Comments on Terminology

Because of the confusion in terminology concerning the bone dysplasias, a sub-committee of the European Society of Pediatric Radiology met in Paris in 1969 and elaborated a nomenclature that divided the Constitutional (Intrinsic) diseases of bone into those with unknown and known pathogenesis. The unknown group were further divided into osteochondrodysplasias (abnormalities of cartilage and/or bone growth and development), dysostoses (malformation of individual bones, single or in combination), and other groups.
The osteochondrodysplasias were subdivided into three groups, the first of which were defects of growth of tubular bones and/or spine that manifested (A) at birth and (B) in later life.

This portion of the nomenclature is given below:

Constitutional Diseases of Bones with Unknown Pathogenesis.

Osteochondrodysplasias (abnormalities of cartilage and/or bone growth and development)

1. Defects of growth of tubular bones and/or spine.

A) Manifested at birth

1. Achondrogenesis
2. Thanatophoric dwarfism
3. Achondroplasia
4. Chondrodysplasia punctata (formerly stippled epiphyses) (several forms)
5. Metatropic dwarfism
6. Diastrophic dwarfism
7. Chondro-ectodermal dysplasia (ELLIS-VAN CREVELD)
8. Asphyxiating thoracic dysplasia (JEUNE)
9. Spondylo-epiphyseal dysplasia congenita
10. Mesomelic dwarfism: type NIEVERGELT; type LANGER
11. Cleido-cranial dysplasia (formerly cleido-cranial dysostosis).

B) Manifested in later life

1. Hypochondroplasia
2. Dyschondosteosis
3. Metaphyseal chondro-dysplasia type JANSEN

4. Metaphyseal chondro-dysplasia type SCHMID
5. Metaphyseal chondro-dysplasia type MCKUSICK
   (formerly cartilage-hair hypoplasia)
6. Metaphyseal chondro-dysplasia with malabsorption and neutropenia
7. Metaphyseal chondro-dysplasia with thymolymphopenia
8. Spondylo-metaphyseal dysplasia (KOZLOWSKI)
9. Multiple epiphyseal dysplasia (several forms)
10. Hereditary arthro-opthalmopathy
11. Pseudo-achondroplasic dysplasia
    (formerly spondylo-epiphyseal pseudo-achondroplasic dysplasia)
12. Spondylo-epiphyseal dysplasia tarda
13. Acrodysplasia
    a) Rhino-trico-phalangeal syndrome (GIEDION)
    b) Epiphyseal (THIEMANN)
    c) Epiphyso-metaphyseal (BRAILSFORD)

The nomenclature was revised in Paris at a further meeting in 1977 and the proposed first portion is given below:

1. Defects of growth of tubular bones and/or spine.

A) Identifiable at birth

1. Achondrogenesis type I (PARENTI-FRACCARO)
2. Achondrogenesis type II (LANGER-SALDINO)
3. Thanatophoric dysplasia
4. Thanatophoric dysplasia with Clover-leaf Skull
5. Short rib-polydactyly syndrome type I (SALDINO-NOONAN)
   (perhaps several forms)
6. Short rib-polydactyly syndrome type II (MAJEWSKI)
7. Chondrodystrophia punctata
   a) Rhizomelic type
   b) Dominant type
   c) Other types
   d) Exclude symptomatic stippling in other disorders
      (ZELLWEGER syndrome, Warfarin embryopathy and others)
8. Campomelic dysplasia
9. Other dysplasias with congenital bowing of long bones
10. Achondroplasia
11. Diastrophic dysplasia
12. Metatropic dysplasia (several forms)
13. Chondro-ectodermal dysplasia (ELLIS-VAN CREVELD)
14. Asphyxiating thoracic dysplasia (JEUNE)
15. Spondylo-epiphyseal dysplasia congenita (SPRANGER-WIEDEMANN)
16. Other spondylo-epiphyseal dysplasias recognizable at birth
17. KNIEST dysplasia
18. Mesomelic dysplasia
    a) type NIEVERGELT
    b) type LANGER (probable homozygous dyschondrosteosis)

c) type ROBINOW
d) type RHEINARDT
e) Others
19. Acromesomelic dysplasia
20. Cleido-cranial dysplasia
21. LARSEN syndrome
22. Oto-palato-digital syndrome

B) Identifiable in later life

1. Hypochondroplasia
2. Dyschondrosteosis
3. Metaphyseal chondrodysplasia type JANSEN
4. Metaphyseal chondrodysplasia type SCHMID
5. Metaphyseal chondrodysplasia type MCKUSICK
6. Metaphyseal chondrodysplasia with exocrine pancreatic insufficiency and cyclic neutropenia
7. Spondylo-metaphyseal dysplasia
   a) type KOZLOWSKI
   b) Other forms
8. Multiple epiphyseal dysplasia
   a) type FAIRBANK
   b) Others
9. Arthro-ophthalmopathy (STICKLER)
10. Pseudo-achondroplasia
    a) Dominant
    b) Recessive
11. Spondylo-epiphyseal tarda
12. Spondylo-epiphyseal dysplasia (other types)
13. DYGGVE-MELCHIOR-CLAUSEN dysplasia
14. Spondylo-epi-metaphyseal dysplasia (several types)
15. Myotonic chondrodysplasia (CATEL-SCHWARTZ-JAMPEL)
16. Parastremmatic dysplasia
17. Tricho-rhino-phalangeal dysplasia
18. Acrodysplasia with retinitis pigmentosa and nephropathy (SALDINO-MAINZER).

This book concerns itself only with the conditions in Group (A) i. e. those manifested at birth. It follows closely the updated nomenclature but conditions such as the oto-palato-digital syndrome have been omitted as their diagnosis may be largely clinical.

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