

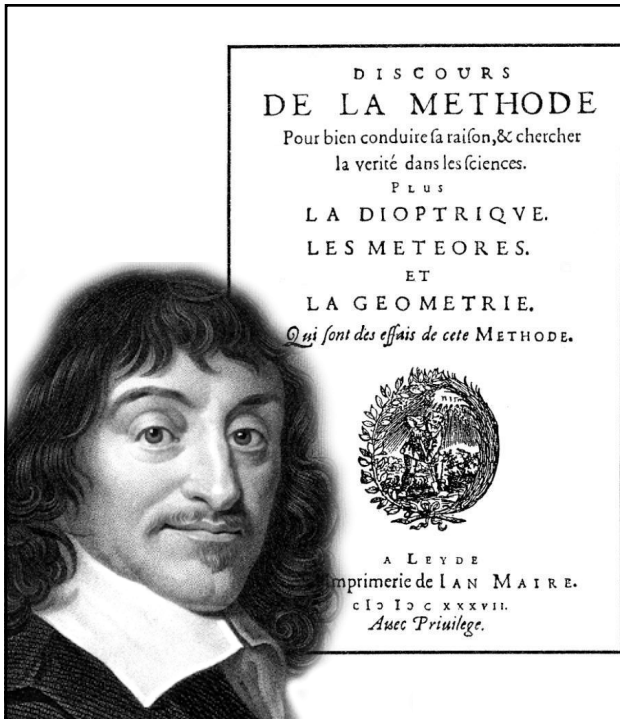
Pathologie dermatologique anténatale

Pierre VABRES
CHU Dijon Bourgogne

1



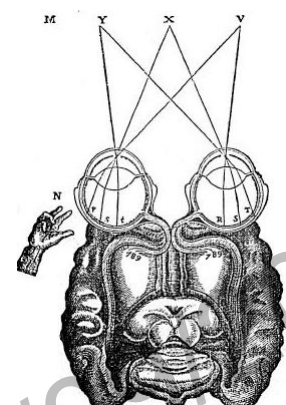
2



DISCOURS
DE LA METHODE
Pour bien conduire sa raison, & chercher
la vérité dans les sciences.
PLUS
LA DIOPTRIQUE.
LES METEORES.
ET
LA GEOMETRIE.
Qui sont des essais de cete METHODE.

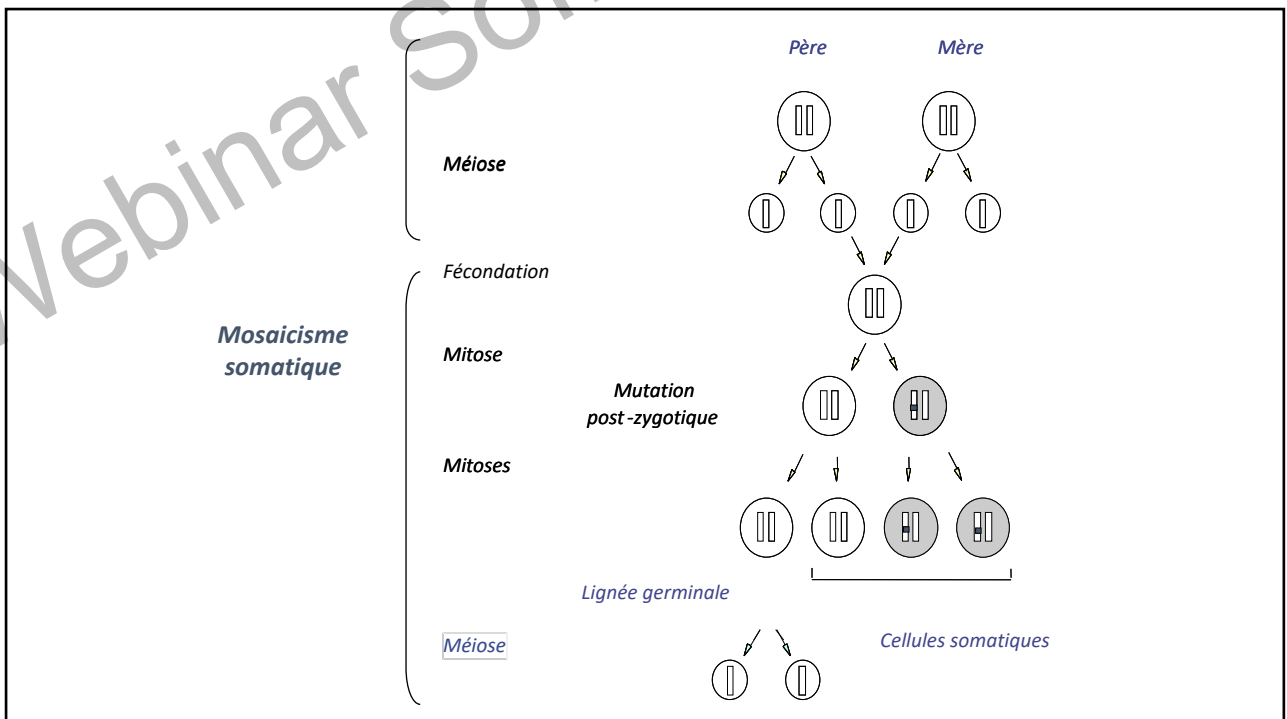
A LEYDE
Imprimerie de IAN MAIRE.
C 1637.
Avec Privilège.

“Même je pourrais, encore plus outre, vous montrer comment quelquefois elle peut passer de là par les artères d'une femme enceinte, jusques à quelque membre déterminé de l'enfant qu'elle porte en ses entrailles, et y former ces marques d'envie, qui causent tant d'admiration à tous les Doctes.”



René Descartes (1637) La Dioptrique. Cinquième discours “Des images qui se forment sur le fond de l’œil”

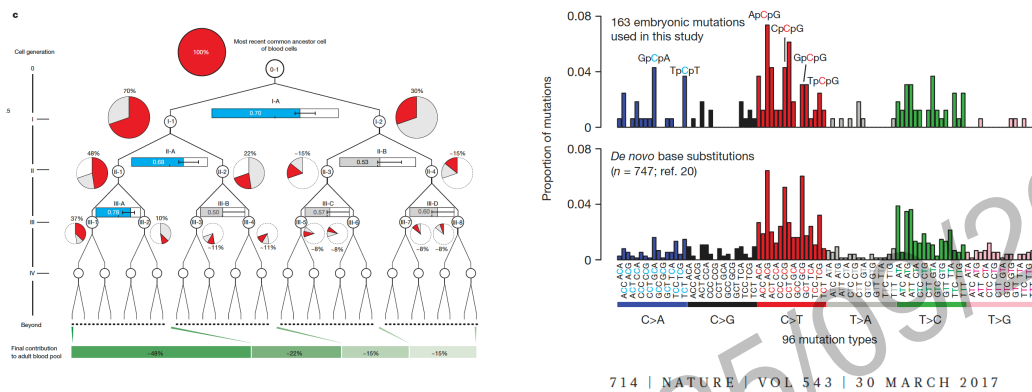
3



4

Somatic mutations reveal asymmetric cellular dynamics in the early human embryo

Young Seok Ju^{1,2}, Inigo Martincorena¹, Moritz Gerstung^{1,3}, Mia Petljak¹, Ludmil B. Alexandrov^{1,4}, Raheleh Rahbari⁵, David C. Wedge^{1,6}, Helen R. Davies¹, Manasa Ramakrishna¹, Anthony Fullam¹, Sancha Martin¹, Christopher Alder¹, Nikita Patel¹, Steve Gamble¹, Sarah O'Meara¹, Dilip D. Giri⁷, Torril Sauer⁸, Sarah E. Pinder⁹, Colin A. Purdie¹⁰, Åke Borg^{11,12,13}, Henk Stunnenberg¹⁴, Marc van de Vijver¹⁵, Benita K. T. Tan¹⁶, Carlos Caldas¹⁷, Andrew Tutt^{18,19}, Naoto T. Ueno²⁰, Laura J. van 't Veer²¹, John W. M. Martens²², Christos Sotiriou²³, Stian Knappskog^{24,25}, Paul N. Span²⁶, Sunil R. Lakhani^{27,28,29}, Jórunn Erla Eyfjörð³⁰, Anne-Lise Børresen-Dale^{31,32}, Andrea Richardson³³, Alastair M. Thompson³⁴, Alain Viari³⁵, Matthew E. Hurles⁵, Serena Nik-Zainal¹, Peter J. Campbell¹ & Michael R. Stratton¹



5

VIEWPOINT/ORIGINAL ARTICLE

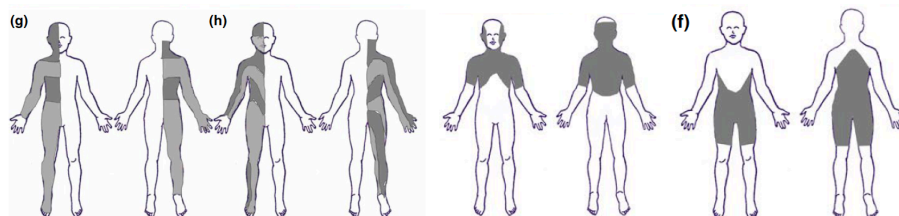
WILEY

The patterns of birthmarks suggest a novel population of melanocyte precursors arising around the time of gastrulation

Veronica A. Kinsler^{1,2} | Lionel Larue^{3,4,5}

Pigment Cell Melanoma Res. 2017;1-15.

wileyonlinelibrary.com/journal/pcmr | 1

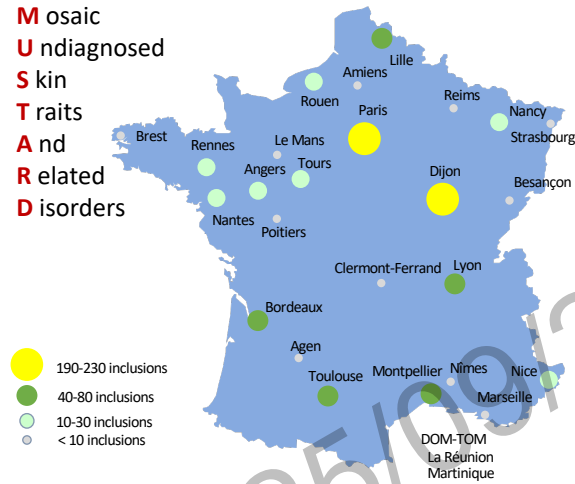


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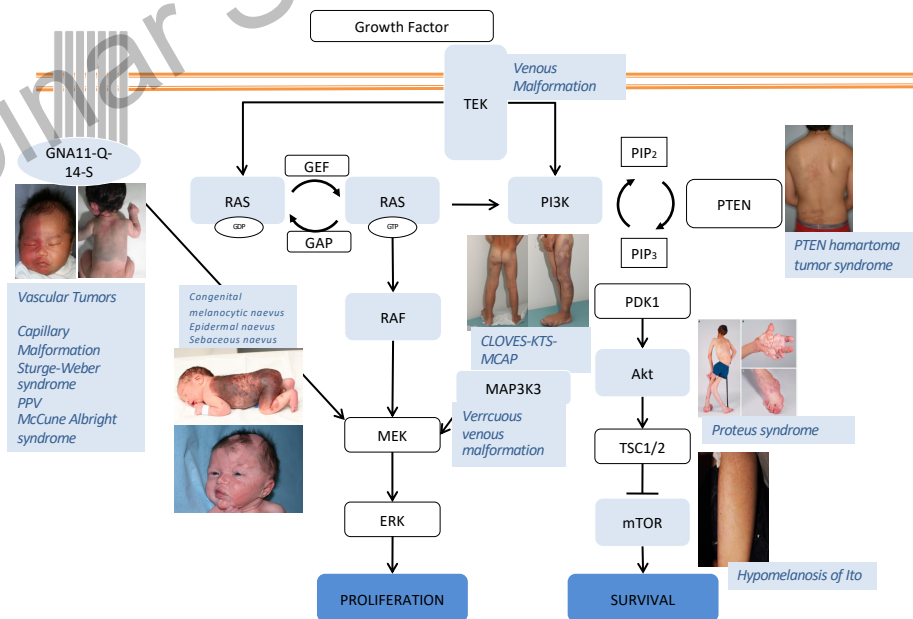
MAGEC-Mosaic centre / TRANSLAD federation (Dijon)

- FIMARAD national network
- Innovative diagnostic approach
- Innovative drug trials
- MUSTARD cohort:
 - 964 index cases
 - 2408 biologic specimens including 678 skin biopsies
- MUSTARD NCT01950975
- MOSAIC NCT01950975

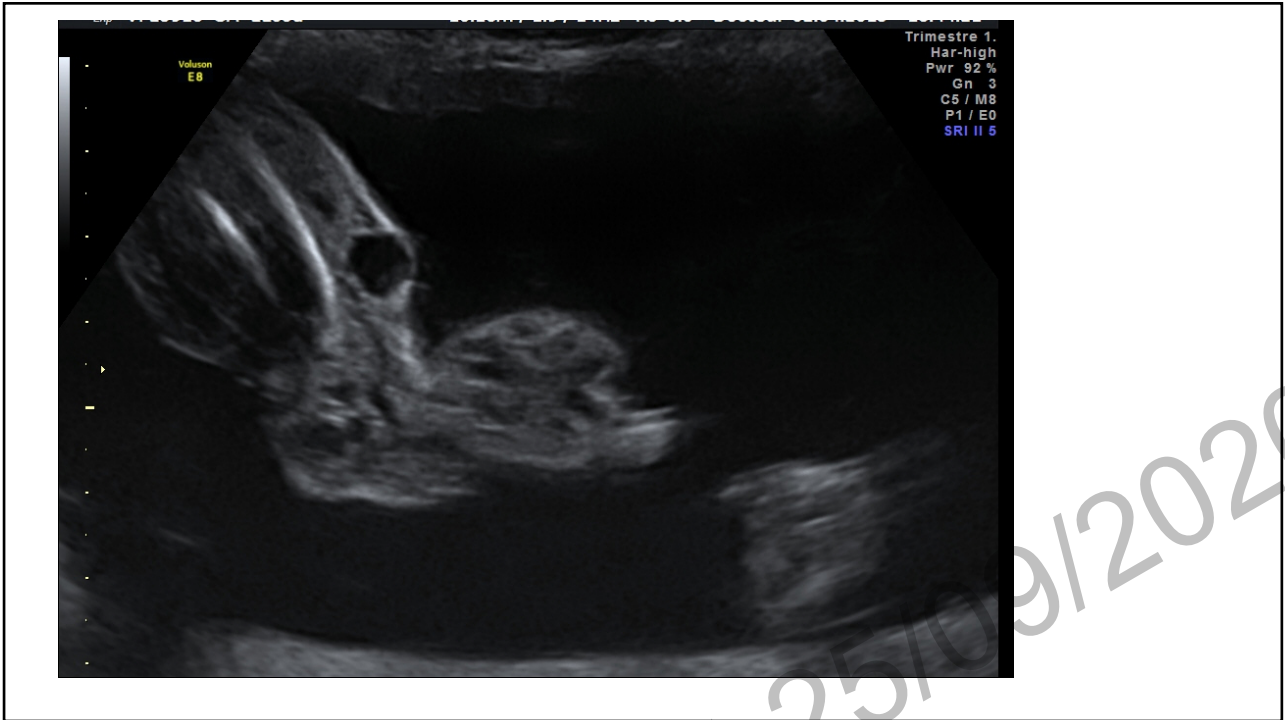
Mosaic
Undiagnosed
Skin
Traits
And
Related
Disorders



7



8



9



10

SCALP syndrome: Sebaceous nevus syndrome, CNS malformations, aplasia cutis congenita, limbal dermoid, and pigmented nevus (giant congenital melanocytic nevus) with neurocutaneous melanosis: A distinct syndromic entity

Joseph Lam, MD,^a Magdalene A. Dohil, MD,^b Lawrence F. Eichenfield, MD,^b and Bari B. Cunningham, MD^b
San Diego, California



11

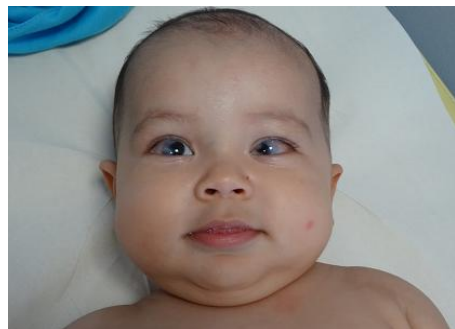
RAPID COMMUNICATION

AMERICAN JOURNAL OF **medical genetics** PART A

Oculoectodermal Syndrome is a Mosaic RASopathy Associated with *KRAS* Alterations

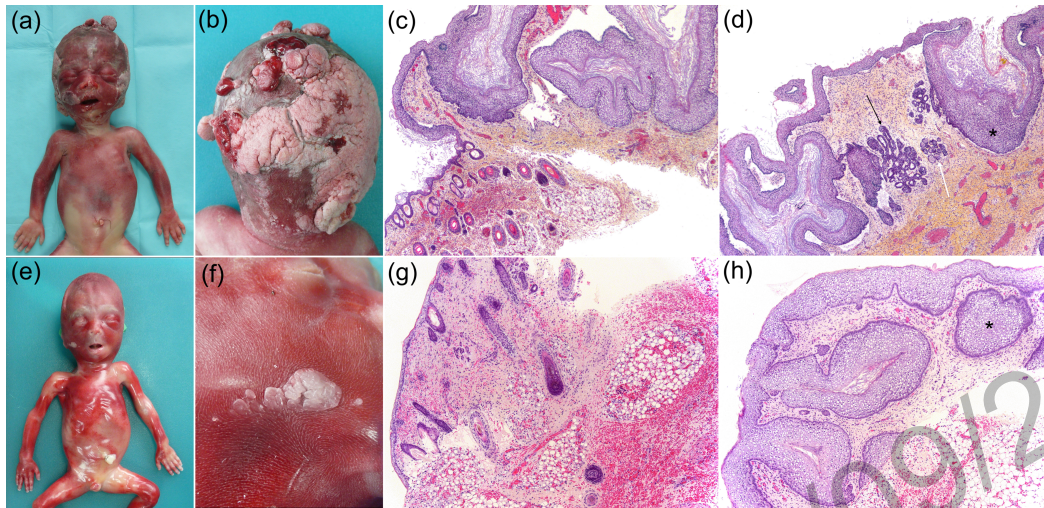


Courtesy of C. Chiaverini, Nice



12

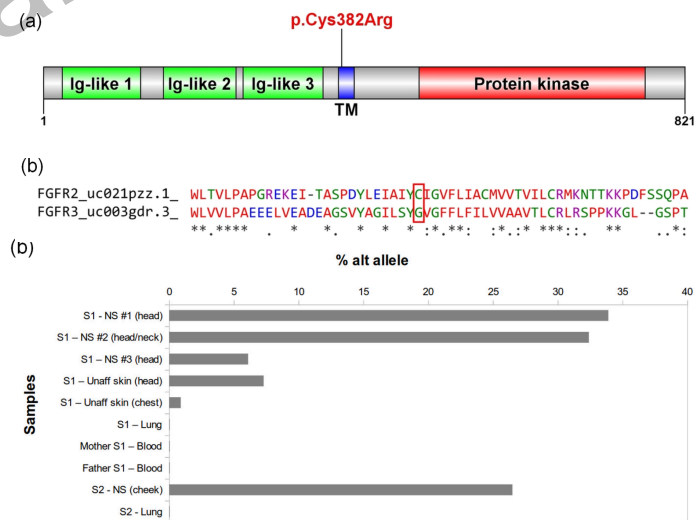
Papillomatous pedunculated sebaceous naevus



Kuentz et al. Br J Dermatol 2016

13

Postzygotic FGFR2 mutations in PPSN



Kuentz et al. Br J Dermatol 2016

14

Pediatric Dermatology Vol. 25 No. 3 355-358, 2008

Large, Papillomatous, Pedunculated Nevus Sebaceous: A New Phenotype

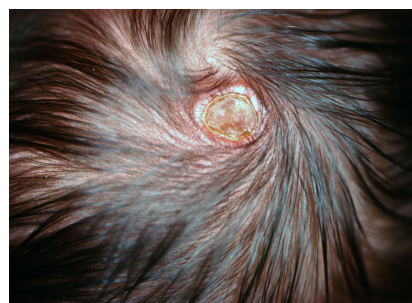
Dana Correale, M.D.,* Franziska Ringpfeil, M.D.,* and Maureen Rogers, FACP†

*Department of Dermatology and Cutaneous Biology, Thomas Jefferson University Hospital, Philadelphia, Pennsylvania, †Royal Alexandra Hospital for Children, Sydney, Australia



15

Scalp ACC in the neonate:
punched out ulceration / necrosis



16

Hair collar sign



17



18

ACC with sinus pericranii



19



20



21

Anterior + posterior midline ACC



22

Imperforate anus



23

Scalp ACC + cutis marmorata : Adams-Oliver



Dehdashtian A, Dehdashtian M. Pediatric Reports 2016; 8:6517

24



25



26

ACC + EB = Bart syndrome

- AD dystrophic EB +++
- AR dystrophic EB

Bullae absent
Variable nail involvement
Favourable outcome



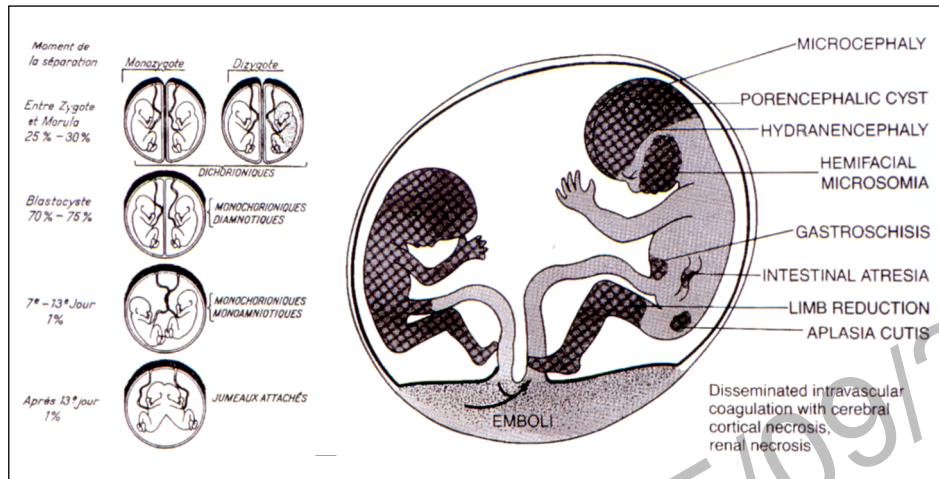
27

ACC with foetus papyraceus /placental ischemia



28

Fetus papyraceus / « vanishing twin »



29



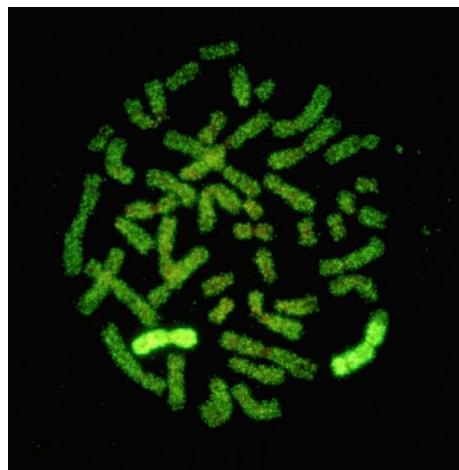
Courtesy of A. Maruani, MD, Tours

30



31

*M*icrophthalmia *D*ermal *A*plasia *S*clerocornea



Xp22 deletion (HCCS gene)

32



33



34

X-linked chondrodysplasia punctata (EBP)



35



36



37



38

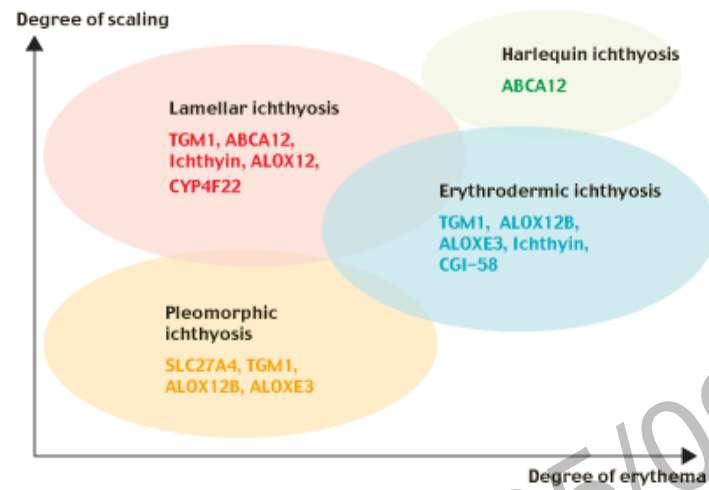


39



40

Gene panel for autosomal recessive congenital ichthyoses



<http://www.socialstyrelsen.se/rarediseases/ichthyosis>