FACTSHEET #4
FACIAL FEATURES

Noonan Syndrome Association

People with Noonan syndrome have characteristic facial features, particularly during childhood, which change with age and may be insignificant in adult life.

In early childhood, ptosis (drooping eyelids), low set ears, short neck, and low hairline are characteristic.

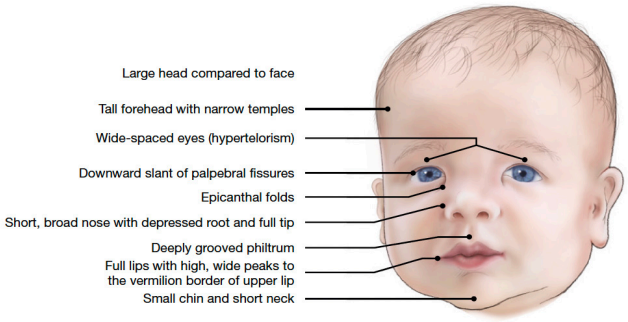
The facial features include:

All ages

- Eyes: May be strikingly blue with arched or diamond-shaped eyebrows
- Ears: Low set, posteriorly rotated, thick helices

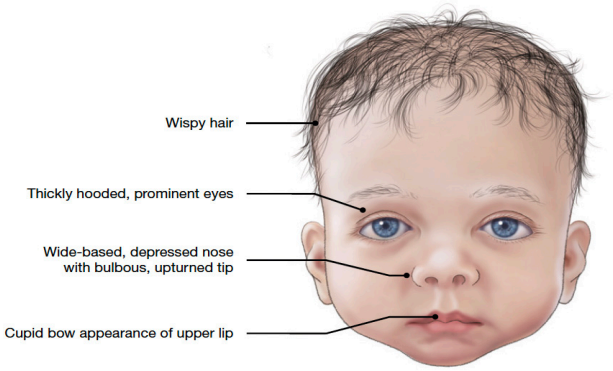
Newborn baby*

**Features can be subtle or absent.*



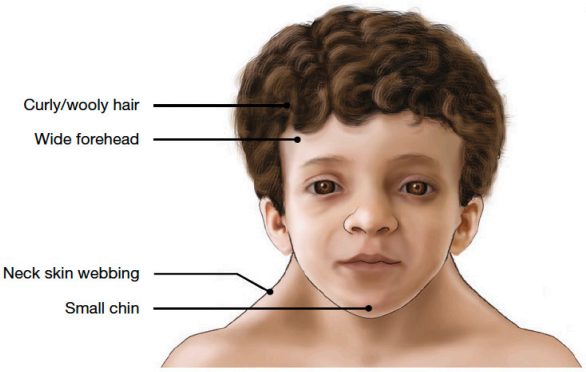
- Forehead, face, hair: Tall forehead, low posterior hairline
- Eyes: Widely spaced, drooping eyelids (ptosis), epicanthal folds (skin fold of the upper eyelid covering the inner corner of the eye)
- Nose: Short and broad, depressed root, upturned tip
- Mouth: Deep groove between the mouth and nose (the philtrum), small lower jaw (micrognathia)
- Neck: Excessive skin at the back of the neck

Infancy (2–12 months)



- Forehead, face, hair: Large head, tall and prominent forehead
- Eyes: Widely spaced, drooping of the upper eyelid, or thick-hooded eyelids
- Nose: Short, wide, depressed nasal root

Childhood (1–12 years)



- Forehead, face, hair: Features might appear coarse, triangular face
- Neck: Webbing may be obvious

FACTSHEET #4
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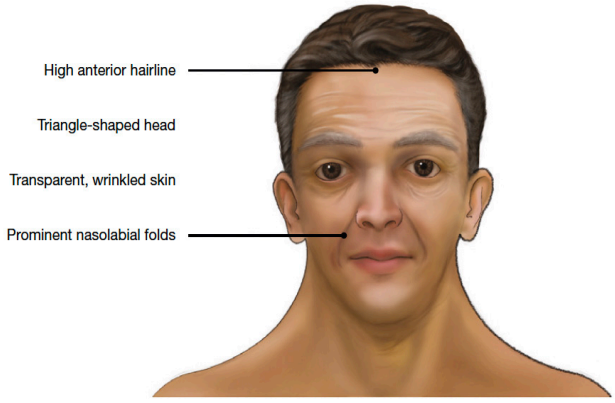
Noonan Syndrome Association

Adolescence & Adulthood



Aolescence (12–18 years)

- Forehead, face, hair: Expressionless face
- Nose: Bridge is high and thin
- Neck: Webbing may be obvious

Adulthood (>18 years)

- Forehead, face, hair: Distinguishing facial features are subtle, skin appears thin and transparent
- Nose: Prominent nasolabial fold

All Images courtesy: National Human Genome Research Institute, www.genome.gov

A 2017 study used facial analysis technology to examine 161 people with Noonan syndrome from 5 different global populations – Caucasian, African and African American, Asian and Latin American.

They found that the facial features were very similar across all the populations, and the technology could diagnose patients from all population groups with a sensitivity and specificity of 88% and 89%, respectively.

The researchers proposed that the technology could support clinicians in making accurate Noonan syndrome diagnoses, helping with earlier detection and increased recognition of the syndrome throughout the world.

REFERENCES/FURTHER READING

Roberts AE, Allanson JE, Tartaglia M, Gelb BD. Noonan syndrome. Lancet. 2013;381(9863):333-342.

Kruszka P, et al. Noonan syndrome in diverse populations. Am J Med Genet 2017; 173(9):2323-2334.

Hammond P, et al. 3D analysis of facial morphology. Am J Med Genet 2004;126A(4):339–348.

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