SYNDROMES OF EAR, NOSE & THROAT

THESIS

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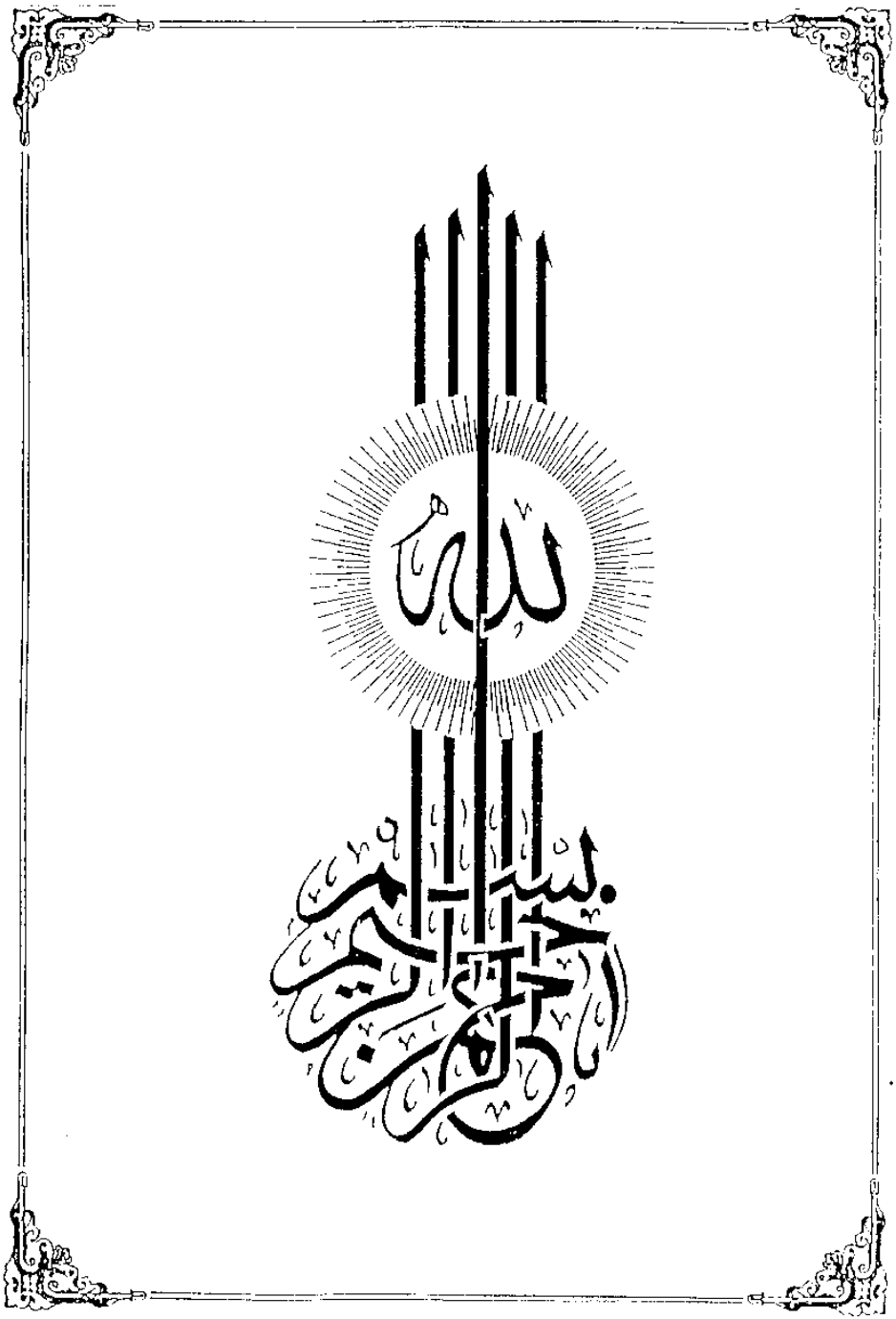

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Introduction

The advance in medicine in the last years have produced an enormous awareness of those factors make it possible for an investigator to recognize and delineate new syndromes.

With the discovery of new syndromes almost daily, the number of designations is expanding to the point where even specialists can no longer remember all the syndrome designations or their phenotypic features.

To cope with such confusion it is necessary that our system of classification be flexible and our nomenclature be weel understood.

Because different systems of classifications or nomenclature have been emplyed, a single syndrome may be known by several terms, thus causing confusion, for example:

Hypertelorism- hypospadias syndrome is also known Opitz syndrome and the BBB syndrome.

However when dealing with any a new syndrome there are two problems:

1. The morphology of a syndrome is not constant e.g. the features of the disorder may alter with time (in infant with the cri-due-chat syndrome, the mewing cry and the full round face disappear within the first year of life.
2. Not all patients with the syndrome possess the full picture also those who possess it may have not the same degree.

However a new syndrome may be introduced and labelled by following one of the following methods:

a) Eponym.

b) Mention one or more of the striking features.

c) Acronyms.

In general a new syndrome may be denoted by many ways:

1. **Eponym**

these method involve the name of the first person who discover the disorder (or the syndrome).

Sometimes 2 or more names are found in some eponyms and this may indicates:

a. Colloberation as in Smith-Lemli-Opitz syndrome.

b. Independent simultangeous discovery as in Moriqu-Branls Ford syndrome.

c. Revival by some one as in.

Pentz-Jeghers syndrome (revival by Jeghers).

Advantages:

- Avoid anatomic description (the syndrome can be expanded as a new features are recognized.
- Do not bias the phenotypic spectrum of future reports because there are no features of the syndrome in the designation.

Disadvantages:

- More than one syndrome may be named after the same individual. A single clinician may discovers . more then one syndrome.

- Frequently the individual for whom the syndrome was named is not the first to describe it.

2. *One or more of striking features.*

In this method we usually mention the name by which the syndrome can be remembered with ease.

Advantage:

They are descriptive term, thus they can aid the clinician in remembering some of the presumably prominent features of a given syndrome.

Disadvantages:

- the name may be too general and non specific.
- Some designations are simply incorrect.
- Introducing anatomic terminology into the designation biases the phenotypic spectrum of future reports.
- Anatomic designations do not allow for further syndrome delineation within the name itself.

3. Acronyms

Using the initials of the original patient surnames to form a name for the syndrome as VATER syndrome (V=Vertebral defect., A=Anal Atresia, T-E=Tracheoesophageal fistula, R= Renal Dysplasia).

Advantages:

- Readily lend themselves to computer diagnosis and information retrieval systems.
- Do not bias the phenotypic spectrum of future reports.

Disadvantage:

They have not been accepted by most workers in the field.

DEFINITIONS AND KEY WORDS

Malformation

A primary structural defect that results from a localized error of morphogenesis e.g cleft lip.

It is distinguised from deformation, an alternation in shape and or structure of a priviously normally formed part e.g torticollis.

Anomalad

A pattern of morphologic defects stemming from a single localized structural anomaly which resulted in a cascade of consequent defect. The ending ad is meant to imply more than one defect. e.g Robin anomalad.

Association

A recognized pattern of malformations which currently is not considered to constitue a syndrome or an anomalad, an osociation may be classified as a syndrome or as an anomalad, e.g hemihypertrophy with Wilms tumour.

Syndrome

A recognized pattern of malformation presumably having the same etiology and currently not interpreted as the consequence of a single localized error in morphogenesis e.g, Down syndrome.

Congenital Malformation

It is a malformation present at birt. The term congenital does not mean or exclude genetic etiology i.e. congenital malformations (disorders of morphogenesis may be due to genetic or non genetic factors.

Chromosomal Disorders:

It is present as the result of the developing organism's effort to cope with the duflicated or lost genes.

Autosomal Inheritance

Autosomal dominant: the dominant gene shows its effect shether in the homozygous or in the heterozygous state.
Autosomal recessive: the recessive gene is only evident when present in the homozygous state.

X-Linked Inheritance

X-linked dominant: the disorder is transmitted from an affected female to helf of her son and half of her daughters. The affected male transmit the disease to all of his daughters and non of his sons.
X-linked recessive: the genes are carried by females as well as males, but in affected persons are mostly males.

CLASSIFICATION

In fact classifying some hundreds of different types of syndromes is not an easy task. e.g. We can depend on:

A) **Aetiology:** Wheather it is known or unkown, further more it needs more explanation to show exactly where it lie in that big group. e.g. We can say it is genetic, but this is not enough as it may be enetic but

1. Autosomal dominant.
2. Autosomal recessive.
3. X-linked.
4. Chromosomal.
5. Genetic uncertain.

B) **Hereditory or non hereditary.**

C) **Stressing the facial feature:** adding to it the basic defects in other body sites e.g.

1. Facial plus limb anomalies with other associated defects.
2. Facial plus other associated defects.
3. Craniofacial with associated defects.
4. Craniofacial plus cardiac anomalies.
5. Craniofacial plus genital anomalies.
6. Craniofacial plus limb anomalies.
7. Craniosynostosis with other associated defect mainly involving the lims.
8. Cleft lipl palate with limb abnormalities and other associated defects.

8. Cleft lipl palate with limb abnormalities and other associated defects.
10. Eye.
11. Hair.
12. Meabolic.
13. Muscle.
14. Neurologic.
15 Hamartoses.
16. Osteochondrodysplasia.
17. Skin.
18. Pre-mature aging features with other associated defects.

D) Anatomical Classification:

E.g Nose: 1. Nares: Anteverted, asymmetric, dorsally tented, flared, narrow, notched, prominent, single, small, widely spaced.

2. Nasal bridge: broad, depressed, flat, high, low, prominent.

3. Nasal Columella: hypoplastic, rounded, short, small.

4. Nsal septum: below alae, deviated, edamatous, short, thick.

5. Nasal tip: dimpled, hanging, notched.

6 Nasal shape: beaking, broad, bulbous, flat, long, mutilated, pinched, prominent, short, small, thin, wide.

7. Skin lesion on nose: telengiectasia, haemangiomas.

Ear: External: Low-set, microtia, meatal atresia, cup-shaped, preauricular pits, thickened lobes. Middle: incudo-stapedial abnormalities.

However, we found that the easier way of classification to follow is :

I. Genetic Syndromes:

1. Autosomal dominent.
2. Autosomal recessive.
3. X-linked.
4. Chromosomal.
5. Genetic uncertain.

II. Syndromes of systemic diseases with relation to ENT:

1. Respiratory.
2. Muxculoskeletal.
3. Nervous system.
4. Renal.
5. Metabolic.
6. Eye.

III. Syndromes of cranial nerves:

1. Syndromes of trigeminal nerve.
2. Syndromes of other cranial nerves.

IV. Specific ENT syndromes.

V. Post. operative syndromes.

VI. Miscellaneous syndromes.

And to put it in A,B manner at the index.

PART I

GENCTIC SYNDROMES

AUTOSOMAL DOMINENT SYNDROME

* ACHONDROPLASIA.
* Acroosteolysis (arthrodento osseous dysplasia).
* Albright's syndrome (cherubism).
* Apert syndrome (acrocephalosyndactyly type I).
* Arthrogryposis with facial and limb anomalies.
* Blepharo naso facial syndrome.
* Branchio-oto-renal syndrome (FOR syndrome).
* Cleft lip and/or palate with paramedian pits of lower lip.
* Cleft palate-lateral synechiae syndrome (CPLS syndrome).
* Cledocranial dysplasia.
* Congenital contractural arachnodactyly syndrome.
* Cowden syndrome (multiple hamartoma syndrome).
* Craniofacial deafness syndrome.
* Craniometaphyseal dysplasia.*
* Cutis laxa.
* Cylindromatosis (Turban tumours).
* Ear pits-deafness syndrome.
* Ectrodactyly-ectodermal dysplasia-clefting (EEC) syndrome
* Ehlers-danlos syndrome
* Epithelioma adenoids cysticum (Brooke=Fordyce Trichoepitheliomas).
* Facioscapulohumeral muscular dystrophy.
* Familial benign cervical lipomatosis.
* Fetal face syndrome (Robinow syndrome).
* Freeman-sheldon syndrome (Whistling face syndrome)(Cranio carpotarsal dysplasia).

* Frontometaphyseal dysplasia.
* G syndrome (hypertelorism with esphegeal abnormality and hypospadias)(hyposfadias-dysphagia) syndrome.
* Gardner syndrome (intestinal polyposis III).
* Gingival fibromatosis, hypertrichosis, epilepsy, and mental retardation.
* Huntington drsease (rigid form).
* Hypertrichosis universalis congenita.
* Inability to open mouth fully, pseudo camptodactyly and short stoture.
* Infantile cortical hyperostosis (caftey disease).
* KBG syndrom (short stature, mental retardation, mecrodontia and unusual facies)
* Leopard syndrome (multiple lentigines syndrome).
* Marcus gunn and inverse marcus gunn syndromes (Jow-winking and winking-jow syndromes).
* Marfan Syndrome
* Multiple mucosal neuroma syndrome.
* Myotonic dystrophy (steinert disease).
* Nemaline myopathy.
* Neurofibromatosis (von recklinghausen disease).
* Nevoid basal cell carcinoma syndrome (Gorlin's nevoid basal cell carcinoma).
* Noonan syndrome.
* ODO syndrome (oculo dento osseous dysplasia).
* Osler-Rendu-Weber syndrom (hereditary hemorrhagic telangiectasia).