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## Craniofacial Abnormalities

Craniofacial is a medical term that relates to the bones of the skull and face. Craniofacial abnormalities are birth defects [<https://medlineplus.gov/birthdefects.html>] of the face or head. Some, like cleft lip and palate [<https://medlineplus.gov/cleftlipandpalate.html>], are among the most common of all birth defects. Others are very rare. Most of them affect how a person's face or head looks. These conditions may also affect other parts of the body.

Treatment depends on the type of problem. Plastic and reconstructive surgery may help the person's appearance.





### Treatments and Therapies

- Craniofacial reconstruction - series [[https://medlineplus.gov/ency/presentations/100058\\_1.htm](https://medlineplus.gov/ency/presentations/100058_1.htm)] (Medical Encyclopedia)  
Also in Spanish [[https://medlineplus.gov/spanish/ency/esp\\_presentations/100058\\_1.htm](https://medlineplus.gov/spanish/ency/esp_presentations/100058_1.htm)]

### Related Issues






















- Baby's Head Shape: Cause for Concern? [<https://www.mayoclinic.org/healthy-lifestyle/infant-and-toddler-health/in-depth/healthy-baby/art-20045964?p=1>] (Mayo Foundation for Medical Education and Research)  
Also in Spanish [<https://www.mayoclinic.org/es/healthy-lifestyle/infant-and-toddler-health/in-depth/healthy-baby/art-20045964?p=1>]






















### Specifics

- Birth Disorders of the Brain and Spinal Cord [<https://www.ninds.nih.gov/health-information/disorders/birth-disorders-brain-and-spinal-cord>]  (National Institute of Neurological Disorders and Stroke)
- Coffin-Lowry Syndrome [<https://www.ninds.nih.gov/health-information/disorders/coffin-lowry-syndrome>]  (National Institute of Neurological Disorders and Stroke)
- Craniosynostosis [<https://www.mayoclinic.org/diseases-conditions/craniosynostosis/symptoms-causes/syc-20354513?p=1>] (Mayo Foundation for Medical Education and Research)
- Flat Head Syndrome (Positional Plagiocephaly) [<https://kidshealth.org/en/parents/positional-plagiocephaly.html>] (Nemours Foundation)  
Also in Spanish [<https://kidshealth.org/es/parents/positional-plagiocephaly.html>]
- Goldenhar Syndrome (For Parents) [<https://kidshealth.org/en/parents/goldenhar-syndrome.html>] (Nemours Foundation)
- Guide to Understanding Apert Syndrome [[https://www.ccakids.org/assets/syndromebk\\_apert.pdf](https://www.ccakids.org/assets/syndromebk_apert.pdf)] (Children's Craniofacial Association) - **PDF**
- Guide to Understanding Pfeiffer Syndrome [[https://ccakids.org/wp-content/uploads/2022/01/pfeiffer-guide\\_2018.pdf](https://ccakids.org/wp-content/uploads/2022/01/pfeiffer-guide_2018.pdf)] (Children's Craniofacial Association) - **PDF**
- Hemifacial Microsomia [<https://www.nidcr.nih.gov/health-info/hemifacial-microsomia>]  (National Institute of Dental and Craniofacial Research)  
Also in Spanish [<https://www.nidcr.nih.gov/espanol/temas-de-salud/la-microsomia-hemifacial>]
- Learning about Holoprosencephaly (HPE) [<https://www.genome.gov/Genetic-Disorders/Holoprosencephaly>]  (National Human Genome Research Institute)

- Noonan Syndrome [<https://www.mayoclinic.org/diseases-conditions/noonan-syndrome/symptoms-causes/syc-20354422?p=1>] (Mayo Foundation for Medical Education and Research)
- Treacher Collins Syndrome [<https://kidshealth.org/en/parents/tcs.html>] (Nemours Foundation)


## Genetics

- 3MC syndrome: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/3mc-syndrome>]  (National Library of Medicine)
- Aarskog-Scott syndrome: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/aarskog-scott-syndrome>]  (National Library of Medicine)
- Acrocallosal syndrome: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/acrocallosal-syndrome>]  (National Library of Medicine)
- Apert syndrome: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/apert-syndrome>]  (National Library of Medicine)
- Baller-Gerold syndrome: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/baller-gerold-syndrome>]  (National Library of Medicine)
- Beare-Stevenson cutis gyrata syndrome: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/beare-stevenson-cutis-gyrata-syndrome>]  (National Library of Medicine)
- Blepharocheilodontic syndrome: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/blepharocheilodontic-syndrome>]  (National Library of Medicine)
- Bohring-Opitz syndrome: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/bohring-opitz-syndrome>]  (National Library of Medicine)
- Bosma arhinia microphthalmia syndrome: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/bosma-arhinia-microphthalmia-syndrome>]  (National Library of Medicine)
- Bowen-Conradi syndrome: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/bowen-conradi-syndrome>]  (National Library of Medicine)
- Branchio-oculo-facial syndrome: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/branchio-oculo-facial-syndrome>]  (National Library of Medicine)
- Burn-McKeown syndrome: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/burn-mckeown-syndrome>]  (National Library of Medicine)
- Cardiofaciocutaneous syndrome: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/cardiofaciocutaneous-syndrome>]  (National Library of Medicine)
- Carpenter syndrome: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/carpenter-syndrome>]  (National Library of Medicine)
- Cerebro-facio-thoracic dysplasia: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/cerebro-facio-thoracic-dysplasia>]  (National Library of Medicine)
- Cherubism: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/cherubism>]  (National Library of Medicine)
- Cleidocranial dysplasia: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/cleidocranial-dysplasia>]  (National Library of Medicine)
- Craniofacial microsomia: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/craniofacial-microsomia>]  (National Library of Medicine)
- Craniofrontonasal syndrome: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/craniofrontonasal-syndrome>]  (National Library of Medicine)
- Craniometaphyseal dysplasia: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/craniometaphyseal-dysplasia>]  (National Library of Medicine)
- Crouzon syndrome with acanthosis nigricans: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/crouzon-syndrome-with-acanthosis-nigricans>]  (National Library of Medicine)

- Crouzon syndrome: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/crouzon-syndrome>]  (National Library of Medicine)
- Enlarged parietal foramina: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/enlarged-parietal-foramina>]  (National Library of Medicine)
- Frontonasal dysplasia: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/frontonasal-dysplasia>]  (National Library of Medicine)
- Gorlin-Chaudhry-Moss syndrome: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/gorlin-chaudhry-moss-syndrome>]  (National Library of Medicine)
- Greig cephalopolysyndactyly syndrome: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/greig-cephalopolysyndactyly-syndrome>]  (National Library of Medicine)
- Hartsfield syndrome: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/hartsfield-syndrome>]  (National Library of Medicine)
- Isolated Pierre Robin sequence: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/isolated-pierre-robin-sequence>]  (National Library of Medicine)
- Jackson-Weiss syndrome: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/jackson-weiss-syndrome>]  (National Library of Medicine)
- Mandibulofacial dysostosis with microcephaly: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/mandibulofacial-dysostosis-with-microcephaly>]  (National Library of Medicine)
- Miller syndrome: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/miller-syndrome>]  (National Library of Medicine)
- Muenke syndrome: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/muenke-syndrome>]  (National Library of Medicine)
- Nager syndrome: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/nager-syndrome>]  (National Library of Medicine)
- Oral-facial-digital syndrome: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/oral-facial-digital-syndrome>]  (National Library of Medicine)
- Pfeiffer syndrome: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/pfeiffer-syndrome>]  (National Library of Medicine)
- Robinow syndrome: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/robinow-syndrome>]  (National Library of Medicine)
- Saethre-Chotzen syndrome: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/saethre-chotzen-syndrome>]  (National Library of Medicine)
- Shprintzen-Goldberg syndrome: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/shprintzen-goldberg-syndrome>]  (National Library of Medicine)
- Sotos syndrome: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/sotos-syndrome>]  (National Library of Medicine)
- Thanatophoric dysplasia: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/thanatophoric-dysplasia>]  (National Library of Medicine)
- Treacher Collins syndrome: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/treacher-collins-syndrome>]  (National Library of Medicine)
- Weaver syndrome: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/weaver-syndrome>]  (National Library of Medicine)

## Clinical Trials

- ClinicalTrials.gov: Craniofacial Abnormalities [<https://clinicaltrials.gov/search?cond=%22Craniofacial+Abnormalities%22&aggFilters=status:not%20rec>]  (National Institutes of Health)


ClinicalTrials.gov: Craniosynostoses [https://clinicaltrials.gov/search?cond=%22Craniosynostoses%22&aggFilters=status:not%20rec]  (National Institutes of Health)

## Journal Articles

References and abstracts from MEDLINE/PubMed (National Library of Medicine)

- Article: Craniomaxillofacial-Derived MSCs in Congenital Defect Reconstruction. [https://www.ncbi.nlm.nih.gov/pubmed/40723825]
- Article: Surgical treatment of first branchial cleft anomalies using retrograde facial nerve... [https://www.ncbi.nlm.nih.gov/pubmed/40694114]
- Article: Characterizing healthcare resource utilization in two rare diseases (Kleefstra syndrome and... [https://www.ncbi.nlm.nih.gov/pubmed/40624551]
- Craniofacial Abnormalities -- see more articles [https://pubmed.ncbi.nlm.nih.gov/?term=%22Craniofacial+Abnormalities%22%5Bmajr%3Anoexp%5D+AND+humans%5Bmh%5D+AND+english%5Bla%5D+AND+%22last+1+Year%22+%5Bedat%5D+NOT+%28letter%5Bpt%5D+OR+case+reports%5Bpt%5D+OR+editorial%5Bpt%5D+OR+comment%5Bpt%5D%29+AND+free+full+text%5Bsb%5D+]

## Find an Expert

- American Academy of Facial Plastic and Reconstructive Surgery [https://www.aafprs.org/]
- National Institute of Dental and Craniofacial Research [https://www.nidcr.nih.gov/]   
Also in Spanish [https://www.nidcr.nih.gov/espanol]

## Patient Handouts

- Apert syndrome [https://medlineplus.gov/ency/article/001581.htm] (Medical Encyclopedia)  
Also in Spanish [https://medlineplus.gov/spanish/ency/article/001581.htm]
- Cleidocranial dysostosis [https://medlineplus.gov/ency/article/001589.htm] (Medical Encyclopedia)  
Also in Spanish [https://medlineplus.gov/spanish/ency/article/001589.htm]
- Craniosynostosis [https://medlineplus.gov/ency/article/001590.htm] (Medical Encyclopedia)  
Also in Spanish [https://medlineplus.gov/spanish/ency/article/001590.htm]
- Craniosynostosis repair [https://medlineplus.gov/ency/article/007364.htm] (Medical Encyclopedia)  
Also in Spanish [https://medlineplus.gov/spanish/ency/article/007364.htm]
- Head and face reconstruction [https://medlineplus.gov/ency/article/002980.htm] (Medical Encyclopedia)  
Also in Spanish [https://medlineplus.gov/spanish/ency/article/002980.htm]
- Pierre Robin sequence [https://medlineplus.gov/ency/article/001607.htm] (Medical Encyclopedia)  
Also in Spanish [https://medlineplus.gov/spanish/ency/article/001607.htm]



Apert syndrome [<https://medlineplus.gov/ency/article/001581.htm>]

Cleidocranial dysostosis [<https://medlineplus.gov/ency/article/001589.htm>]

Craniofacial reconstruction - series [[https://medlineplus.gov/ency/presentations/100058\\_1.htm](https://medlineplus.gov/ency/presentations/100058_1.htm)]

Craniosynostosis [<https://medlineplus.gov/ency/article/001590.htm>]

Craniosynostosis repair [<https://medlineplus.gov/ency/article/007364.htm>]

Craniosynostosis repair - discharge [<https://medlineplus.gov/ency/patientinstructions/000067.htm>]

Head and face reconstruction [<https://medlineplus.gov/ency/article/002980.htm>]

Increased head circumference [<https://medlineplus.gov/ency/article/003305.htm>]

Metopic ridge [<https://medlineplus.gov/ency/article/003312.htm>]

Micrognathia [<https://medlineplus.gov/ency/article/003306.htm>]

Noonan syndrome [<https://medlineplus.gov/ency/article/001656.htm>]

Pierre Robin sequence [<https://medlineplus.gov/ency/article/001607.htm>]

Prognathism [<https://medlineplus.gov/ency/article/003026.htm>]

Short philtrum [<https://medlineplus.gov/ency/article/003302.htm>]

Skull x-ray [<https://medlineplus.gov/ency/article/003802.htm>]

Sutures - ridged [<https://medlineplus.gov/ency/article/003308.htm>]

Treacher Collins syndrome [<https://medlineplus.gov/ency/article/001659.htm>]

## Related Health Topics

Brain Malformations [<https://medlineplus.gov/brainmalformations.html>]

Cleft Lip and Palate [<https://medlineplus.gov/cleftlipandpalate.html>]

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