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?
Mild micrognathia
Brachycephaly
Low posterior hair line
10 years old patient (full face and lateral view)
Hypertelorism
Pointed nasal tip, anteverted nostrils
Flat philtrum
Thin upper vermilion border
Novel syndrome: AR or XR?
Sparse lateral eyebrows
Protruding and malformed ears
Absence / dysfunction of lacrimal structures
Midface prominence
Hypodontia
High myopia
Mild hearing impairment
Mild micrognathia
pleasant, joyful
Learning difficulties
Low posterior hair line
10 years old patient (full face and lateral view)
New Syndrome

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

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We report on two brothers, born to double first cousin Jordanian Arab parents, with a syndrome comprising severe hypertelorism with upsailed 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
Mutations in IRX5 impair craniofacial development and germ cell migration via SDF1

Carine Bonnard1, Anna C Strobi2, Mohammad Shboul1, Hane Lee3, Barry Merriman3, Stanley F Nelson3, Osama H Ababneh4, Elif Uz5,6, Tülay Güran7, Hülya Kayserili8, Hanan Hamamy9,10 & Bruno Reversade1,11

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 punctae (b, inset), anteverted nostrils, pointed nasal tip, flat philtrum, thin upper vermilion 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 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
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
The first five years of a preventive programme for haemoglobinopathies in Northeastern Iraq

Nasir AS Al-Allawi¹, Sana D Jalal², Najmaddin H Ahmed³, Azad H Faraj³, Awaz Shalli² and Hanan Hamamy⁴

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% of 5000), 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, MD1, Stylianos E. Antinarakis, MD, DSc1, Luigi Luca Cavalli-Sforza, MD2, Samia Temtamy, MD, PhD3, Giovanni Romeo, MD4, Leo P. Ten Kate, MD, PhD5, Robin L. Bennett, DSc6, Alison Shaw, PhD7, Andre Megarbane, MD, PhD8, Cornelia van Duijn, PhD9, Heli Bathija, MD10, Siv Fokstuen, MD10, Eric Engel, MD11, Joel Zlotogora, MD, PhD11, Emmanouil Dermitzakis, PhD1, Armand Boitani, MD1, Sophie Dahoun, MD1, Michael A. Morris, DPhD12, Steve Arsenault, BSc13, Mona S. Aglan, MD13, Mubasshir Ajaz, BSc, MSc14, Ayad Alkalami, MD15, Dhehra Alnagib, MD16, Mohamed K. Alwastyah, PhD17, Nawful Anwer, MD, FIBOG18, Rawan Awwad, MS19, Melissa Bonnefin, BA, MA20, Peter Corry, MD, FRPCPCH21, Lorraine Gwanmesia, MD21, Gulshan A. Karbani, BSc, MSc22, Maryam Mostafavi, MD22, Tommaso Pippucci, PhD23, Emmanuelle Ranza-Boscardin, MD24, Bruno Reversade, PhD24, Saghiria M. Sharif, BSc, PGCLTIE25, Marieke E. Teetwil, MD26, and Alan H. Bittles, PhD, ScD27.

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 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.


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

Genetics IN Medicine • Volume 13, Number 9, September 2011
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

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

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

Procedures
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
Background and purpose of study

Sample and methods

Results

Conclusion on findings should reflect the words of the purpose.

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

<table>
<thead>
<tr>
<th>Question</th>
<th>Details</th>
</tr>
</thead>
<tbody>
<tr>
<td>What prompted me to do the work and to write</td>
<td></td>
</tr>
<tr>
<td>What does previous research say about the issue</td>
<td></td>
</tr>
<tr>
<td>What is the work setting and nature of study</td>
<td></td>
</tr>
<tr>
<td>What are my methods</td>
<td></td>
</tr>
<tr>
<td>What are my major findings</td>
<td></td>
</tr>
<tr>
<td>What did I learn that I can tell others</td>
<td></td>
</tr>
<tr>
<td>What do I plan to do next</td>
<td></td>
</tr>
<tr>
<td>How are my findings of benefit to patients, doctors, public</td>
<td></td>
</tr>
</tbody>
</table>
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

- Decide on Title & purpose
- Develop outline
- Write first draft
  - Revise 3X
- Format to journal
- Give to a colleague to read
  - Did he get your message???
  - send
- Give to coauthors
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

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