

24.1

CLINICAL APPLICATION

IT'S ALL IN THE GENES

Do you have uncombable hair, misshapen toes or teeth, a pigmented tongue tip, or an inability to smell skunk? Do you lack teeth, eyebrows, eyelashes, nasal bones, thumbnails, or fingerprints? Can you wiggle your ears? If so, you may find your unusual trait described in *Online Mendelian Inheritance in Man*, which catalogs more than 10,000 known human genetic variants (www.ncbi.nlm.nih.gov/entrez/query.fcgi?db=OMIM). Most of the entries include family histories, clinical descriptions, molecular information, and how the trait is transmitted. Amidst the medical terminology can be found some fascinating inherited traits in humans, from top to toes.

Genes control whether hair is blond, brown, or black, whether or not it has red highlights, and whether it is straight, curly, or kinky. Widow's peaks, cowlicks, a whorl in the eyebrow, and white forelocks run in families, as do hairs with triangular cross sections. Some people have multicolored hairs like cats, and others have hair in odd places, such as on the elbows, nosetip, knuckles, palms of the hands, or soles of the feet. Teeth can be missing or extra, protuberant or fused, present at birth, or "shovel-shaped" or "snow-capped." A person can have a grooved tongue, duckbill lips, flared ears, egg-shaped pupils, three rows of eyelashes, spotted nails, or "broad thumbs and great toes." Extra

breasts have been observed in humans and guinea pigs, and one family's claim to fame is a double nail on the littlest toes.

Unusual genetic variants can affect metabolism, sometimes with noticeable effects. Members of some families experience "urinary excretion of odoriferous component of asparagus" or "urinary excretion of beet pigment" after eating the implicated vegetables. In "blue diaper syndrome," an infant's inherited inability to break down an amino acid turns urine blue on contact with air.

One father and son could not open their mouths completely. "Dysmelodia" is the inability to carry a tune. Those with "odor blindness" cannot smell musk, skunk, cyanide, or freesia flowers. Motion sickness, migraine headaches, and stuttering may be inherited. Uncontrollable sneezing may be due to Achoo syndrome (an acronym for "autosomal dominant compelling helioophthalmic outburst" syndrome). Figure 24A illustrates some more common genetic traits. ■

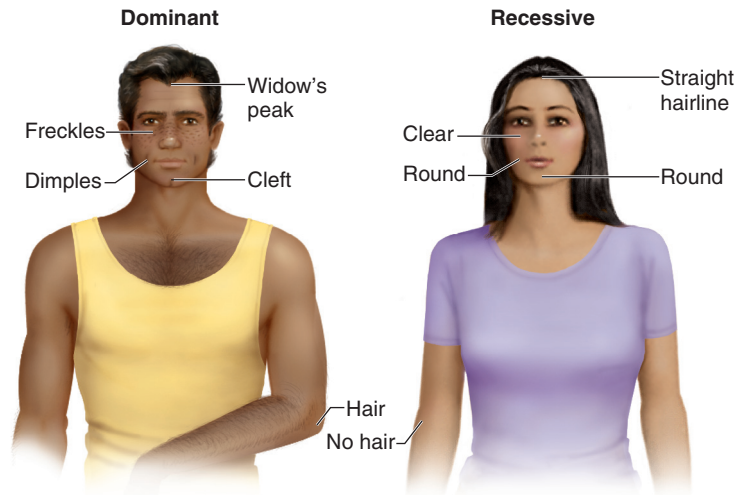


FIGURE 24A

Inheritance of some common traits: freckles, dimples, widow's peak, hairy elbows, and a cleft chin.