

How to become a dysmorphologist

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1.

Passion

&

a little bit of talent

What is passion?



Paul A. O'Keefe is a social psychologist and a Professor of Organisational Behaviour at the University of Exeter Business School

His research focuses on how mindsets, interest, and passion influence motivation, goal pursuit, and achievement.

*Passion is defined as a **strong inclination** toward an activity that people like, that they find important, and in which **they invest time and energy**.*

What is passion?

A. Discovery theory



- *You discover something and immediately feel enthusiastic*
- *You can experience a strong emotional response*
- *You think: “This is really for me!” (sometimes you might realize it years later)*

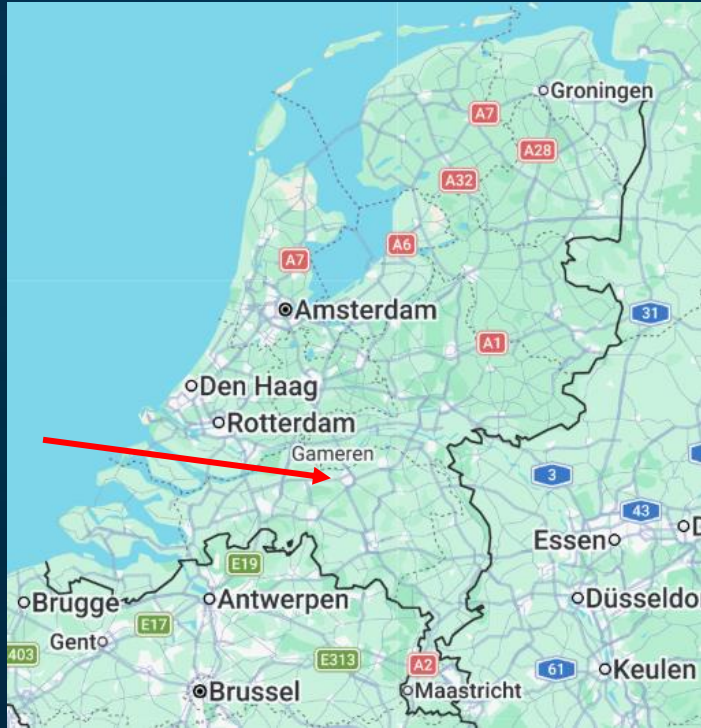
What is passion?

B. Developmental theory



- *You are curious about a certain topic*
- *Repeated exposure*
- *Positive experiences*
- *Growing competence*

Where I grew up



Gamarén:

- 2000 inhabitants
- Christian village
- One primary school



Arjan

- *Went to School with the Bible*
- *Liked animals, nature and cooking*
- *Wasn't very good at playing instruments*

Janey

- *Suffered from severe asphyxia*
- *Feeding problems*
- *Did not meet developmental milestones*
- *Attended special education*

Janey

- *Very happy and social child*
- *Loved all kinds of music: participated at marching band parade*
- *Short stature*
- *Learning problems persisted*



What is passion?

A. Discovery theory



- *You discover something and immediately feel enthusiastic*
- *You can experience a strong emotional response*
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Janey

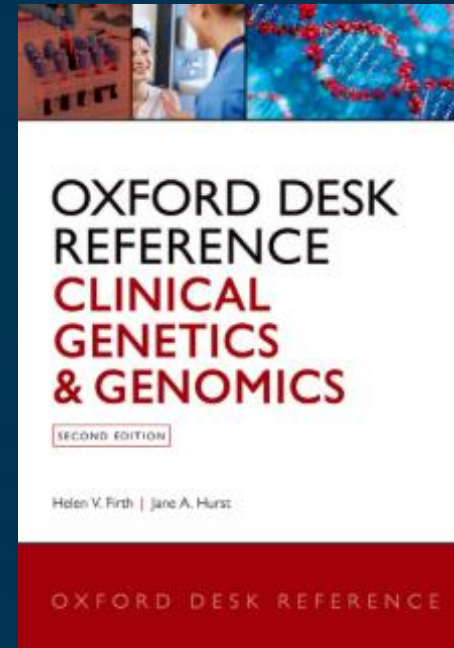
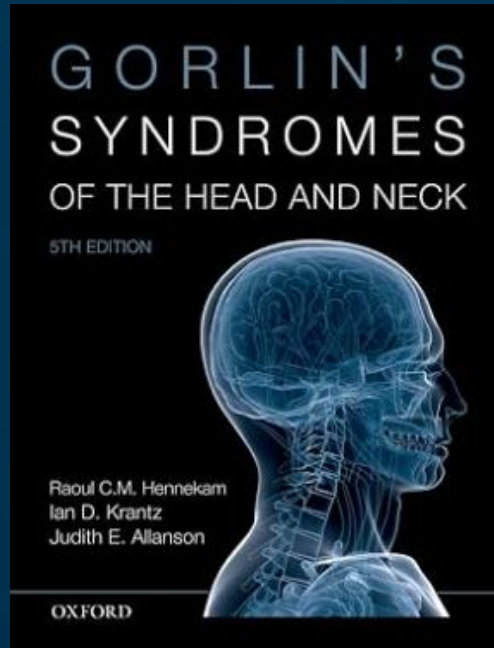
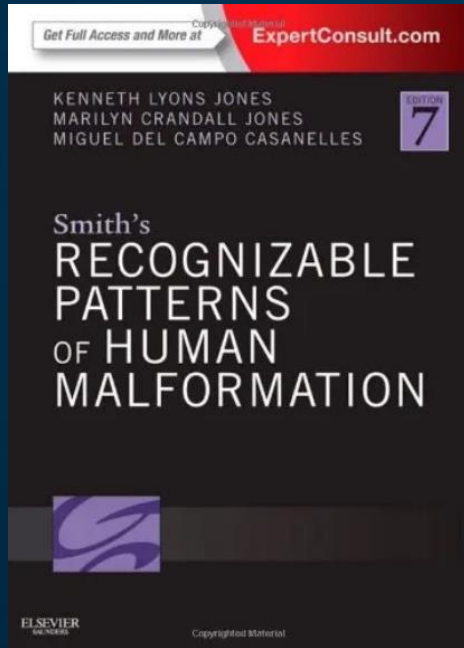
- *She lives on a care farm*
- *Works in a animal shop*
- *She is still very happy and sociable*
- *Early greying*



2.

Training

Training: books



Training: print and read Elements of Morphology

RESEARCH ARTICLE

AMERICAN JOURNAL OF
medical genetics PART
A

Elements of Morphology: Standard Terminology for the Head and Face

Judith E. Allanson,^{1*} Christopher Cunliffe,² H. Eugene Hoyne,³ Julie McGaughan,⁴
Max Muenke,⁵ and Giovanni Neri⁶



RESEARCH ARTICLE

AMERICAN JOURNAL OF
medical genetics PART
A

Elements of Morphology: Standard Terminology for the Ear

Alasdair Hunter,^{1*} Jaime L. Frias,² Gabriele Gillesen-Kaesbach,³ Helen Hughes,⁴
Kenneth Lyons Jones,⁵ and Louise Wilson⁶

Training: HPO-terms

<https://hpo.jax.org/>



Training: attend courses and meetings

MANCHESTER ESHG

SYNDROMOLOGY AND DYSMORPHOLOGY COURSE 2026

14–16 October 2026 • The Manchester Museum, Oxford Road, Manchester M13 9PL



I02

Dysmorphology 1

S. Douzgou Houge,
A. Bouman, P. Krawitz



Title: [SDG Autumn 2026 Meeting - Virtual](#)

Date: Thursday, 12 November, 2026 - 13:00

End Date: Friday, 13 November, 2026 - 09:00

Location: Virtual Meeting over Zoom

Training: write case reports

Contents lists available at ScienceDirect

 **ELSEVIER**

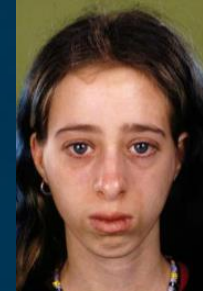
European Journal of Medical Genetics

journal homepage: www.elsevier.com/locate/ejmg



Bloom syndrome does not always present with sun-sensitive facial erythema

Arjan Bouman^{a,b}, Silvana van Koningsbruggen^a, M. Barış Karakullukcu^c, Willem Hans Schreuder^c,
Phillis Lakeman^{a,*}



CLINICAL REPORT

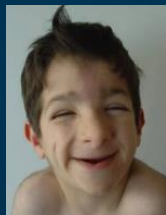
AMERICAN JOURNAL OF 
medical genetics

Trisomy 4 Mosaicism: Delineation of the Phenotype

Arjan Bouman,¹ Anne-Marie van der Kevie-Kersemaekers,¹ Karin Huijsdens-van Amsterdam,¹
Nordin Dahhan,² Lia Knecht,¹ Fleur Vansenne,³ and Jan Maarten Cobben^{2*}

¹Department of Clinical Genetics, Academic Medical Center, Amsterdam, The Netherlands

Training: patients, patients, patients



Training: be playful and kind



Training: counseling





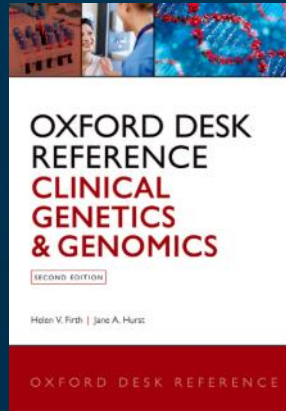
2.

Training

Let's practise!

Training

- A. Try to recognize hallmark features of a syndrome or group of syndromes
- B. **Always** make a differential diagnosis based on specific clinical features



Training

Clinical feature 1:

Fifth finger clinodactyly

- A) Silver-Russell syndrome
- B) Coffin-Siris syndrome
- C) Cornelia de Lange syndrome
- D) All of the above



Training

Clinical feature 1:

Fifth finger clinodactyly

- A) Silver-Russell syndrome
- B) Coffin-Siris syndrome
- C) Cornelia de Lange syndrome
- D) All of the above



Training

Clinical feature 2:

Synophrys

- A) Wiedemann-Steiner syndrome
- B) KBG syndrome
- C) Cornelia de Lange syndrome
- D) All of the above



Training

Clinical feature 2:

Synophrys

- A) Wiedemann-Steiner syndrome
- B) KBG syndrome
- C) Cornelia de Lange syndrome
- D) All of the above



Training

Clinical feature 3:

Synophrys + macrodontia upper incisors

- A) Wiedemann-Steiner syndrome
- B) KBG syndrome
- C) Cornelia de Lange syndrome
- D) All of the above



Training

Clinical feature 3:

Synophrys + macrodontia upper incisors

- A) Wiedemann-Steiner syndrome
- B) KBG syndrome
- C) Cornelia de Lange syndrome
- D) All of the above



Training

Clinical feature 4:

Sparse temporal hair

- A) Costello syndrome
- B) Pallister-Killian syndrome
- C) Kabuki syndrome
- D) All of the above



Training

Clinical feature 4:

Sparse temporal hair

- A) Costello syndrome
- B) **Pallister-Killian syndrome**
- C) Kabuki syndrome
- D) All of the above



Training

Clinical feature 5:

Hairy elbows

- A) Cantu syndrome
- B) Wiedemann-Steiner syndrome
- C) Cornelia de Lange syndrome
- D) All of the above



Training

Clinical feature 5:

Hairy elbows

- A) Cantu syndrome
- B) Wiedemann-Steiner syndrome
- C) Cornelia de Lange syndrome
- D) All of the above

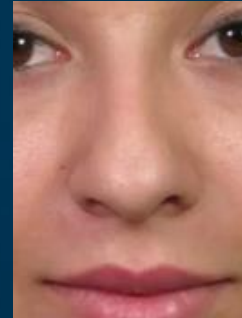


Training

Clinical feature 6:

Broad nasal tip

- A) Hyper-IgE syndrome
- B) Floating Harbor syndrome
- C) Tricho-rhino-phalangeal syndrome
- D) All of the above

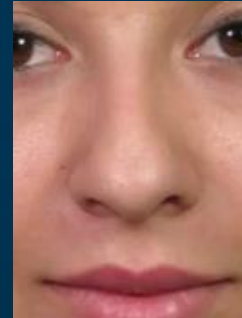


Training

Clinical feature 6:

Broad nasal tip

- A) Hyper-IgE syndrome
- B) Floating Harbor syndrome
- C) Tricho-rhino-phalangeal syndrome
- D) All of the above



Training

Clinical feature 7:

Broad nasal tip + dystrophic nails

- A) Hyper-IgE syndrome
- B) Floating Harbor syndrome
- C) Tricho-rhino-phalangeal syndrome
- D) All of the above



Training

Clinical feature 7:

Broad nasal tip + dystrophic nails

- A) Hyper-IgE syndrome
- B) Floating Harbor syndrome
- C) Tricho-rhino-phalangeal syndrome
- D) All of the above



Training

Clinical feature 8:

Acro-osteolysis

- A) Osteogenesis imperfecta
- B) Achondroplasia
- C) Hajdu-Cheney syndrome
- D) All of the above



Training

Clinical feature 8:

Acro-osteolysis

- A) Osteogenesis imperfecta
- B) Achondroplasia
- C) Hajdu-Cheney syndrome
- D) All of the above



Training

Clinical feature 9:

Long toes

- A) Marfan syndrome
- B) Loeys-Dietz syndrome
- C) 22q11 microdeletion syndrome
- D) All of the above



Training

Clinical feature 9:

Long toes

- A) Marfan syndrome
- B) Loeys-Dietz syndrome
- C) 22q11 microdeletion syndrome
- D) All of the above

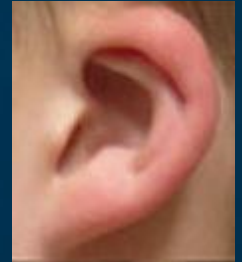


Training

Clinical feature 10:

Long toes + abnormally shaped ear

- A) Marfan syndrome
- B) Loeys-Dietz syndrome
- C) 22q11 microdeletion syndrome
- D) All of the above

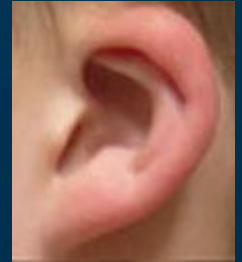


Training

Clinical feature 10:

Long toes + abnormally shaped ear

- A) Marfan syndrome
- B) Loeys-Dietz syndrome
- C) 22q11 microdeletion syndrome
- D) All of the above



questions





Dysmorphology Interactive Session
Gothenburg, June 2026

Case/cohort

Known/Unknown

Your name

Your role

Your email contact





ESHG2026

EUROPEAN HUMAN GENETICS CONFERENCE

June 13-16 | Svenska Mässan, Gothenburg | Sweden



Declaration of conflict of interest

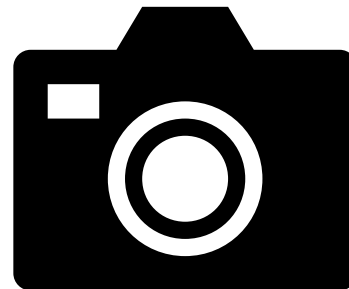
Type	Company
Employment full time / part time	Company name/None
Research Grant (P.I., collaborator or consultant; pending and received grants)	Company name/None
Other research support	Company name/None
Speakers Bureau / Honoraria	Company name/None
Ownership interest (stock, stock-options, patent or intellectual property)	Company name/None
Consultant / advisory board	Company name/None

Clinical presentation and Photographs/Video

1. Summarise the issues/reasons for/at referral/presentation
2. General status about major fields of development:
age/growth/developmental milestones/malformations/morbidity/behaviour
3. Important to include photographs already on the 1st slide.
Describe/illustrate craniofacial and other dysmorphisms using/through the
photographs/videos and avoid repeating them as a written list

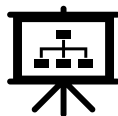
(example) 6 year old boy

- Short stature (height on 3rd percentile)
- Congenital, bilateral hearing loss
- Mild-to-moderate, global developmental delay
- Ataxia
-



Relevant Family tree/history:

-

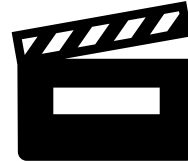
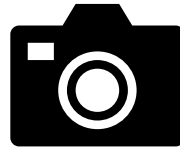


Clinical history and Imaging/Video

1. Summarise the clinical history of this individual i.e. prenatal exposures/feeding/any progressive symptoms/changes
2. Show pictures of relevant imaging studies i.e. MR brain

(example) 6 year old boy

- Epilepsy onset...
- Medications..
- Operated for..
- Tendency to form cheloids..
- Sociable behavior with a shift towards self-aggression around ..
-

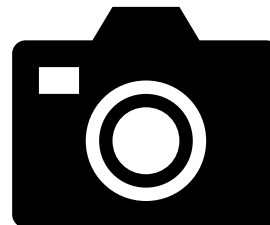


Laboratory investigations (and a clinical photograph)

1. When showing the genetic investigations, it is **necessary** to include platforms/methods used
2. When showing genetic variants, it is **necessary** to use the HUGO standards
3. When showing pathogenicity interpretations, it is **necessary** to use ACMG or ABC standards
4. Include all relevant laboratory investigations, not just genetic testing

(example)

- Micro-array (SNP): Normale female pattern
- Clinical exome, trio: XYZ gene:, heterozygous, de novo, disease-causing variant
- Re-analysis...
- Other relevant investigations performed:
-

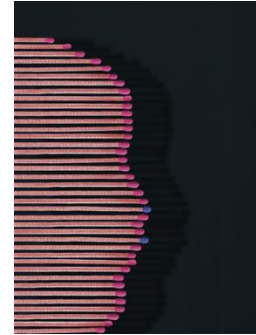


Differential diagnosis and GestaltMatcher analysis

1. Which genetic or other conditions did you consider when designing the approach to investigations/follow-up/treatment/family counselling
2. If you considered other dysmorphic genetic conditions, show why/how do the features compare with the ones of this individual i.e. show a comparison to published facial photographs/list of features
3. Presentations that include GestaltMatcher analysis will be prioritised

(example)

- XYZ (genetic) syndrome
- Prenatal exposure to...
- Multiple conditions
- ...



Summary/Conclusion

(example)

- A 6 year old boy with XXX/unknown syndrome confirmed by/normal genetic investigations...
- Genetic Variant of possible significance...
- Take home message.....
- ...

1. *Why did you choose to present this case?*
2. *How can this workshop be helpful in this case?*
3. *Any learning points?*

