Scientific Research Writing for Publication

Professor Hanan Hamamy

Why is writing and publishing important???

- Communicate knowledge to others
- Self satisfaction
- Career development/promotion
- Influence policy & practice
- Enjoy writing
- Establish scientific identity/contribution
- Part of your job/degree training

Your views

- Have you published/attempted to publish scientific work previously?
- If yes, how have you found the process?
- Are you intending to write up your project for publication?
- If yes, why?
- If no, Why not?

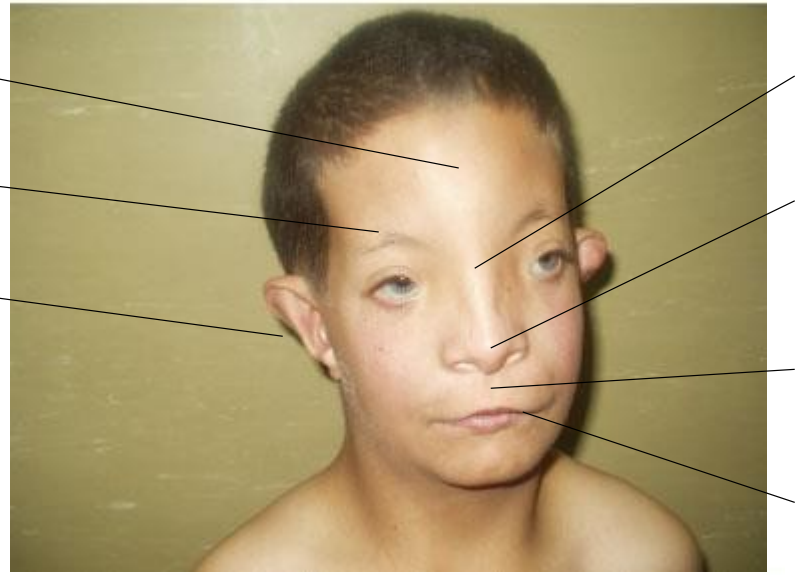
Novel syndrome: AR or XR?

Midface prominence

Sparse lateral eyebrows

Protruding and malformed ears

Absence / dysfunction of lacrimal structures

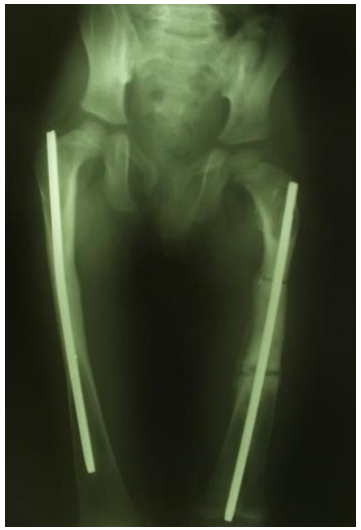


Hypertelorism

Pointed nasal tip, anteverted nostrils

Flat philtrum

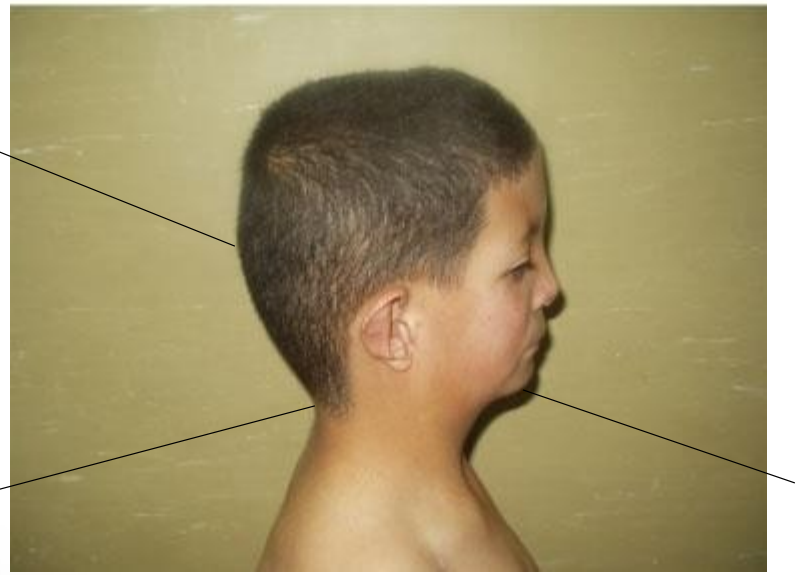
Thin upper vermilion border



Brachycephaly

pleasant, joyful
Learning difficulties

Low posterior hair line

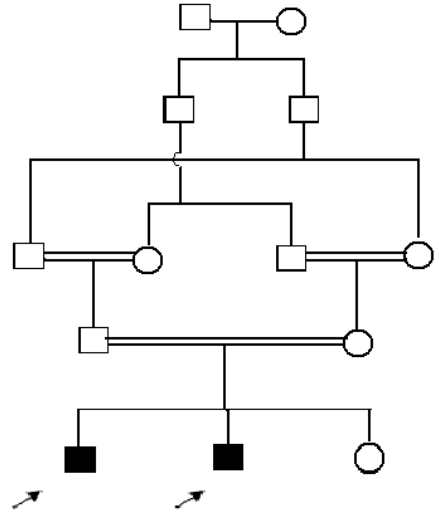


Hypodontia

High myopia

Mild hearing impairment

Mild micrognathia



10 years old patient (full face and lateral view)

American Journal of Medical Genetics Part A 143A:229–234 (2007)

New Syndrome

Severe Hypertelorism, Midface Prominence, Prominent/Simple Ears, Severe Myopia, Borderline Intelligence, and Bone Fragility in Two Brothers: New Syndrome?

Hanan A. Hamamy,[1] Ahmad S. Teebi,[2]* Kamaldine Oudjhane,[3] N.N. Shegem,[1] and K.M. Ajlouni[1]

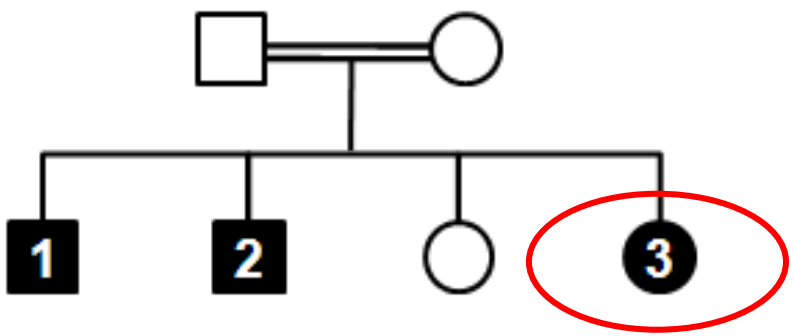
[1]National Center for Diabetes, Endocrinology and Genetics, Amman, Jordan
[2]Department of Pediatrics, Weill Cornell Medical College in Qatar, Doha, Qatar
[3]Department of Diagnostic Imaging, The Hospital for Sick Children, Toronto, Canada

Received 28 July 2006; Accepted 23 October 2006

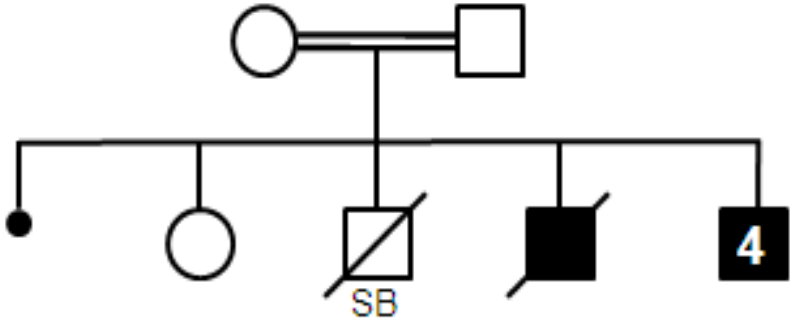
We report on two brothers, born to double first cousin Jordanian Arab parents, with a syndrome comprising severe hypertelorism with upslanted palpebral fissures, brachycephaly, abnormal ears, sloping shoulders, enamel hypoplasia, and osteopenia with repeated fractures. Both have severe arrangements. The father has mild hypertelorism but the family history is otherwise unremarkable. We think that this represents a previously unrecognized autosomal or X-linked recessive syndrome. © 2007 Wiley-Liss, Inc.

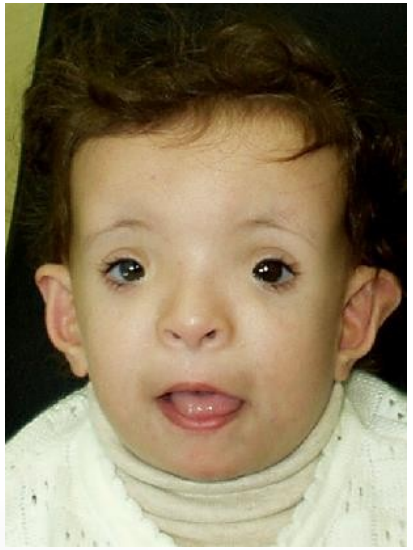
Genetic studies initiated

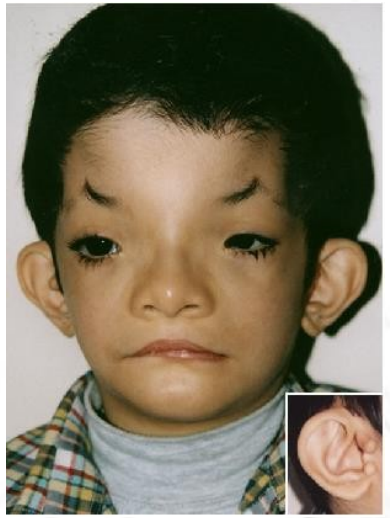
Jordanian family



Turkish family







nature genetics

Nat Genet. 2012 May 13. doi: 10.1038/ng.2259. [Epub ahead of print]

Mutations in *IRX5* impair craniofacial development and germ cell migration via SDF1

Carine Bonnard[1], Anna C Strobl[2], Mohammad Shboul[1], Hane Lee[3], Barry Merriman[3], Stanley F Nelson[3], Osama H Ababneh[4], Elif Uz[5,6], Tülay Güran[7], Hülya Kayserili[8], Hanan Hamamy[9,10] & Bruno Reversade[1,11]

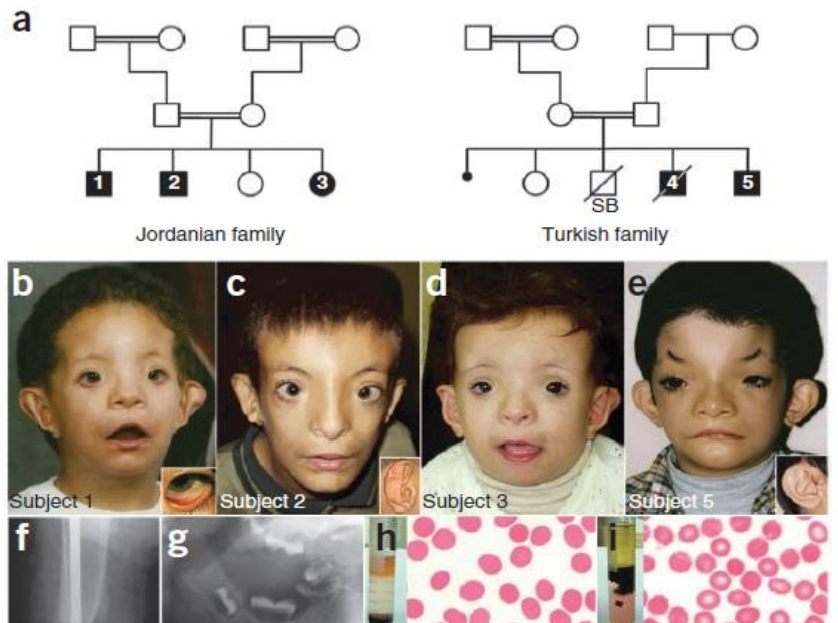


Figure 1 Clinical and genetic findings in five probands diagnosed with Hamamy syndrome. (**a**) Pedigrees of two inbred families from Jordan and Turkey. Filled black symbols, affected individuals (1–5). Crossed symbols, individual deceased. Small black circle, miscarriage (sex unspecified). SB, stillborn. (**b–e**) Full facial photographs of affected individuals with common craniofacial dysmorphisms, including midface prominence, sparse lateral eyebrows, severe telecanthus, agenesis of lacrimal punctuae (**b**, inset), anteverted nostrils, pointed nasal tip, flat philtrum, thin upper vermillion border and protruding ears (**c**,**e**, inset). Participants gave consent for publication of all photos. (**f**) Femoral radiograph of subject 5 showing multiple fractures and corrective osteotomy. (**g**) Panoramic radiograph of maxilla and mandible of subject 5 showing loss of lamina dura and teeth malocclusion. (**h**,**i**) Blood cell separation by density-gradient centrifugation and blood smear of mother of subject 5 (**h**) and subject 5 (**i**) show hypochromic microcytic anemia. Scale bar, 10 μm. (**j**) Electrocardiogram of subjects 1–3 showing left intraventricular conduction delay (QRS complex >100 ms). V1, fourth intercostal right chest lead. (**k**) Homozygosity mapping delineated a single candidate locus encompassing 73 genes on chromosome 16q12.2-q21 (Chr. 16). Locus capture followed by massive parallel resequencing in subject 1 identified a single biallelic base pair change (c.498C>A) in the *IRX5* gene, leading

Consanguinity research project
Department of Genetic medicine and Development
Geneva University

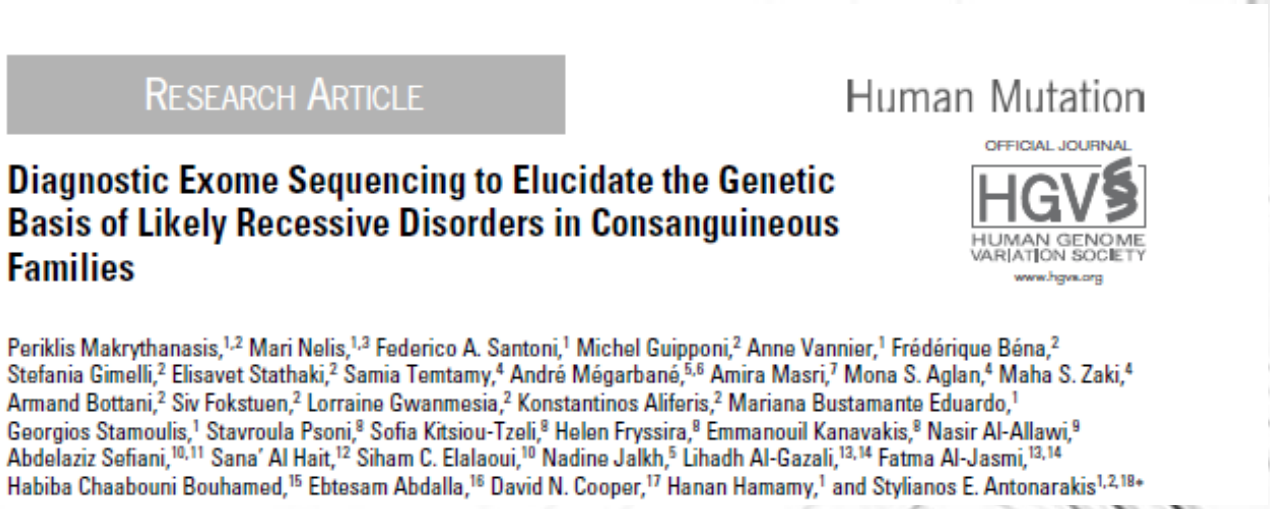
- Inclusion criteria
  - parents are consanguineous
  - having at least 2 affected children
  - no diagnosis
  - Any Phenotype

- Total number of families : 51
- 39 with children having syndromic or non-syndromic intellectual disability/developmental delay

Samples were received from our collaborators in Egypt 20, Jordan 17, Lebanon 4, Greece 4, Iraq 2, Morocco 1, UAE 1, Tunisia 1, Switzerland 1

Results

- ✓The putatively pathogenic homozygous variant was found in known disease-causing genes in 18 families by exome sequencing and one family by array CGH. The diagnostic yield was 37%.
- ✓In 26 families (51%) candidate genes were identified
- ✓In only 5 families no diagnosis or candidates were found



RESEARCH ARTICLE

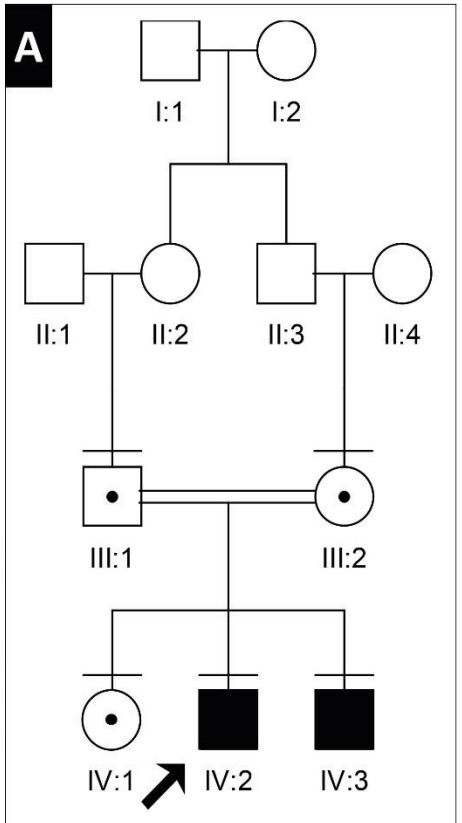
Human Mutation

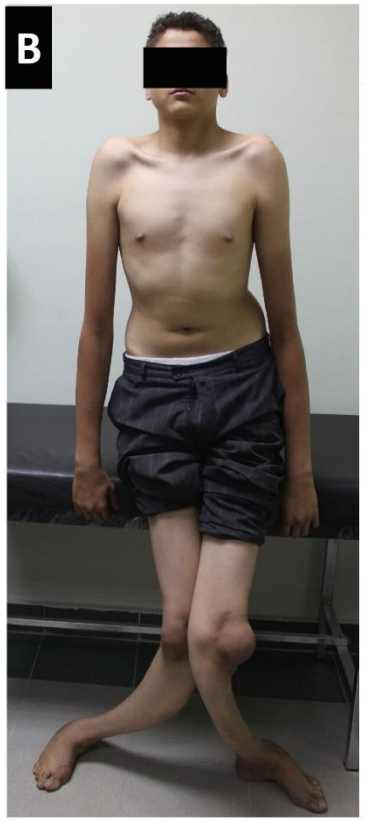
OFFICIAL JOURNAL HGVS HUMAN GENOME VARIATION SOCIETY www.hgvs.org

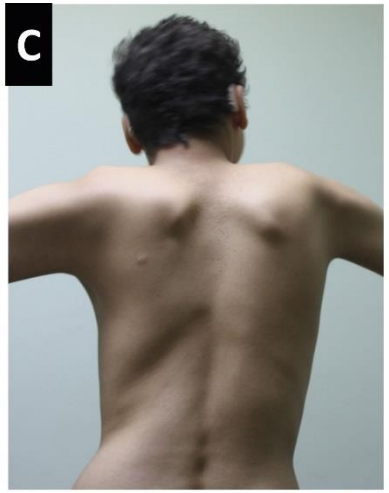
Diagnostic Exome Sequencing to Elucidate the Genetic Basis of Likely Recessive Disorders in Consanguineous Families

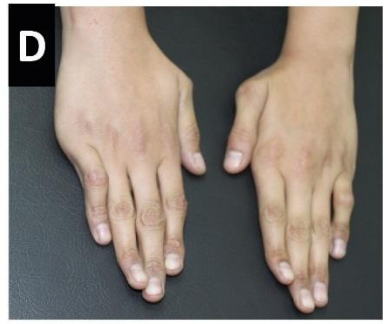
Periklis Makrythanasis,[1,2] Mari Nelis,[1,3] Federico A. Santoni,[1] Michel Guipponi,[2] Anne Vannier,[1] Frédérique Béna,[2] Stefania Gimelli,[2] Elisavet Stathaki,[2] Samia Temtamy,[4] André Mégarbané,[5,6] Amira Masri,[7] Mona S. Aglan,[4] Maha S. Zaki,[4] Armand Bottani,[2] Siv Fokstuen,[2] Lorraine Gwanmesia,[2] Konstantinos Aliferis,[2] Mariana Bustamante Eduardo,[1] Georgios Stamoulis,[1] Stavroula Psoni,[8] Sofia Kitsiou-Tzeli,[8] Helen Fryssira,[8] Emmanouil Kanavakis,[8] Nasir Al-Allawi,[9] Abdelaziz Sefiani,[10,11] Sana' Al Hait,[12] Siham C. Elalaoui,[10] Nadine Jalkh,[5] Lihadh Al-Gazali,[13,14] Fatma Al-Jasmi,[13,14] Habiba Chaabouni Bouhamed,[15] Ebtesam Abdalla,[16] David N. Cooper,[17] Hanan Hamamy,[1] and Stylianos E. Antonarakis[1,2,18]*

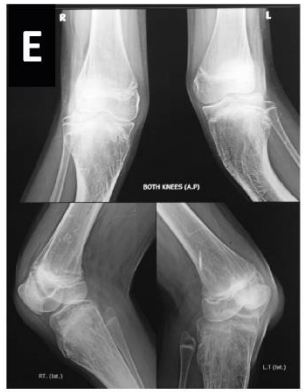
Tall stature, severe lateral tibial deviation, scoliosis, hearing impairment, camptodactyly and arachnodactyly

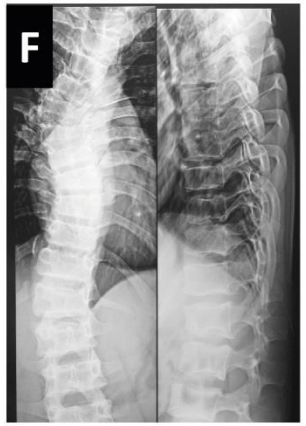


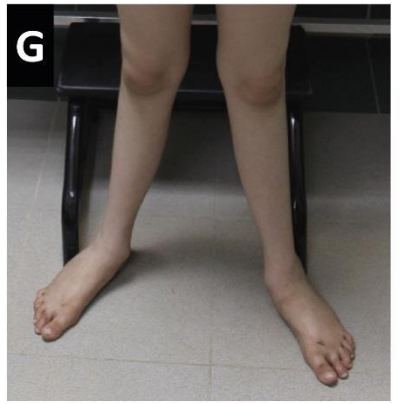


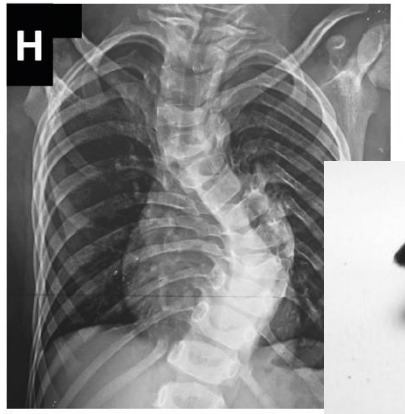






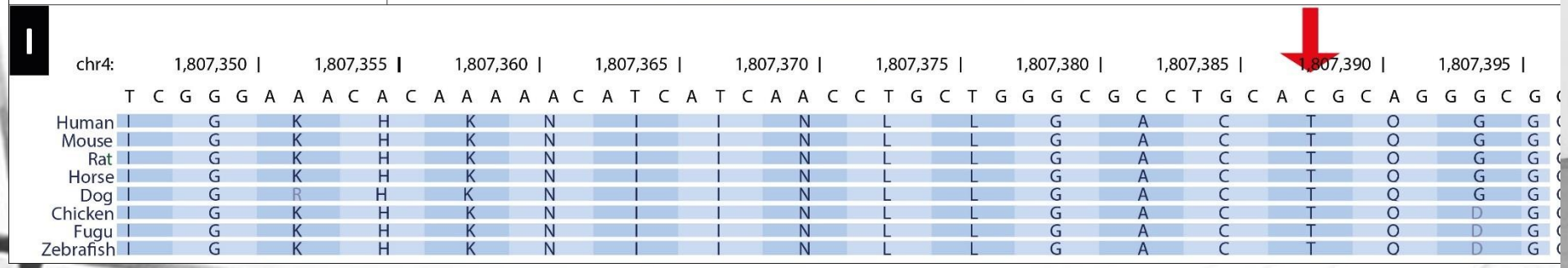






A novel homozygous mutation in *FGFR3 gene*

Spider lamb syndrome



HUMAN MUTATION, Vol. 35, No. 8, 959–963, 2014

J Med Screen
20(4) 171–176

DOI: 10.1177/0969141313508105
msc.sagepub.com
SAGE

The first five years of a preventive programme for haemoglobinopathies in Northeastern Iraq

Nasir AS Al-Allawi[1], Sana D Jalal[2], Najmaddin H Ahmed[3], Azad H Faraj[3], Awaz Shalli[2] and Hanan Hamamy[4]

Abstract

Objective: To evaluate the feasibility and effectiveness of a preventive programme for haemoglobinopathies in a single centre in Northeastern Iraq.

Methods: Premarital screening, genetic counselling and prenatal diagnosis (PND) were implemented over a 5 year period.

Results: Among a total of 108,264 screened individuals (54,132 couples), β-thalassaemia trait, δβ-thalassaemia trait, and sickle cell trait were diagnosed in 3.98%, 0.11% and 0.07%, respectively. Of 130 at risk couples (2.4/1000), 107 (82%) were available for follow up, with 105 couples (98.1%) proceeding with their marriage after counselling. In the 125 registered pregnancies in the latter couples, PND was performed in 85 (in 80 couples, uptake 76%). Selective termination was chosen in 10 of the 11 pregnancies with an affected fetus. Six affected babies were born among couples who declined PND. At the same time 30 already married couples with at least one thalassaemic child underwent PND, revealing three affected fetuses; all three pregnancies were terminated.

Conclusion: The programme revealed that most at risk couples diagnosed by premarital screening chose to proceed with their marriage, with 76% seeking PND followed by selective termination of an affected fetus. A 65% reduction in number of affected births was reported over the 5 year period. This regional programme could serve as a prototype for a national haemoglobinopathy prevention programme.

Consanguineous marriages, pearls and perils: Geneva International Consanguinity Workshop Report

Hanan Hamamy, MD[1], Stylianos E. Antonarakis, MD, DSc[1], Luigi Luca Cavalli-Sforza, MD[2], Samia Temtamy, MD, PhD[3], Giovanni Romeo, MD[4], Leo P. Ten Kate, MD, PhD[5], Robin L. Bennett, DSc[6], Alison Shaw, PhD[7], Andre Megarbane, MD, PhD[8], Cornelia van Duijn, PhD[9], Heli Bathija, MD[10], Siv Fokstuen, MD[1], Eric Engel, MD[1], Joel Zlotogora, MD, PhD[11], Emmanouil Dermitzakis, PhD[1], Armand Bottani, MD[1], Sophie Dahoun, MD[1], Michael A. Morris, DPhil[12], Steve Arsenault, BSc[13], Mona S. Aglan, MD[3], Mubasshir Ajaz, BSc, MSc[14], Ayad Alkalamchi, MD[15], Dhekra Alnaqeb, MD[16], Mohamed K. Alwasiyah, PhD[17], Nawfal Anwer, MD, FIBOG[18], Rawan Awwad, MS[19], Melissa Bonnefin, BA, MA[20], Peter Corry, MD, FRCPCH[21], Lorraine Gwanmesia, MD[1], Gulshan A. Karbani, BSc, MSc[22], Maryam Mostafavi, MD[1], Tommaso Pippucci, PhD[23], Emmanuelle Ranza-Boscardin, MD[1], Bruno Reversade, PhD[24], Saghira M. Sharif, BSc, PGCLTHE[25], Marieke E. Teeuw, MD[26], and Alan H. Bittles, PhD, ScD[27]

Abstract: Approximately 1.1 billion people currently live in countries where consanguineous marriages are customary, and among them one in every three marriages is between cousins. Opinions diverge between those warning of the possible health risks to offspring and others who highlight the social benefits of consanguineous marriages. A consanguinity study group of international experts and counselors met at the Geneva International Consanguinity Workshop from May 3 2010, to May 7, 2010, to discuss the known and presumptive risks and benefits of close kin marriages and to identify important future areas for research strong preferential culture of close kin marriages in many societies, and among migrant communities in Western countries, merits an equivalently detailed assessment of the social and genetic benefits of consanguinity in future studies. ***Genet Med* 2011:13(9):841–847.**

Key Words: *consanguinity, consanguineous marriage, inbreeding, endogamy, fertility, stillbirths, infant mortality, congenital disorders, genetic counseling*

What gets published?

Good papers:

- Relevant to the audience
- Important
- Original
- Methodologically sound
- Ethically sound

BUT .. The definition of 'good, important, sound' etc can be subjective

Step 1: Formulate a specific question

- Most important step!
- Must be CLEAR and CONCISE
- Must be stated clearly with a hypothesis

Formulate a research question related to community genetics

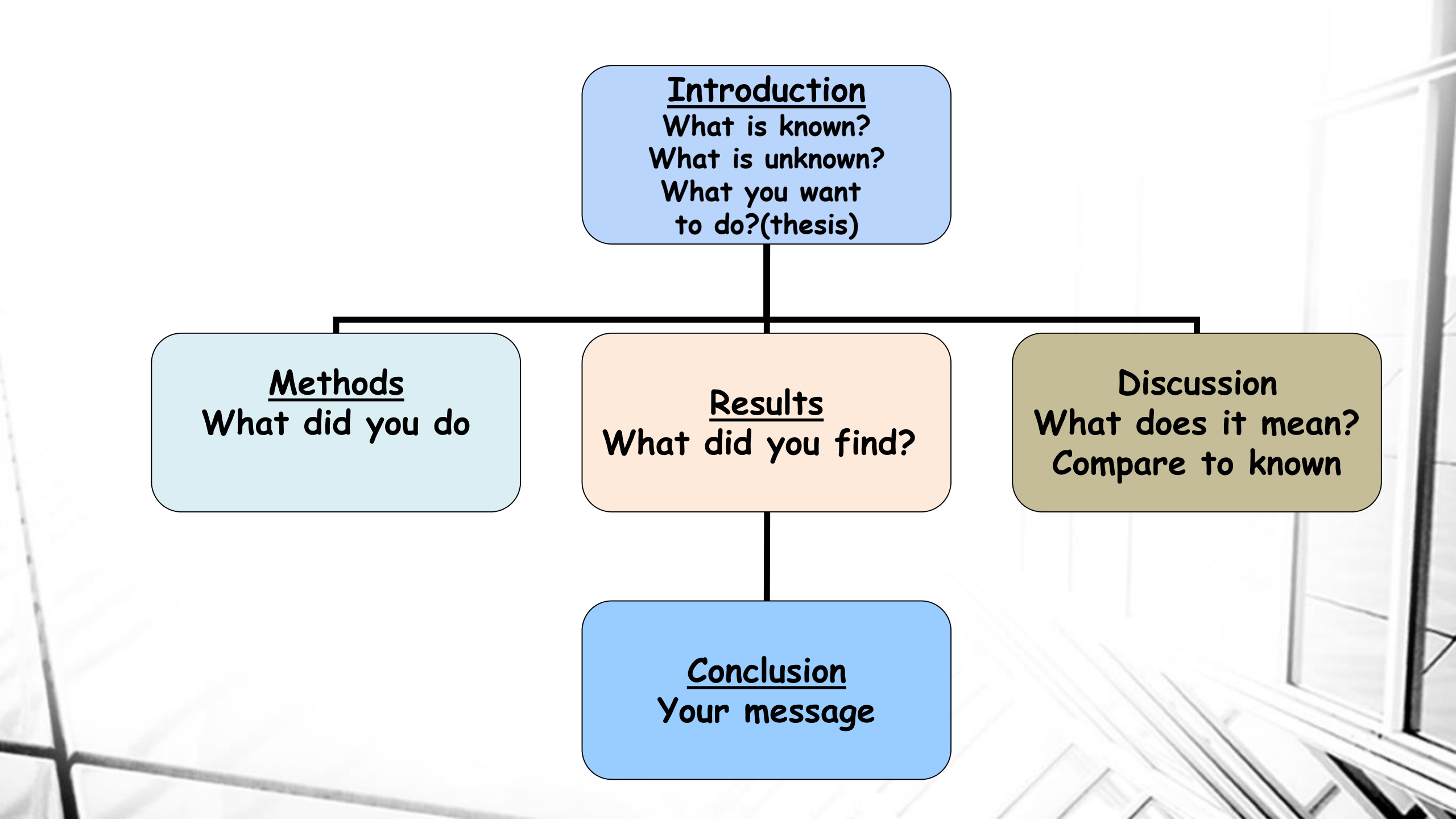
Prevalence of birth defects (affected and carriers)	• Specific defects • Infant and child causes of mortality • Consanguinity rates and effects
What genetic services are available	• Specialized genetic services • Preconception care • Lab facilities
Human resources	• Numbers of genetics specialists • Number of lab personnel for diagnosis of birth defects
Education and training	• College and university curricula in community genetics • Workshops and training courses for health care personnel • Media messages
Facilities for the care and rehabilitation of affected	• Special schools • Medical care

Format of manuscript

- Original research
- Review
- Case report
- Brief communication
- Letter to the editor

Sections of a scientific manuscript IMRaD Principle

- Title
- Abstract
- Keywords
- Introduction
- Material and methods
- Results
- Discussion
- Conclusion
- Acknowledgements
- Conflicts of interest
- References


Introduction
What is known?
What is unknown?
What you want
to do?(thesis)
Methods
What did you do
Results
What did you find?
Discussion
What does it mean?
Compare to known
Conclusion
Your message

Introduction

1. Statement of the issue
2. Why is your paper needed
3. Your purpose and hypothesis

Introduction

- First paragraph introduces the issue your paper will address . The first sentences should be attractive to catch the readers attention
- Second paragraph includes reasonable literature review and why your paper is needed
- Third paragraph includes the purpose of your study. The last and most important sentence in the introduction begins with : ***"The purpose of this study is….."***
- This sentence includes the hypothesis you are putting or your research question. The editor searches here for your input. It is called the ***thesis***

Methods

4-8 paragraphs (most important section)

- ✓ Subjects
- ✓ Procedures
- ✓ Terms and measures
- ✓ Data Collection
- ✓ statistics

Subjects

- Design: should be indicated in first sentence:
  - Case report
  - Case series
  - Case control study
  - Cohort
  - Clinical trial
- Collection of data: prospective or retrospective
- Criteria for inclusion and exclusion
- Demographics of studied population is in methods section for retrospective studies and in results section for prospective studies. They include
- Number, age, sex, symptoms, presentation, choice of controls..

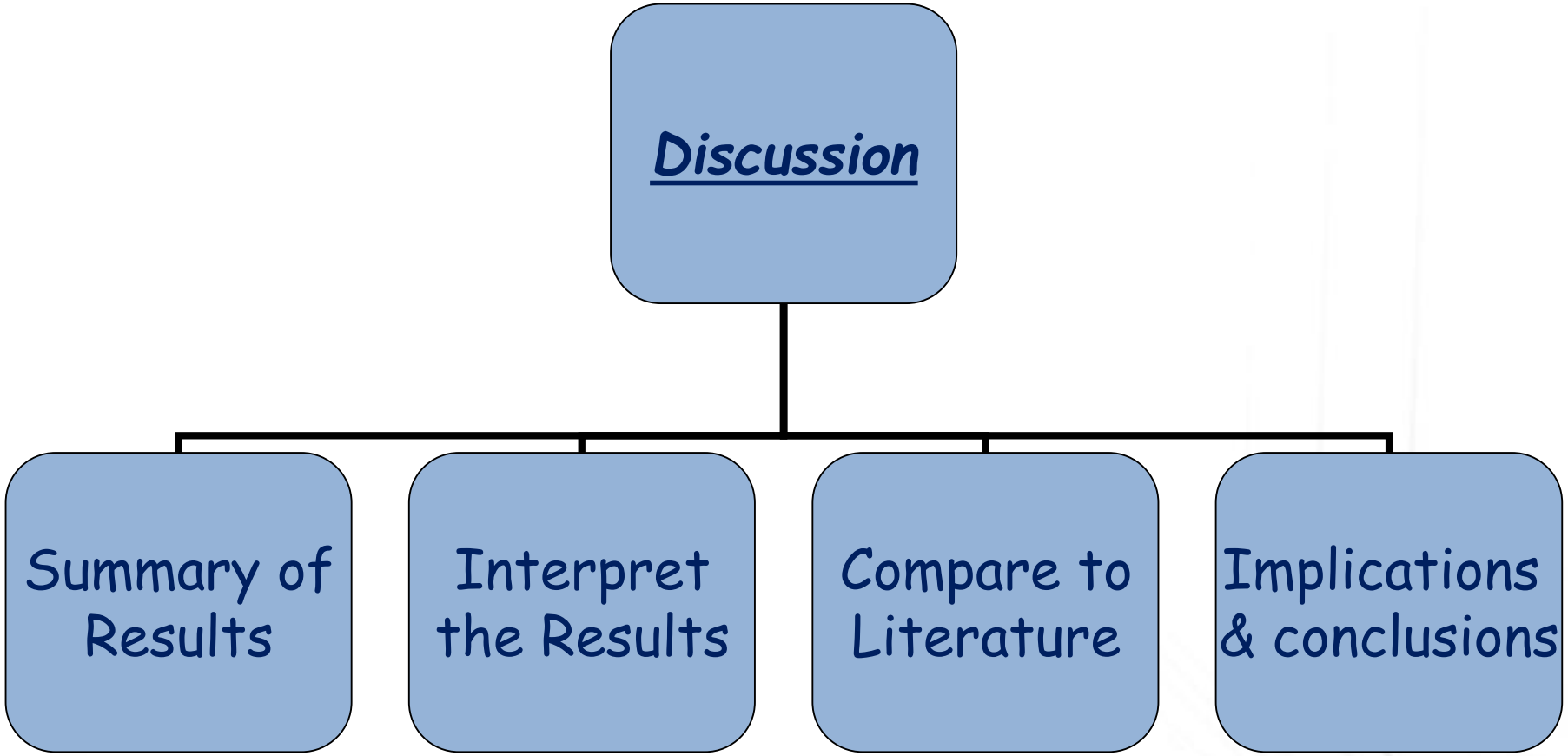
Procedures

Details of all procedures should be explained in the order they were applied

- clinical
- laboratory investigations
- radiology
- Others
- Provide definitions, measures and statistical proceduresclearly

Results

- Report all results as facts and numbers without interpretation
- Units should be accurate
- Make sure numbers add up correctly
- Data either put as text or tables and figures, do not repeat
- About 5-7 paragraphs


Discussion
Summary of Results
Interpret the Results
Compare to Literature
Implications & conclusions

Discussion

- First paragraph: summarize your principal and most important findings according to your thesis, do not repeat all data from result section, do not add new data, do not start with literature review
- Second paragraph: interpretation of your results
- Third paragraph: Compare and contrast your findings and interpretations with other studies from the literature and suggest reasons. Do not repeat the literature review from the introduction
- Fourth paragraph: indicate the limitations of your study without undermining its strength
- Fifth paragraph: Summarize your findings and conclusions. Clearly indicate what are the important implications for policy, practice and future research. The last sentence should describe what would be needed next

References

- Include mostly the most recent , the most important and those directly related to your study
- Try not to exceed 30 references for a researched study and less for case report
- Format exactly as instructed in authors guidelines

Abstract: usually written last

- **Background and purpose of study**
- **Sample and methods**
- **Results**
- **Conclusion on findings should reflect the words of the purpose.**

Clin Genet 2005: 67: 511–516
Printed in Singapore. All rights reserved

CLINICAL GENETICS
doi: 10.1111/j.1399-0004.2005.00426.x

Short Report

Consanguineous marriages in Jordan: why is the rate changing with time?

Hamamy H, Jamhawi L, Al-Darawsheh J, Ajlouni K. Consanguineous marriages in Jordan: why is the rate changing with time? Clin Genet 2005: 67: 511–516. © Blackwell Munksgaard, 2005

The objective of this study was to explore the secular trend in consanguinity in Jordan and the subtypes of consanguineous marriages that may be undergoing a change. A total of 1032 individuals attending a diabetic clinic in Amman were interviewed. The questionnaire provided information on consanguinity status and date of marriage among three generations: the persons interviewed, their parents, parents of their spouses and their offspring. Data on consanguinity status among 5401 marriages was obtained. Generations were named generation 1 for marriages contracted before 1950, generation 2 for marriages contracted between 1950 and 1979, and generation 3 for

H Hamamy, L Jamhawi, J Al-Darawsheh and K Ajlouni

National Center for Diabetes, Endocrinology and Genetics, Amman, Jordan

The objective of this study was to explore the secular trend in consanguinity in Jordan and the subtypes of consanguineous marriages that may be undergoing a change.
A total of 1032 individuals attending a diabetic clinic in Amman were interviewed.
The questionnaire provided information on consanguinity status and date of marriage among three generations: the persons interviewed, their parents, parents of their spouses and their offspring.
Data on consanguinity status among 5401 marriages was obtained.
Generations were named generation 1 for marriages contracted before 1950, generation 2 for marriages contracted between 1950 and 1979, and generation 3 for marriages contracted after 1980.
For generations 1, 2, and 3, first-cousin marriages constituted 20.2, 28.5 and 19.5% of all marriages, respectively.
In conclusion, first-cousin marriage rate among a representative population from Amman showed a significant decline among marriages contracted after 1980 compared to marriages contracted between 1950 and 1979, but not to marriages contracted before 1950.

Journals may ask for:

- Ethical approval
- Source of funding
- Participation of each co-author
- Conflicts of interest

Steps for scientific manuscript writing

Decide to write: first steps

- Decide on the topic. 

Must be interesting to you: timely and relevant and not outdated topic, presents a new idea

- Identify a target journal
- Put your own timetable 
- Begin with a thorough literature search and gather relevant articles

Key questions to answer to identify your idea and message

What prompted me to do the work and to write
What does previous research say about the issue
What is the work setting and nature of study
What are my methods
What are my major findings
What did I learn that I can tell others
What do I plan to do next
How are my findings of benefit to patients, doctors, public..

Second steps

- Identify 2,3 articles as the most relevant, read them very carefully and use them as your templates
- Put a preliminary title
- Sketch an outline , mainly with headings, not too detailed
- Write your first draft, don't think too much about grammar at this stage.

Further steps

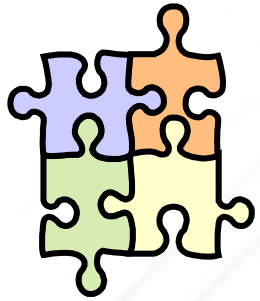
- Revise, revise, revise & give to co-authors or supervisor
  - Say what you mean, mean what you say
  - Words and sentences must be simple, clear and accurate
  - Avoid repetitions
  - Be careful not to commit any scientific fraud

Scientific fraud

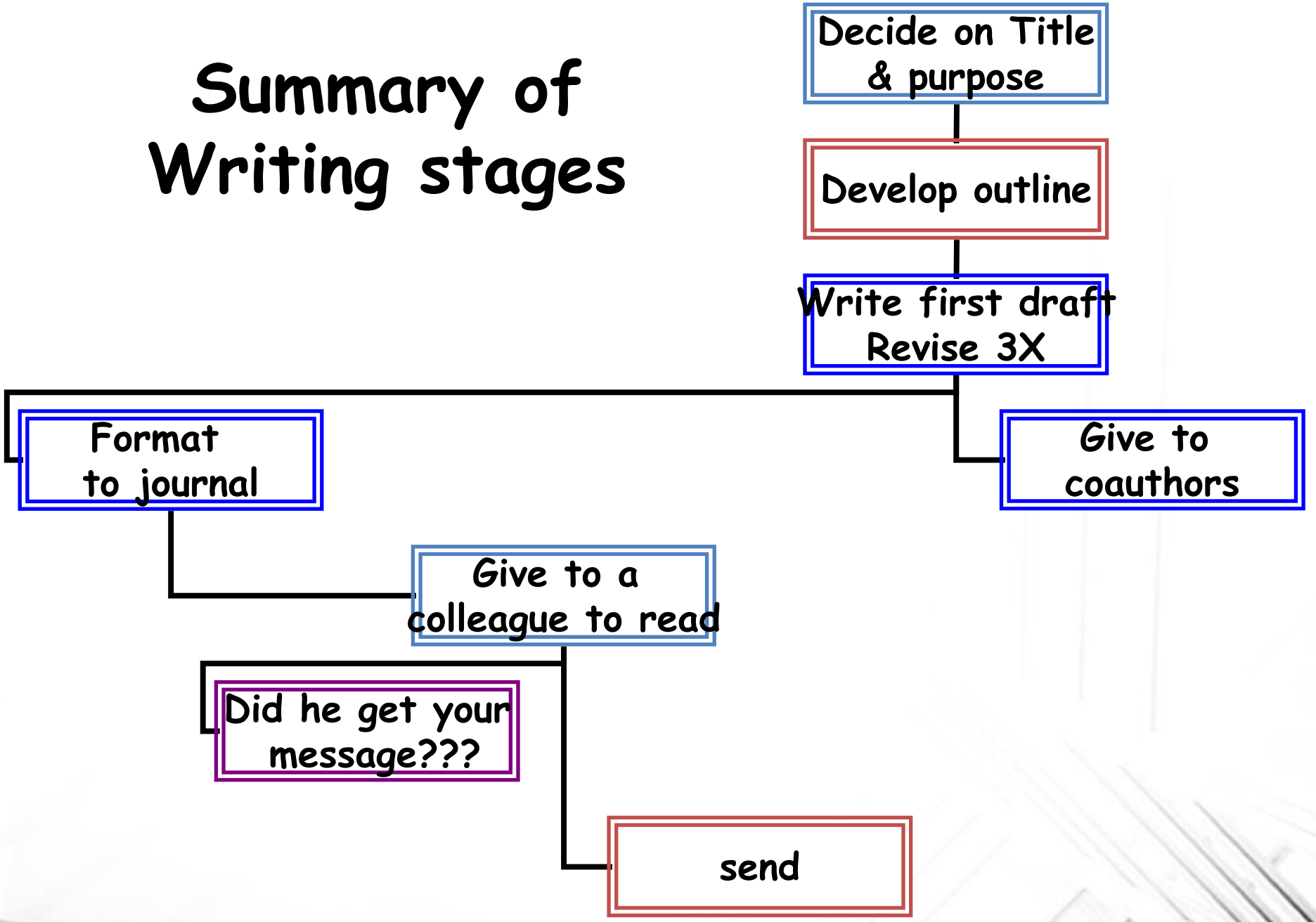
- falsification: altering truthful information
- fabrication: inventing information where none previously existed
- Plagiarism: To use someone else's words or ideas and put them as your own without proper reference

Further steps

- Proper spelling, grammar and punctuation is essential in last draft
- Must follow the journal's instructions to authors (you must read them thoroughly)
- Follow the journal's requirements regarding referencing system /number and style of tables & figures/ word count
- Don't forget: Conflict of interest /funding /acknowledgements /authors' contributions



Summary of Writing stages



Authorship

- As a rule the principal writer should be the first author
- The supervisor or head of research team should be last
- Other authors are listed after the first author in order of their level of contribution

Guidance on the authorship of scientific papers : The International Committee of Medical Journal Editors

Authorship credit should be based on

- substantial contributions to conception and study design, or acquisition of data, or analysis and interpretation of data
- drafting the article or revising it critically for important intellectual content
- final approval of the version to be published.

Authors should meet all three conditions

Letter to the editor

A must!

Must include the main message of the paper (if it is not the same as the title)

Your message should be clear and concise. Does the evidence in the paper support it? Is it interesting? And who would it interest?

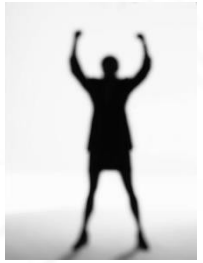
Tell the editor why your paper is worth publishing, how will it add to the evidence, and how it fits with what is usually or previously published in their journal

What is an impact factor

For a particular journal, the journal impact factor (JIF) is defined as the number of citations within a given year (e.g., 2005) cited to all papers published in that journal during the previous 2 years (i.e., 2004 and 2005), divided by the total number of papers published in that journal during those 2 years.

Last steps

- Submit and wait
- Expect:
  - Immediate rejection
  - Late rejection with or without reviewers comments
  - Provisional acceptance with reviewers comments
  - Acceptance without changes



Last steps

- If changes wanted, revise according to reviewers comments and resubmit with cover letter detailing all changes
- If accepted, proof will be sent to you, read thoroughly for any mistakes and send in 2 days
- If rejected, send to another journal after changing format accordingly

What is your reaction when you receive request for changes?

Sometimes the way in which an author responds to reviewers' comments reflects the author's responses to other stressors in life.

Some individuals are unable to accept criticism in a simple manner and essentially revolt at the idea that their work needs improvement

The characteristics that allow an author to successfully revise a manuscript:

- Acceptance of criticism
- Willingness to revise one's position
- Perseverance
- Organizational skills.

Revise according to reviewers comments and resubmit with cover letter detailing all changes

10 principles to assist in the revision process

Principle 1. Decide Whether to Resubmit the Manuscript to the Same Journal

Principle 2. Contact the Editor Regarding Unresolved Issues

Principle 3. Prioritize the Reviewers' Comments

Principle 4. Approach the Reviewer as a Consultant Rather than an Adversary

Principle 5. Deal With Reviewer Comments when one Does Not Agree

Principle 6. Disagree Without Being Disagreeable

Principle 7. Devise a Strategy for Responding to Divergent Comments

Principle 8. Put in the Work and Show All That You Have Done

Principle 9. If Requested, Shorten the Manuscript

Principle 10. Review the Medical Literature Before Resubmission

Provenzale JM. AJR Am J Roentgenol. 2010;195:W382-7

Thank the reviewers

Thank you for giving this manuscript a chance to be reviewed for possible acceptance in your respectful journal . We would like to thank the reviewers for their valuable comments which were all taken into consideration in revising the manuscript. The changes are highlighted in the text. The following is a point by point description of the response to the reviewers' comments:

Most frequent reasons for manuscript rejection

1. Inappropriate or incomplete statistics
2. Over-interpretation of results
3. Inappropriate methods
4. Sample too small or biased
5. Text difficult to follow
6. Problem not stated clearly
7. Inaccurate or insufficient data
8. Incomplete, inaccurate or outdated literature
9. Defective tables or figures

Tips to avoid rejection

- Start with the right study design as this cannot be corrected later
- Describe the methods and results optimally and accurately. Do not interpret in results section
- Discussion should be no longer than necessary, draw the right conclusions from the results
- Say what you mean, mean what you say and don't use big words
- Choice of journal and format

For Novice writers 

- Take a decision to write with persistence
- Get over your internal barriers such as your lack of confidence of being a writer
- Put your own timeline
- Assign a good half an hour each day for writing your paper
- There is no perfectionism , so do not stop in the middle, first drafts do not need good language
- With time you will learn to " *write with a gallop that leaves one's internal critics behind"* (Virginia Woolf)

Conclusions

- It is essential to disseminate information
- Writing scientific manuscripts is a learned process
- The more a person writes, the better writer he becomes
- Writing needs time , effort and accuracy
- Contribution makes you an expert in the field