Ped. Genetik

9

From M. A. Akşit's collection

Odd looking or facies abnormal babies*

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From a discussion of a patient at the Neonatology Intensive Care Unit, with a Nurse (E. Topaloğlu), and Pediatric Genetic Specialist (M. A. Aksit).
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When you want to confirm the abnormalities, mostly indicates not normal, thus, the main description is about the determination of normal, thus malformation I the routine descriptive one. For malformation it is mostly be quite sure on the abnormality, e.g. six fingers are obvious. But when an estimation, we can indicate as odd looking, or something is wrong, not quite physiological estimation and expectations is required. The pathological evidences are not quite indicted the diagnosis, as odd-looking infant, it is just a major finding not a diagnosis.

Systematic survey on the diagnosis of malformations are mostly when obvious ones is confirmed. Odd-looking is somehow not to be quite establishing one. So, in this Unit, by the PossumCore notification, the facial approach to the abnormalities is taken in notice.

In this Unit, we are indicating the odd-looking concept as the facial abnormalities, under the *Possumcore web,* Smith's *Recognizable Patterns of Human Malformations* contributions and "*Genetik Hastalıklarda Klinik Özellikler, Genetik Danışmanlık/Clinic specifications at the genetic diseases*, EsOGÜ Pediatric Genetics Department Guideline, January 2014.

Outline

Odd looking or facies abnormal babies

AIM: Although the most advance ultrasonography, by directly four dimensions looking the face, the abnormalities are hard to confirm. So, the first to be noticeable is suspicious and taken measurement for the remarkable evidence is or not. In this Chapter, the evaluation of odd-looking infant is discussed.

Grounding Aspects: The PossumCore and "*Genetik Hastalıklarda Klinik Özellikler, Genetik Danışmanlık/Clinic specifications at the genetic diseases*, EsOGÜ Pediatric Genetics Department Guideline, January 2014" is the main source of the information.

Introduction: The structure, the constitution of a baby; from genetically parents, thus, several influences are affected the growth and development. Some factors as odd-looing is hard to identified and determined, like polydactyly. The facial evaluation is discussed at this Unit.

<u>General Considerations</u>; The perspective of the face is concern as; significantly abnormal, small, wide, premature looking, course, elfin, expressionless/hypotonic/myopathic. abnormal facial skin folds and creases noticed one.

Proceeding: As the general standardization of facial measurement, and the differentiation of the abnormality is presented.

Notions: In order to be evaluate, the general face structure have been indicated as; interpupillary distance, outer eyeball length, inner eyeball length, nose broad, palpebral fold length, and the ear structure, length, etc.

Conclusion: The facial abnormalities as notices odd-looking is stated at this Chapter.

Key Words: Odd-looking, facial abnormalities as; significantly abnormal, small, wide, premature looking, course, elfin, expressionless/hypotonic/myopathic. abnormal facial skin folds and creases noticed one

Özet

Değişik görünümlü veya yüzünde anormallik olan bebekler

Amaç: En gelişmiş ultrasonlarla doğudan yüz görünümü gözlenen tetkiklerle de anormal görünümü saptamak oldukça zordur. Öncelikle şüphelenerek, ölçümlerin yapılması katkı sağlayabilecektir. Bu Ünite değişik görünümlü bebekler irdelenecektir.

Dayanaklar/Kaynaklar: Web sitesi olarak The PossumCore ve "*Genetik Hastalıklarda Klinik Özellikler, Genetik Danışmanlık*, EsOGÜ Pediatrik Genetik Bilim Dalı Yayını Ocak 2014" teme alınmıştır.

Giriş: Bebeğin genetik yapısı ve fiziksel görünümü aileden gelen özellikler yanında diğer büyüme ve gelişmedeki çeşitli faktörler de etkilemektedir. Yüz görünümde ise fark edilmesi altı parmak gibi kolay olmamaktadır. Bu Bölümde bu konu irdelenmektedir.

<u>Genel Yaklaşım</u>; Yüz görünümlerindeki aykırılıklar, belirgin anormal yüz, ufak, iri ve geniş, kaba, melek tipi, donuk/hipotonik/myopatik görünüm, anormal cilt katlandıkları olması gibi durumlar üzerin eğinilmektedir.

Başlıca boyutlar: İlk planda yüz ölçümleri belirtilmektedir ki farklılıklar saptanabilmesi sağlansın.

Yaklaşım: Bazı ölçümler sunulmaktadır; iki göz arasındaki mesafe, dış göz uzunluğu, göz küre iç uzunluğu, burun genişliği, palpebral katlantı uzunluğu ve kulak yapısı, uzunluk, açı gibi boyutlar sunulmaktadır.

Sonuç: Değişik yüz görünümü olan bebeklerin yüz anormallikleri ele alınarak konu irdelenmektedir.

Anahtar Kelimeler: Yüz görünümü anormal olan bebekler, yüzde anormal olma durumları; belirgin anormal, ufak, geniş, kaba, ince elfin tipi, prematüre görünümlü, Apatik yüz görünümlü ve yüzde farklı katlantı ve kıvrımların olmasıdır.

Odd Looking?

When you mean odd looking, you must indicate the parameters. Thus, in some occasions it is hard to mentioned. So, the measuring and specification what you mean is indicated for understandable concept. If you mentioned as hypertelorism, there must be measurably be a reality, not your subjective indication.

From Possumcore perspective; This Unit these parameters are indicated by picture and graphical standard curves.

General impression

- o Facies significantly abnormal
  - o Round face
  - o Square face

- o Triangular face
- o Long face
- o Flat face
- o Broad cheeks
- o Jowls, sagging cheeks
- o Structural asymmetry of face
- o Facial palsy
- o Boney distortion of face
- o Small face
- o Large face
- o Prematurely aged face
- o Coarse face
- o Fine or elfin face
- o Expressionless/hypotonic/myopathic face
- o Abnormal facial skin folds and creases

Face and Eye Dimensions

The face general measurable parameters are as indicated below.

Common measured lengths

Smith's Recognizable Patterns of Human Malformations

Measurable
A: Outer eyeball

B: Nose broth

C: Pupillary length

D: Nose-lips
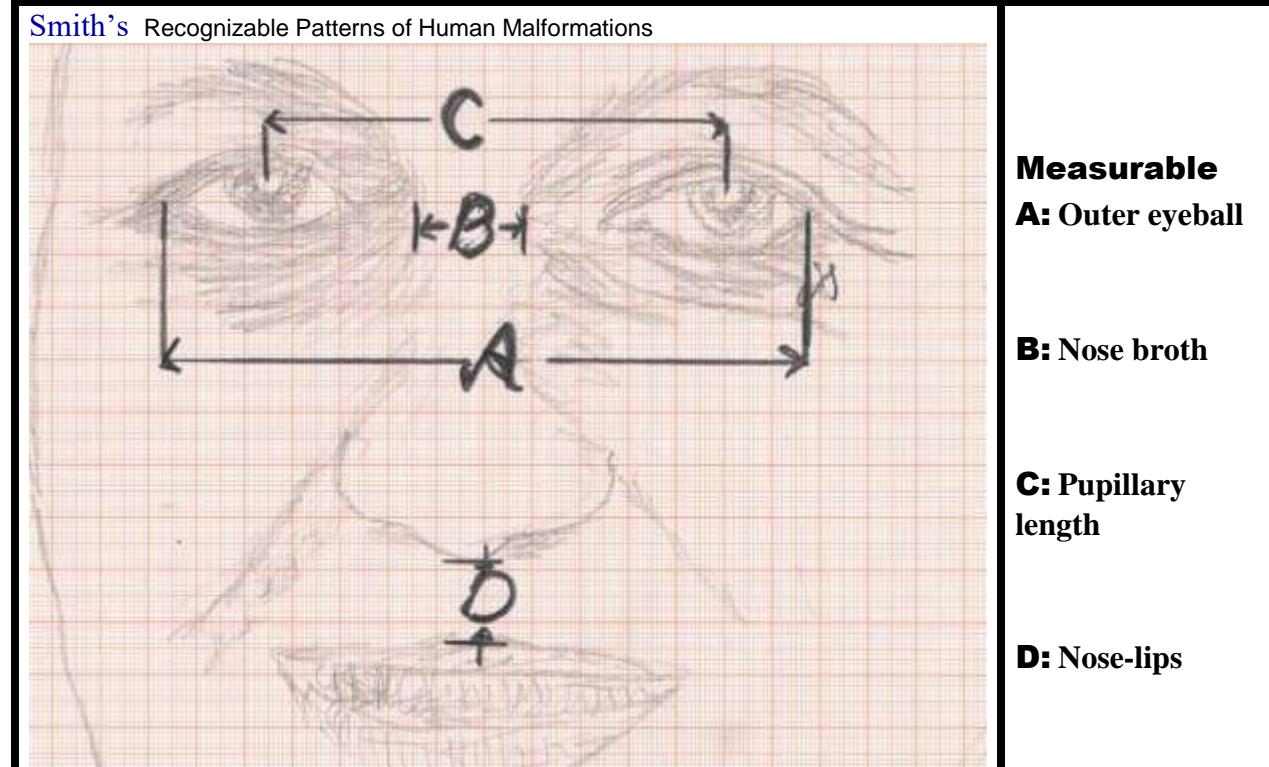
Figure 9-1: The distance between the points to taken care

The detailed measurement is considered at the next figure.

More detailed overview
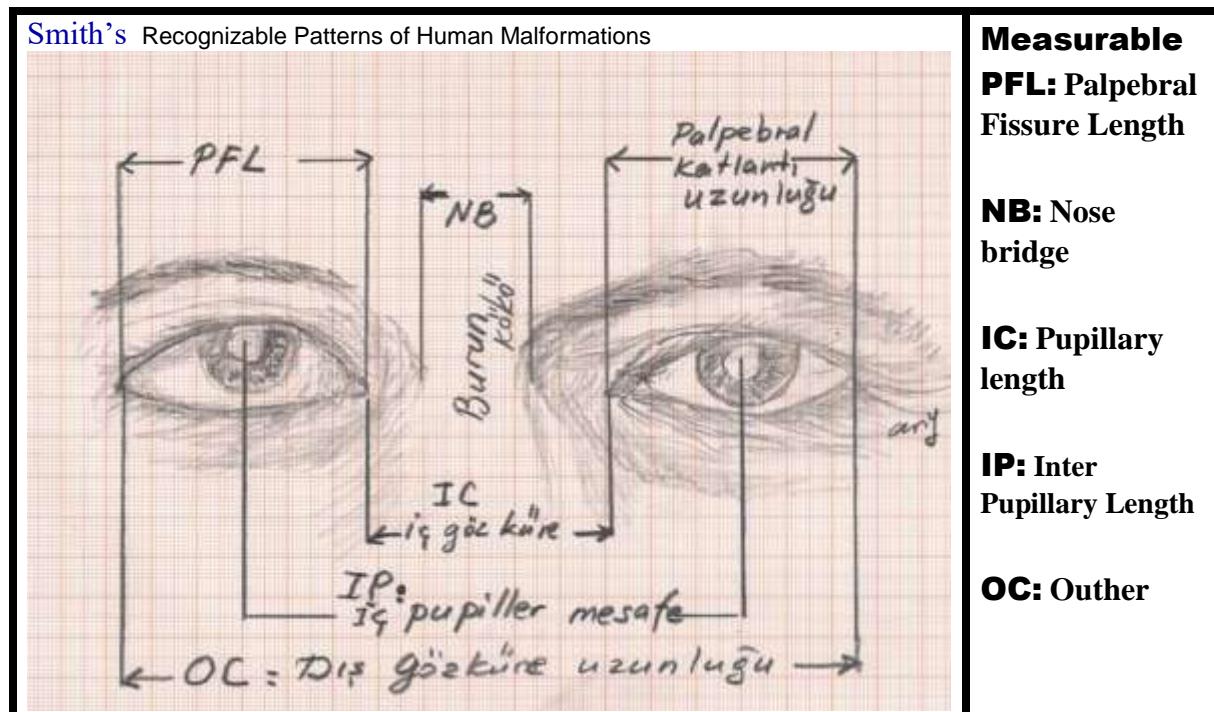
Figure 9-2: The length that can be consider for measurement

These measurements must be plotted to the carts for the evaluation of the standard length.

Palpebral Fissure Length (PFL)
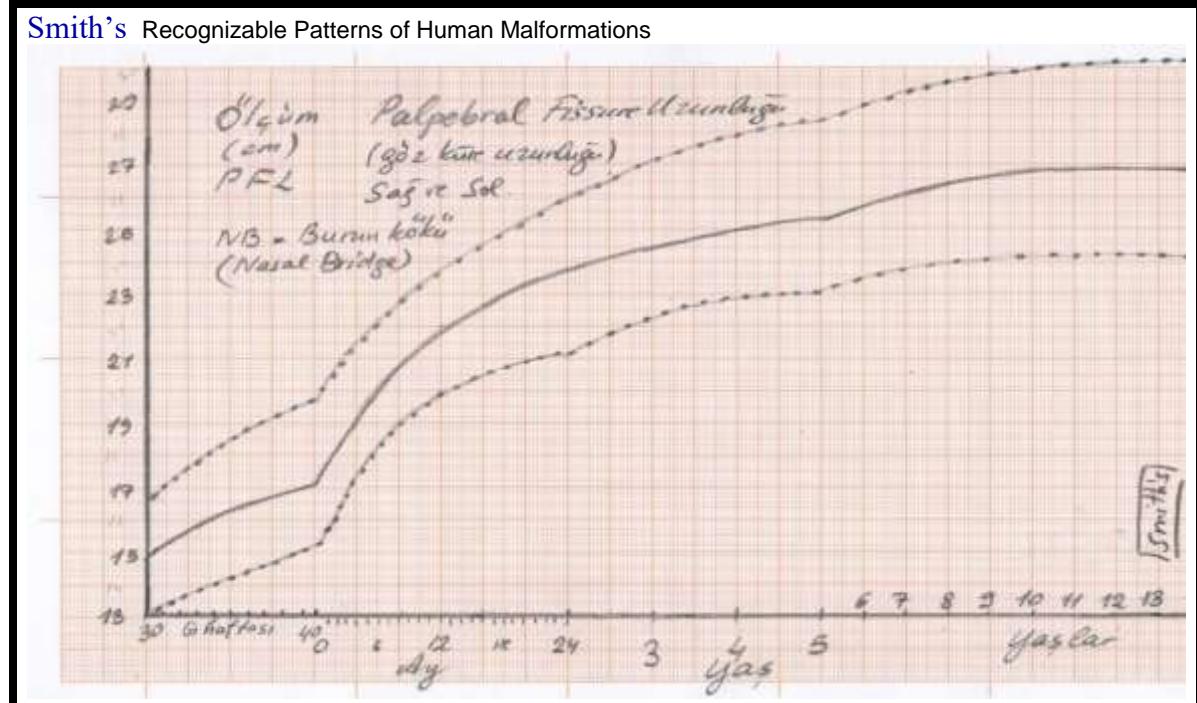
Figure 9-3: The standard curves; minimum, medium and maximum

Inner Central Distance (ICD)
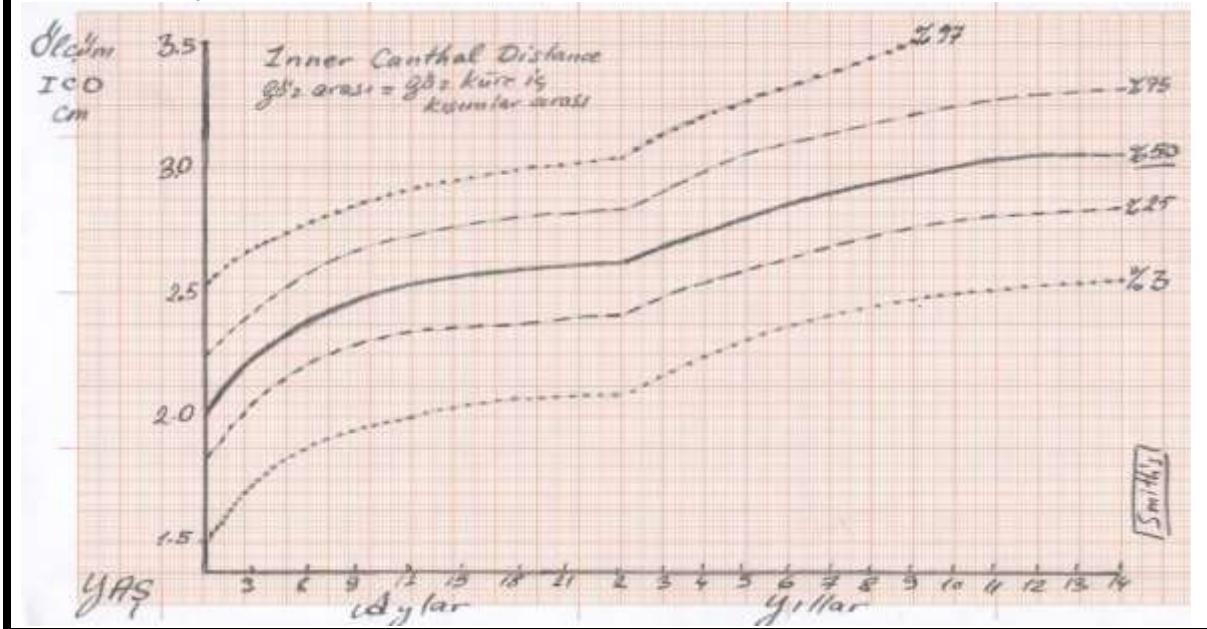
Figure 9-4: The indication of standard deviations, for more precious measurement

Outer Central Distance (OCD)
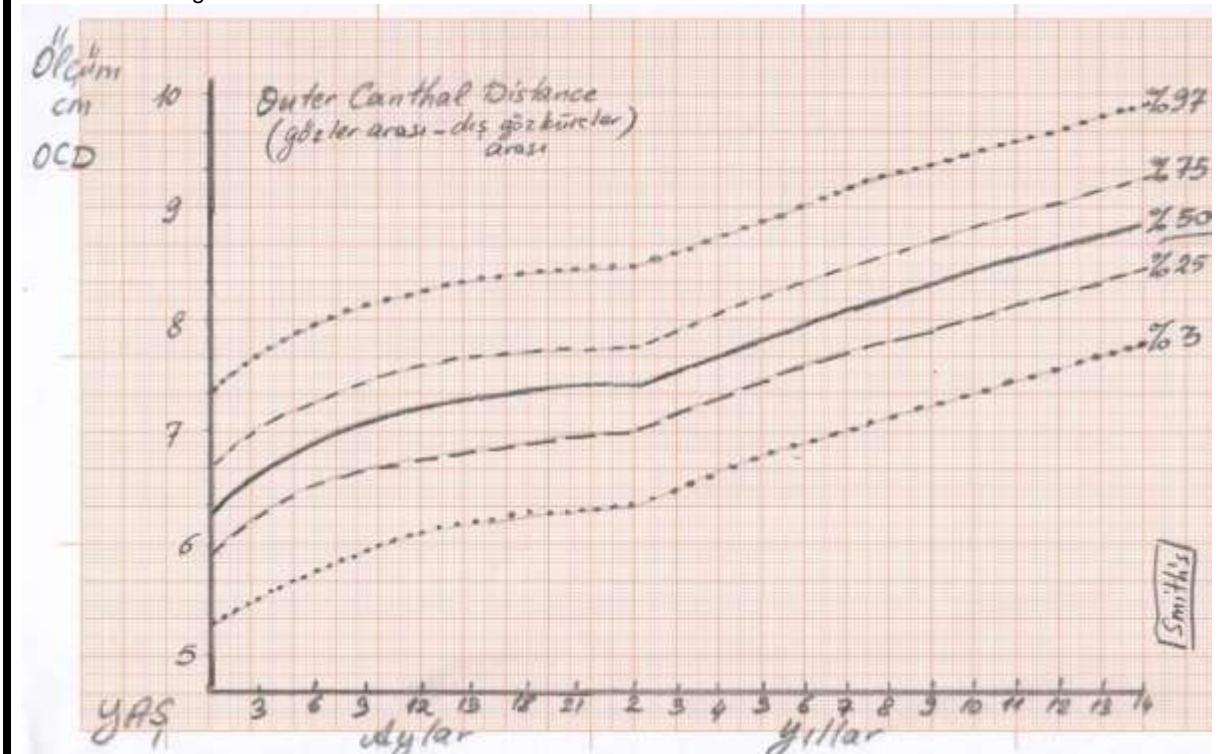
Figure 9-4: The length variations, form 3% to 97 percent

Inter Pupillary Distance (IPD)
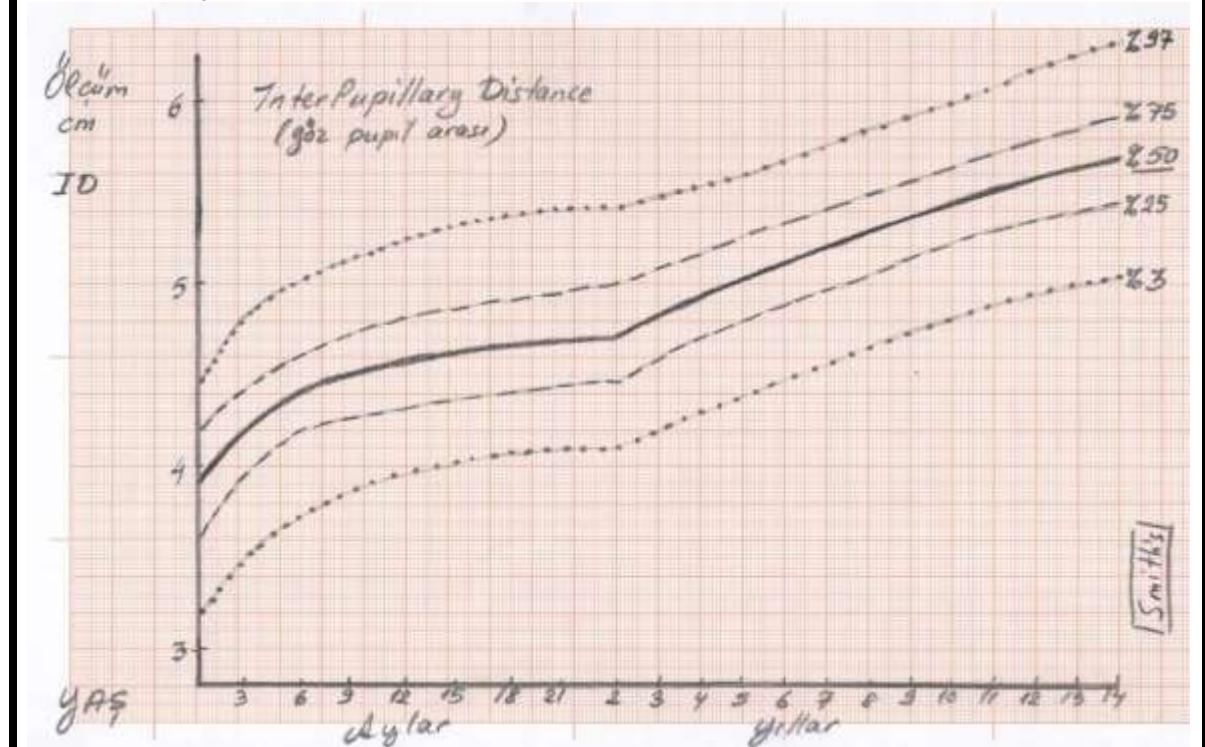
Figure 9-5: Inter Pupillary Distance, according to the ages; moths and years

Palpebral Fissure Length (PFL)
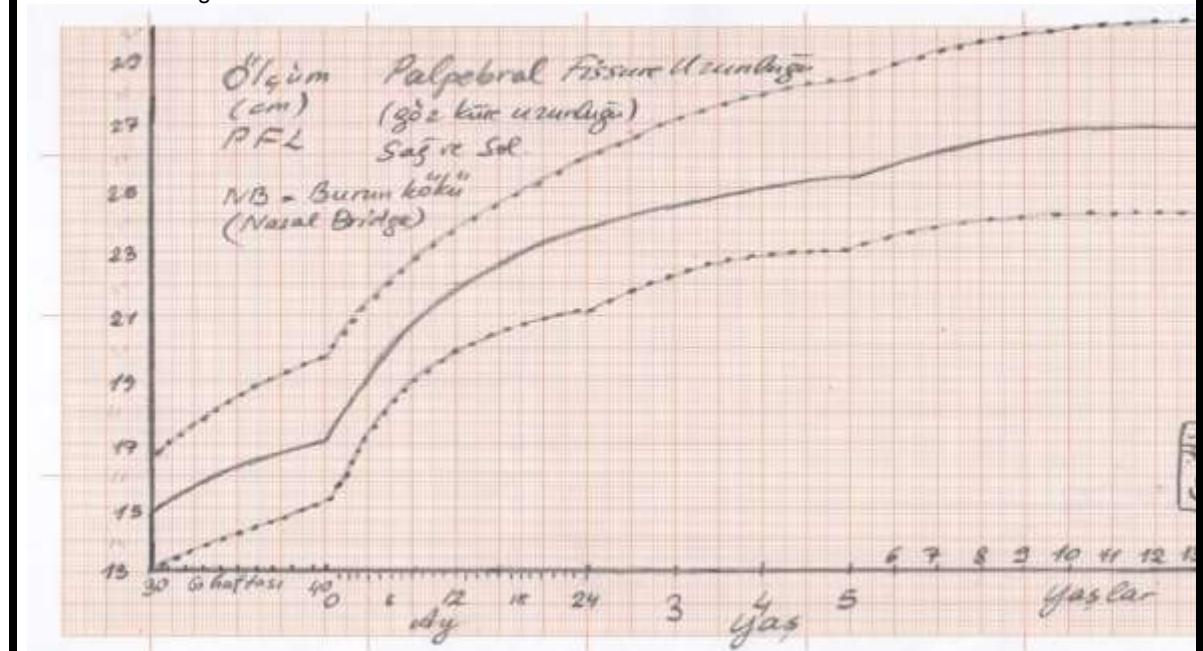
Figure 9-6: Palpebral Fissure Length, due to the gestational weeks

Ear

Ear measurement is quite different then the face and eye, thus must consider in different perspective

Common Ear Measurement

Smith's Recognizable Patterns of Human Malformations
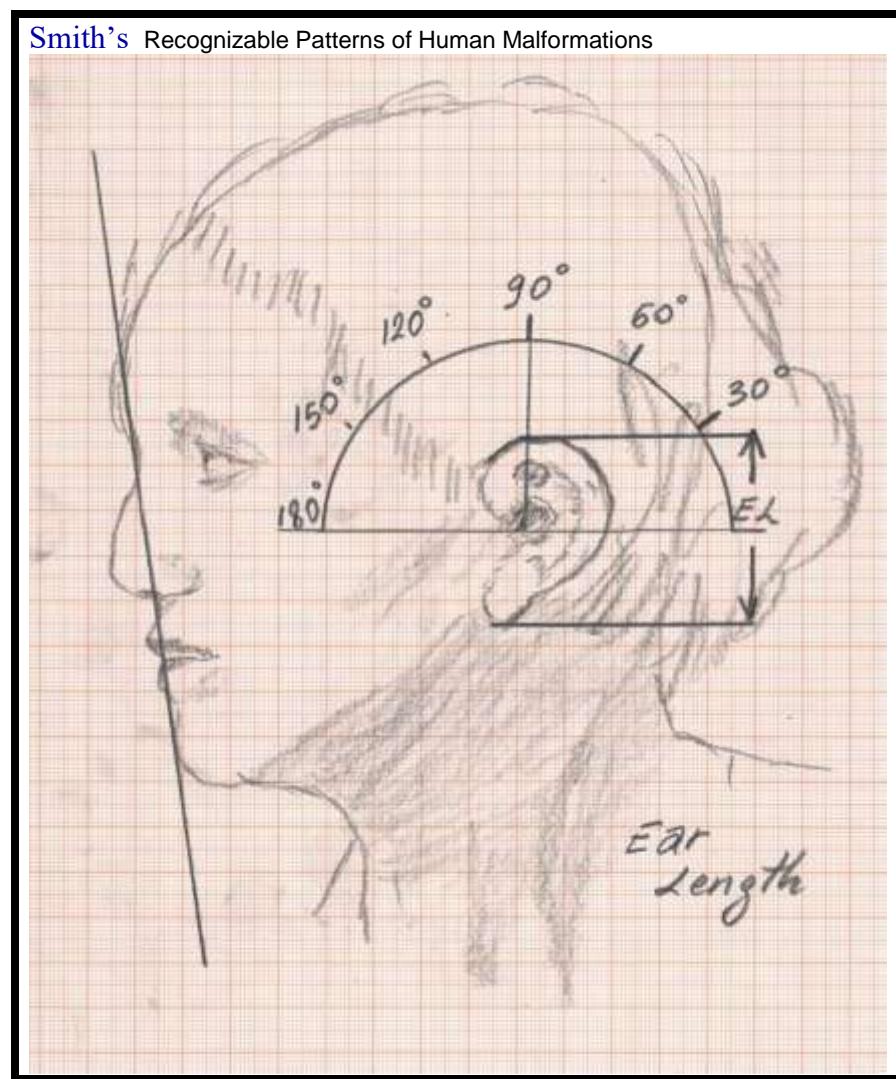
Figure 9-7: Ear Length and degree of the position

Ear is not only for measurement of the length; thus, the rotation is also important.

1) The original fetal rotation is about 150 degrees, so turning to 60 degrees.
2) The fetal ear is broad, later narrowed, more swollen type.
3) The out opening is also important for discussion
4) There are some skin folds and as nipple findings
5) The mandibula and eye line must be in straight

Ear Length (EL)
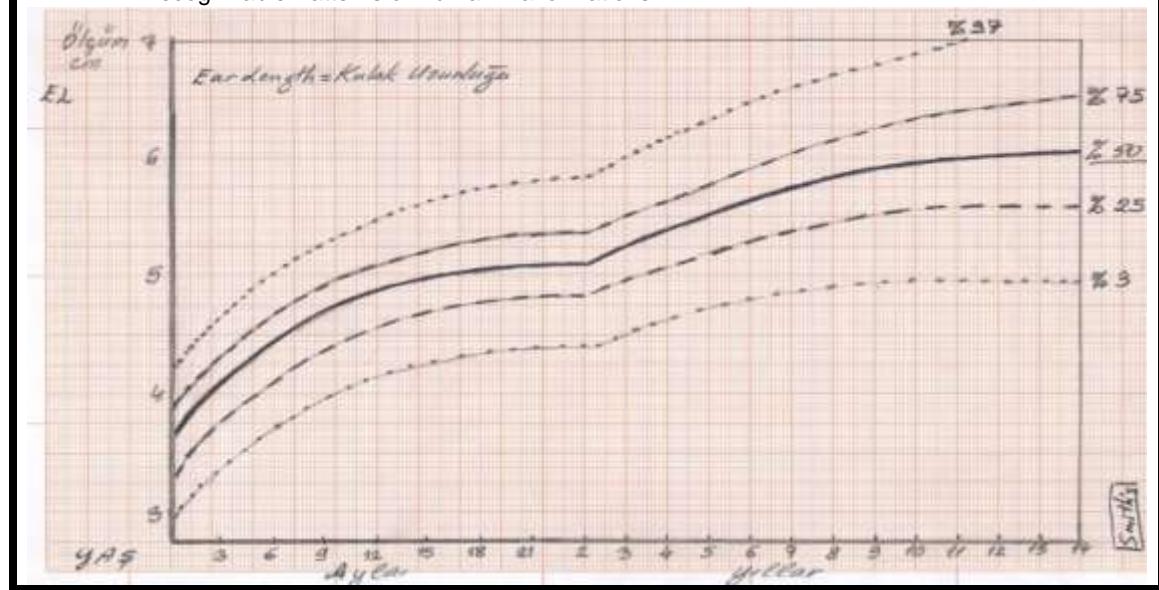
Figure 9-8: Ear Length, according to the ages; moths and years

The length differences are so obvious, as seen Figure 9, at the abnormalities.

## Ear Length, by comparison to Down's Syndrome
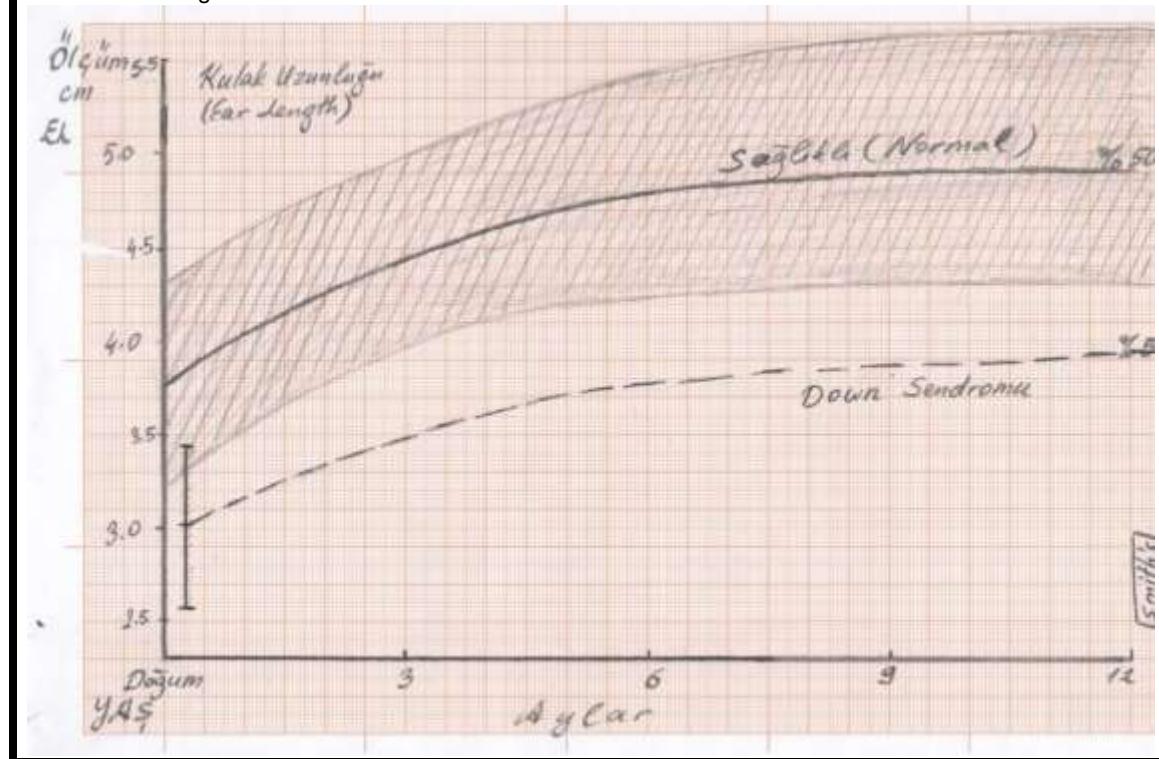
Figure 9-9: The differences of the ear length at Down's and normal babies, by the median line

Universal Appearance

Comment: This is only some characteristics of the faces. The real pictures are taken from Google, pictures.

The heading are as follows:
- o Facies significantly abnormal
    - o Round face
    - o Square face
    - o Triangular face
    - o Long face
    - o Flat face
    - o Broad cheeks
    - o Jowls, sagging cheeks
    - o Structural asymmetry of face
    - o Facial palsy
    - o Boney distortion of face
- o Small face
- o Large face
- o Prematurely aged face
- o Coarse face
- o Fine or elfin face
- o Expressionless/hypotonic/myopathic face
- o Abnormal facial skin folds and creases

Pictures

Commonly Google pictures is used for demonstration of the shapes.
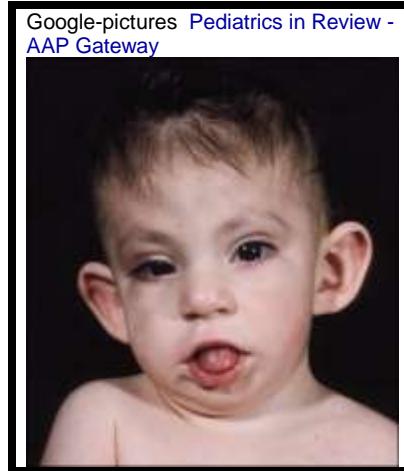
| Facies significantly abnormal	Characteristics of abnormal face
Google-pictures Pediatrics in Review - AAP Gateway	Google-pictures/ Reddit
Figure 9-10:	**Figure 9-11:**
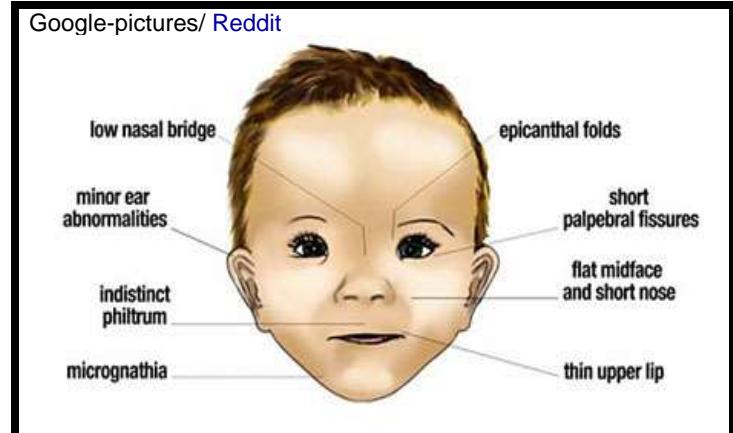
These figures are obviously abnormal faces; thus, physician must indicate the measurements and the findings, not mentioned as abnormality, just some characteristics.

Some of them as you noticed, are from normal person face.

Long, Heart, Oval, Square and Round face

Google-pictures/ Complete Fashion

Oblong

Heart

Oval

Square

Round
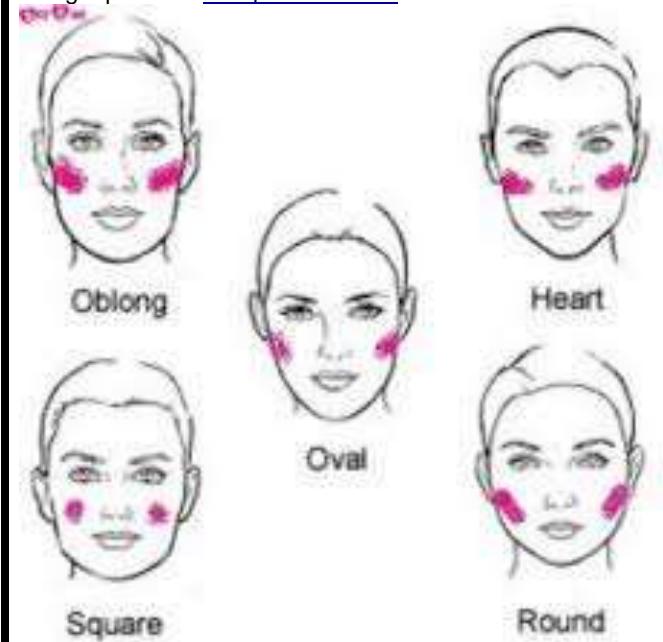
Figure 9-12: Different face perspectives

Broad cheeks

Google-pictures SelectSpecs
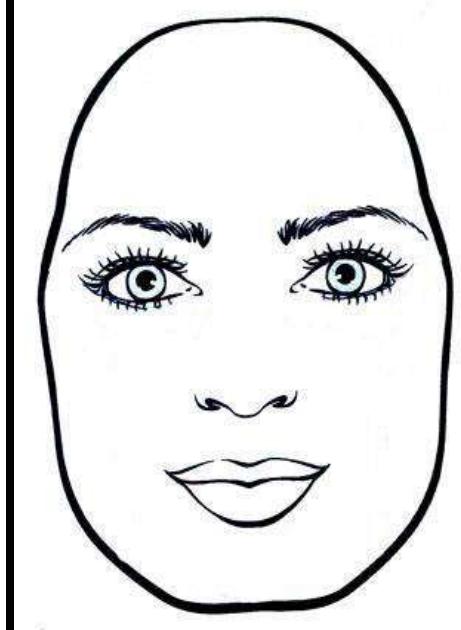
Figure 9-13: Broad Cheeks

Triangular face

Google-pictures cloudmind.info
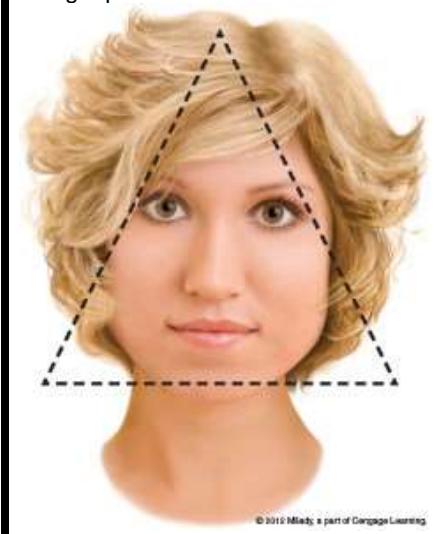
Google-pictures UPMC HealthBeat

Figure 9-14: Triangular face

There are two main different triangular face

Flat face

Google-pictures ID Hospital Korea Plastic Surgery - blogger

Figure 9-15: Flat Face, before and after surgery operation

Jowls, sagging cheeks

Google-pictures/ Pinterest

Figure 9-16: Before and after the Plastic Surgery

Facial palsy

Google-pictures/ Carolina Facial Plastics

Figure 9-17: Characteristics of Facial Palsy

Boney distortion of face

Google-pictures/ Pinterest

Figure 9-18: Boney distortion

## Discriminating Features and Associated ones

Google-pictures/ Reddit
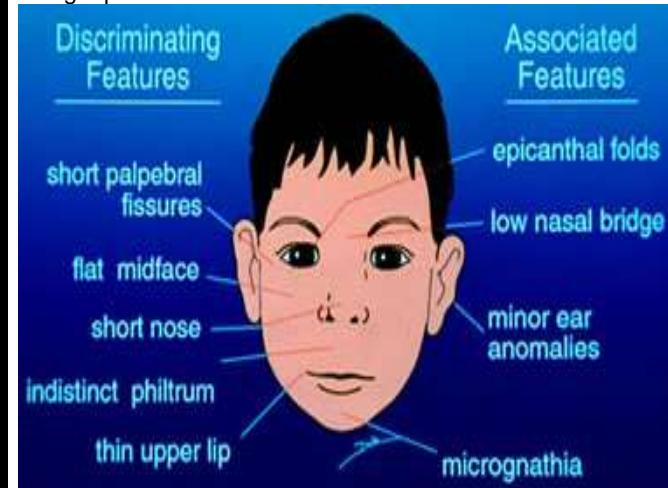
Figure 9-19: Boney distortion

Coarse face

Google-pictures/ Pediatrics Clerkship - University of Chicago
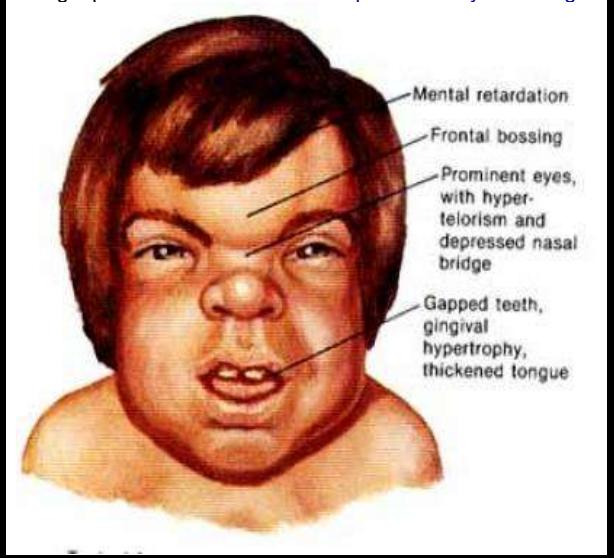
Figure 9-20: Coarse face

Fine or elfin face

Google-pictures/ WordPress.com

Figure 9-21: Twinklerosefairy's Blog | A fine WordPress.com site

The above pictures/drawings are obvious for abnormality; thus, the below ones are near and normal appearances.

Some of them are humor for the artistic commercial ones (Leonardo DiCaprio with a Small Face, for freaking).

Small face

Google-pictures/ Freaking News

Figure 9-22: Leonardo DiCaprio with a Small Face

Large face

Google-pictures/ Pinterest
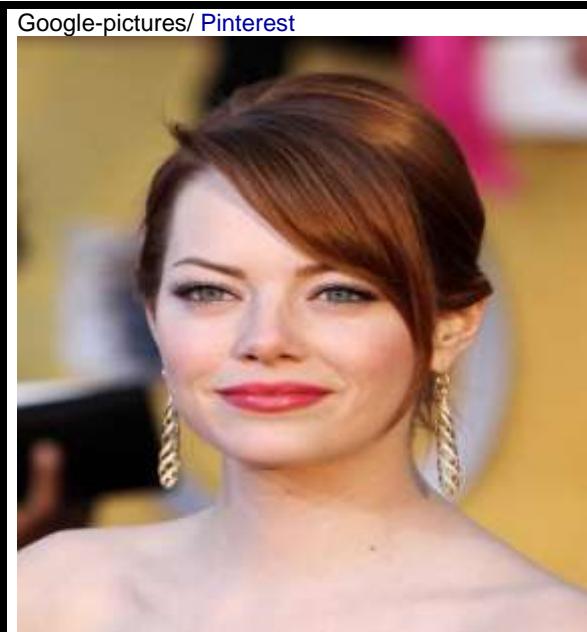
Figure 9-23: Wedding Hairstyle to a large face woman

Face Lines

Google-pictures/ ABG LAB

PERIORBITAL LINES
TEAR TROUGH
CHEEK AUGMENTATION
FACIAL CONTOURING
NASOLABIAL FOLDS
ORAL COMMISSURES

WORRY LINES
GLABELLAR LINES
PERIORAL LINES
SMILE LINES
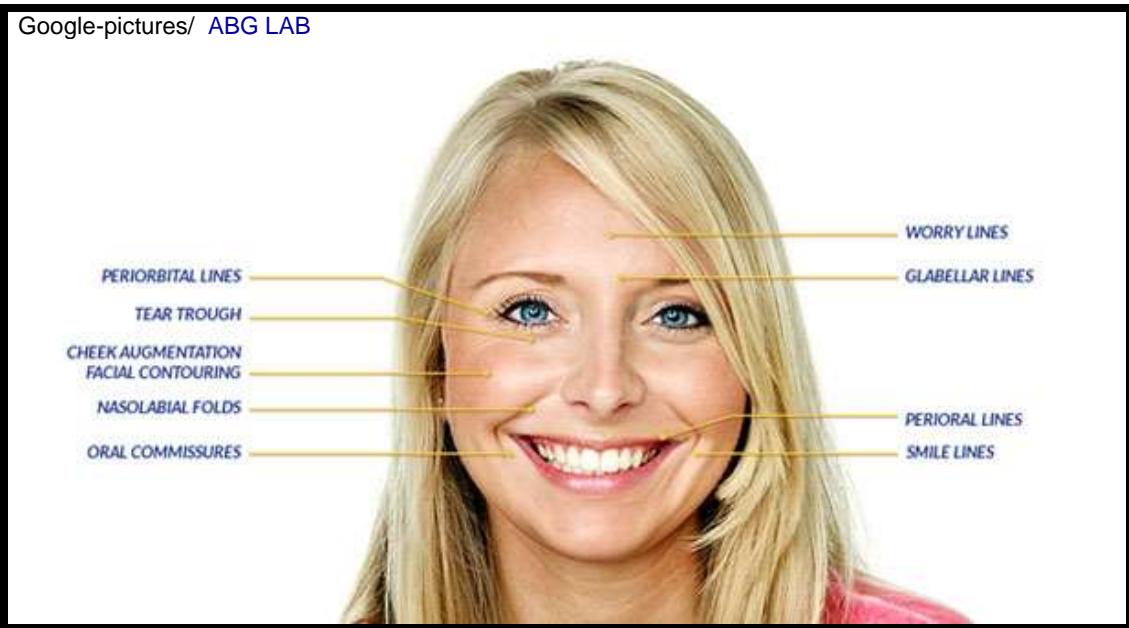
Figure 9-24: Common face lines are demonstrated at the above picture

As seen below picture, palpebral fissures are not the only and main characteristics, it is not specific or diagnostic. The Tatarian face as same, so, only chromosome analysis the only way of diagnosis, Trisomy 21.

Down's syndrome

Google-pictures/ Arquapetrarca
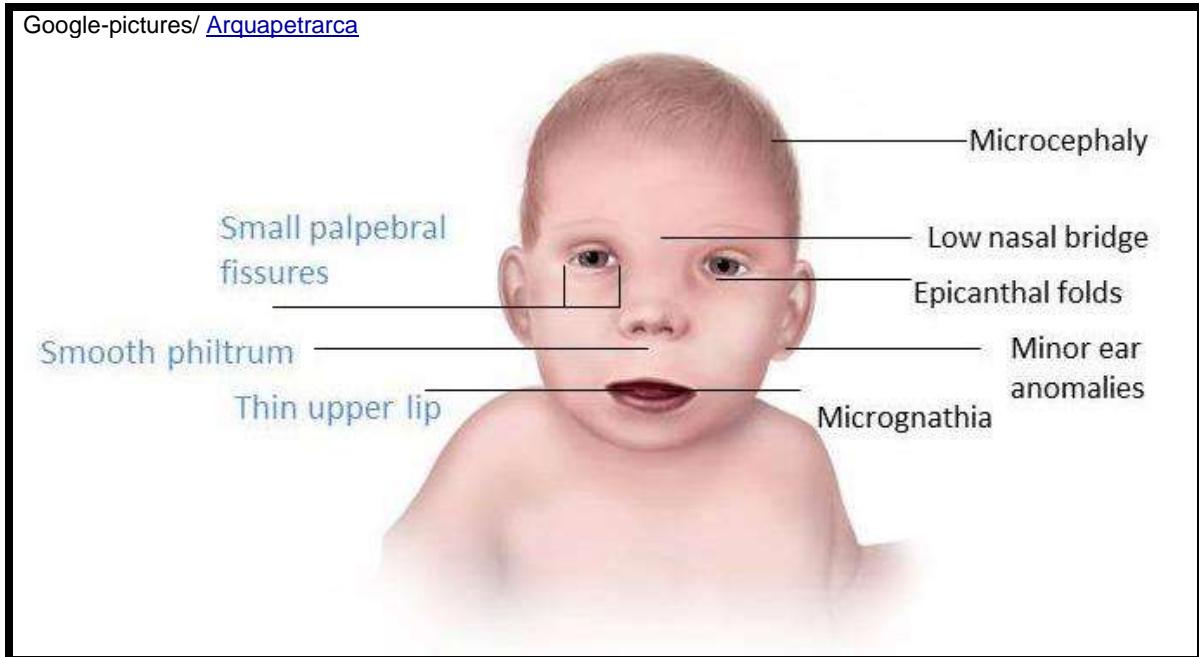
Figure 9-25: Common face appearance of Trisomy 21

Comment

Consider that, even eye slants, are also seen at some Tatarian people, even in Turkey, so, epicanthus is not indicates as Down's syndrome, thus, be a parameter, but, not a diagnostic parameter. So, this face figures must consider only as a characteristically finding, not diagnostic. The specifications as seen, can be seen at normal infants also. Not be quick to indicate them as abnormal finding, unless it is obvious.