al [2013] it is unclear if the CDA completely resolved, as a repeat bone marrow biopsy was not performed.

Prevention of Secondary Complications

Physical therapy can help to prevent contractures.

Surveillance

The following are appropriate:

- Routine CBC to determine if red blood cell transfusion is necessary
- Regular pediatric care

Agents/Circumstances to Avoid

Prolonged bed rest should be avoided because it can result in joint contractures and disuse atrophy.

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 in the US and EU Clinical Trials Register 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.

Other

Three patients were treated with colchicine for four months with no improvement.

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

Majeed syndrome is inherited in an autosomal recessive manner.

Risk to Family Members

Parents of a proband

- The parents of an affected child are obligate heterozygotes (i.e., carriers of one mutated allele).
- Heterozygotes (carriers) do not have Majeed syndrome, but may have psoriasis.

Sibs of a proband

- At conception, each sib of an affected individual has a 25% chance of being affected, a 50% chance of being an asymptomatic carrier, and a 25% chance of being unaffected and not a carrier.
- Once an at-risk sib is known to be unaffected, the risk of his/her being a carrier is 2/3.
- Heterozygotes (carriers) do not develop Majeed syndrome, but may have psoriasis.

Offspring of a proband. The offspring of an individual with Majeed syndrome are obligate heterozygotes (carriers) for a pathogenic variant.

Other family members of a proband. Each sib of the proband's parents is at a 50% risk of being a carrier.

Carrier (Heterozygote) Detection

Carrier testing for at-risk family members is possible once the pathogenic variants have been identified in the family.

In populations with a high carrier rate and/or a high rate of consanguinity, it is possible that the reproductive partner of the proband may be affected or heterozygous. Thus, the risk to offspring is most accurately determined after molecular genetic testing of the proband's reproductive partner.

Related Genetic Counseling Issues

Family planning

- The optimal time for determination of genetic risk, clarification of carrier status, and discussion of the availability of prenatal testing is before pregnancy.
- It is appropriate to offer genetic counseling (including discussion of potential risks to offspring and reproductive options) to young adults who are affected, are carriers, or are at risk of being carriers.

DNA banking is the storage of DNA (typically extracted from white blood cells) for possible future use. Because it is likely that testing methodology and our understanding of genes, allelic variants, and diseases will improve in the future, consideration should be given to banking DNA of affected individuals.

Prenatal Testing and Preimplantation Genetic Diagnosis

Once the pathogenic variants have been identified in an affected family member, prenatal testing for a pregnancy at increased risk and preimplantation genetic diagnosis 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.

- **National Library of Medicine Genetics Home Reference**
  Majeed syndrome
- **Eurofever Registry**
  Largo Gaslini, 5
  Genova 16147
  Italy
  Phone: +39 010 382854; +39 010 393425
  Fax: +39 010 393324; +39 010 4211018
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  Eurofever Registry

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. Majeed Syndrome: Genes and Databases

Gene	Chromosome Locus	Protein	Locus-Specific Databases	HGMD	ClinVar

Table A. continued from previous page.

LPIN2	18p11.31	Phosphatidate phosphatase LPIN2	LPIN2 database The registry of LPIN2 sequence variants	LPIN2	LPIN2

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 Majeed Syndrome (View All in OMIM)

605519	LIPIN 2; LPIN2
609628	MAJEED SYNDROME; MJDS

Gene structure. The genomic sequence of *LPIN2* is approximately 95 kb and comprises 20 exons. Exon 1 and the majority of exon 20 are noncoding (5' and 3' untranslated regions). The mRNA is approximately 6,245 bp and encodes a protein of 896 amino acids. It is expressed in almost all tissues. For a detailed summary of gene and protein information, see Table A, **Gene**.

Pathogenic variants. Three pathogenic variants have been identified: two frameshifts and one splice site change. Four pathogenic variants have been identified: two frameshift variants resulting from a 2-bp deletion, one splice site variant, and one missense variant.

The fourth nucleotide variant is a missense variant that changes an evolutionarily conserved amino acid (see Table 3).

Table 3. Selected *LPIN2* Variants

Variant Classification	DNA Nucleotide Change (Alias [1])	Predicted Protein Change (Alias [1])	Reference Sequences
Benign	c.991G>T	p.Ala331Ser	NM_014646.2 NP_055461.1
	c.1043C>T	p.Pro348Leu	
Pathogenic	c.540_541del	p.Cys181Ter	
	c.2201C>T	p.Ser734Leu	
	c.2327+1G>C	p.Arg776SerfsTer66 [2]	
	c.1316_1317del (1312_1313del)	p.Ser439TrpfsTer15 (Leu438fs+16Ter) [3]	

Variants listed in the table have been provided by the authors. *GeneReviews* staff have not independently verified the classification of variants.
GeneReviews follows the standard naming conventions of the Human Genome Variation Society (varnomen.hgvs.org). See Quick Reference for an explanation of nomenclature.
1. Variant designation that does not conform to current naming conventions
2. Al-Mosawi et al [2007]
3. Herlin et al [2013]

Normal gene product. The protein product comprises 896 amino acids and has a lipin domain and a SMP2 domain. The protein plays a role in lipid metabolism and may play a role in lipodystrophy in mice. In humans it is apparent that it plays a role in inflammation, probably in the innate immune response.

Abnormal gene product. Three of the four identified pathogenic variants produce a truncated protein. The fourth pathogenic variant is a missense variant that changes an evolutionarily conserved amino acid.

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Chapter Notes

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* Professor Hasan A Majeed was a distinguished pediatrician and astute scientist. He was a world-renowned expert on autoinflammatory disorders, contributing tens of manuscripts on the topic in the form of published journal articles, chapters in books, and proceedings in international conferences. Professor Majeed died in November, 2009, at the age of 75 years. He was a great teacher who took pride in educating and training hundreds of physicians. He is greatly missed by his family, friends, colleagues, students, and patients.

Revision History

- 12 September 2019 (ma) Chapter retired: extremely rare
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