DNA Mixture Interpretation Principles: Observations from a NIST Scientific Foundation Review AAFS 2019 Workshop #10 (February 18, 2019; Baltimore, MD)

Things we know from Transfer Studies

Sheila Willis Guest Researcher NIST





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Disclaimer

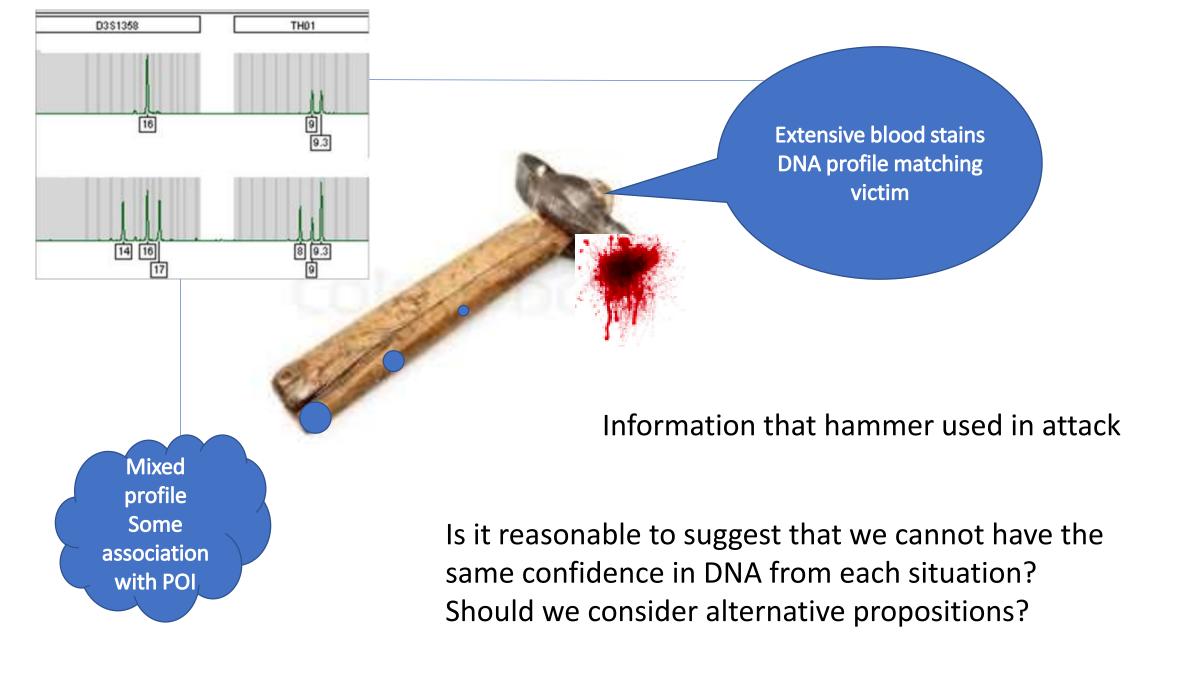
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Why worry about relevance in a mixture workshop?

- There is a growing interest in transfer and persistence studies in the literature in recent years since the ability to profile invisible stains
- It is easy to suggest that this has little to do with the complexity of mixture deconvolution but I suggest the opposite
- We know there are varying numbers of genotypes in a mixture
- Therefore it is arguable more important than ever to consider relevance
- DNA is , as we all know, is a wonderful material to discriminate one person from another
- It readily transfers and we have the tools to detect very small quantities of it
- This later property gives challenges as well as benefits





Relevance affected by when DNA is transferred to crime samples

Before the crime

Crime Event

Processing the samples



Transfers at this stage are the only relevant ones

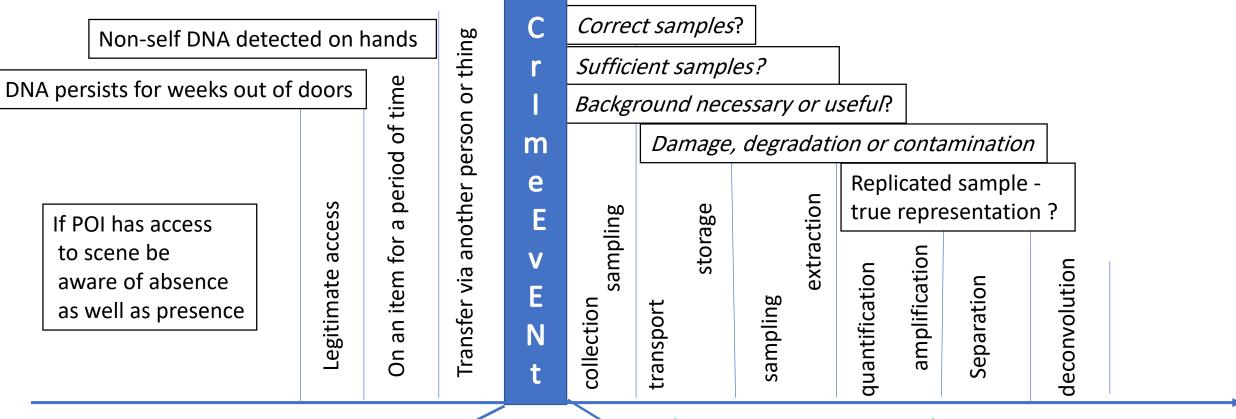


3 Reviews on this topic in the last twelve months
Taylor, D., Kokshoorn, B. and Biedermann, A. (2018) 'Evaluation of forensic genetics findings given activity level propositions: A review', *Forensic Sci Int Genet*, 36, pp. 34-49.

van Oorschot, R. A. H., Szkuta, B., Meakin, G. E., Kokshoorn, B. and Goray, M. (2019) 'DNA transfer in forensic science: A review', *Forensic Sci Int Genet*, 38, pp. 140-166.

Burrill, J., Daniel, B. and Frascione, N. (2018) 'A review of trace "Touch DNA" deposits: Variability factors and an exploration of cellular composition', *Forensic Sci Int Genet*, 39, pp. 8-18.

Relevance and issues that need to be considered



Possible transfer before the crime considered here

Relevant transfers

Studies re contamination avoidance

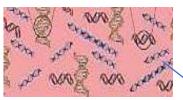
Intra and inter-laboratory validation studies

Context in which crime committed

Potential for errors to be considered here

Potential sources of DNA which could deposit from the hands

Cell free DNA



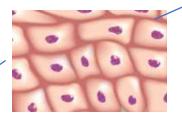
Fragmentassociated residual DNA



Transferred exogenous nucleated cells



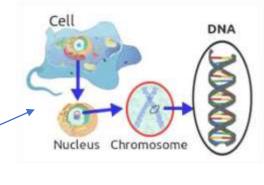
Endogenous nucleated cells



Anucleate corneocytes



Method of deposit not agreed Sweat Sebaceous fluid





Dynamic – transfers in either direction possible

Salivia, sneezing, dandruff

J. Burrill et al, Forensic Science International Genetics 39 (20019) 8-18

Transfer studies - Impact of individual donor



Shedder status – the amount of DNA detected from an individual Most of publications agree that some people

shed more readily than others
Likely to be a continuum rather than strict divide



Factors studied
Gender, Age, Sex
Time since hand washing
Activities, Part of hand

References for some of these studies on your slides

In spite of variation in the studies, support for the view that some individuals shed more than others

Status

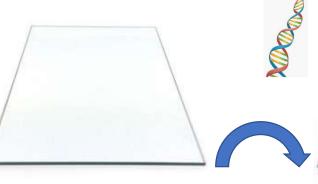
- Affects direct transfer
- Affects likelihood of detection of major profile from last handler
- Affects persistence
- Affects detection of non-self on hands

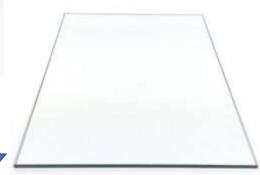


Transfer studies – effect of substrate











Less transferred to hard non-porous surface but lost more quickly from Higher amount transferred to soft porous surface but less transferred

Passive, pressure and friction

Increasing amount of transfer

Moisture

Friction to transfer DNA from non-porous to porous the most efficient chain

DNA transfers affected by

- 1. type of substrate
- 2. Moisture
- 3. pressure

Goray, M., Eken, E., Mitchell, R. J. and van Oorschot, R. A. (2010) 'Secondary DNA transfer of biological substances under varying test conditions', *Forensic Sci Int Genet*, 4(2), pp. 62-7.

Verdon, T. J., Mitchell, R. J. and van Oorschot, R. A. (2013) 'The influence of substrate on DNA transfer and extraction efficiency', *Forensic Sci Int Genet*, 7(1), pp. 167-75.

One Part of a study of Secondary Transfer to Wood Glass and Metal



10 transfer chains for door handle; average of 55ng deposited on handle; 64% transferred to gloves; 32% transferred to cloth.

Fonnelop, A. E., Egeland, T. and Gill, P. (2015) 'Secondary and subsequent DNA transfer during criminal investigation', *Forensic Sci Int Genet*, 17, pp. 155-162.

Research likely influenced by observations on investigation of Merdith Kercher murder resulting in miscarriage of Justice

Gill, P. (2016) 'Analysis and implications of the miscarriages of justice of Amanda Knox and Raffaele Sollecito', *Forensic Sci Int Genet*, 23, pp. 9-18.

Shedder status and Investigation of Self and Non-Self







Last person to handle

- 240 handprints deposited by 10 individuals;
- Analyzed for differences in DNA quantity and type of profile at different times on different days;
- Inter-personal variation higher than intra-personal. 0.05- 5 ng per deposit;
- Mainly 2 person mixtures or non interpretable; non- self in 79%
 Non-self usually the minor component;
- On rare occasion when non-self was major, they were associated with poor depositor/shedder; 7/240 self excluded

Goray, M., Fowler, S., Szkuta, B. and van Oorschot, R. A. H. (2016) 'Shedder status-An analysis of self and non-self DNA in multiple handprints deposited by the same individuals over time', *Forensic Sci Int Genet*, 23, pp. 190-196.

Time since deposit

Raymond, J. J., van Oorschot, R. A. H., Walsh, S. J., Roux, C. and Gunn, P. R. (2009) 'Trace DNA and street robbery: A criminalistic approach to DNA evidence', *Forensic Science International: Genetics Supplement Series*, 2(1), pp. 544-546



Profiles obtained out of doors up to two weeks with technology available in 2009



Taylor, D., Abarno, D., Rowe, E. and Rask-Nielsen, L. (2016) 'Observations of DNA transfer within an operational Forensic Biology Laboratory', *Forensic Sci Int Genet*, 23, pp. 33-49.

Profiles built up over a period of time in laboratory setting again highlighting need for elimination databases

Fewer systematic persistence studies than transfer studies

Persistence of one user following another

- Original user detected vast majority of experiments
- Varied depending on
 - Duration of use by second person
 - Substrate
 - Original handler shedder status
 - Activities /action
 - Duration of use

Study with computer and mouse

Original user detected and transferred to second user up to 8days

Differences in ability to detect initial user reasoned to be due to shedder status

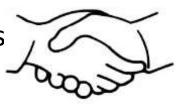




Trend in studies to mimic casework in a broad manner

Experiments with knives

Experiments with knives to check if indirect trans from person shaking hands with handler is detectable – yes handler main profile; 13 /20 5/20 secondary transfer main profile.



Cale, C. M., Earll, M. E., Latham, K. E. and Bush, G. L. (2016) 'Could Secondary DNA Transfer Falsely Place Someone at the Scene of a Crime





Is DNA of nearby person detected in stabbing cases and how much is transferred;

DNA of person handling knife major or single profile 83%; Profiles too complex 5%; Observers profiles not detected.

Samie, L., Hicks, T., Castella, V. and Taroni, F. (2016)

Is profile of regular user detected as well as stabber and hand shaker

Regular user persisted for at least a week; Non-donor DNA co-deposited 5% -declined with time.







Meakin, G. E., Butcher, E. V., van Oorschot, R. A. H. and Morgan, R. M. (2015)

Washing machine experiments



165-171.

DNA profiles from laundered semen stains recovered at least 8 months after deposition.

- •Micrograms of DNA and full DNA profiles recovered, irrespective of wash conditions.
- •No significant decline in DNA quantity and profile quality after multiple washes.
- •Both DNA sources detected on laundered T-shirt with semen stains from two donors.
- •Laundered semen-stained clothing should be examined in sexual assault cases.

Brayley-Morris, H., Sorrell, A., Revoir, A. P., Meakin, G. E., Court, D. S. and Morgan, R. M. (2015) 'Persistence of DNA from laundered semen stains: Implications for child sex trafficking cases', *Forensic Sci Int Genet,* 19, pp.

blo usa epi

Secondary transfer of DNA from blood following washing but no usable profiles from saliva or epithelial abrasions

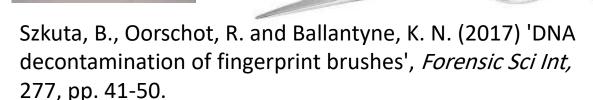
Kamphausen, T., Fandel, S. B., Gutmann, J. S., Bajanowski, T. and Poetsch, M. (2015) 'Everything clean? Transfer of DNA traces between textiles in the washtub', *Int J Legal Med*, 129(4), pp. 709-14.

Contamination studies









Szkuta, B., Harvey, M. L., Ballantyne, K. N. and van Oorschot, R. A. H. (2015) 'Residual DNA on examination tools following use', *Forensic Science International: Genetics Supplement Series,* 5(Supplement C), pp. e495-e497.





Fonnelop, A. E., Johannessen, H., Egeland, T. and Gill, P. (2016) 'Contamination during criminal investigation: Detecting police contamination and secondary DNA transfer from evidence bags', *Forensic Sci Int Genet*, 23, pp. 121-129.

At a minimum need elimination samples needed

Care needed to ensure DNA on window frame, the sample profiled in the lab

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Recap - we have seen

- Variation in shedder status
- Impact of substrate
- Persistence affected by background DNA as well as environment
- Contamination facilitated by many commonly used vectors



In addition we have major difficulties re standardization which hinders use Studies not comparable because of

- Changes in technology
- Impact of different extraction and analytical parameters
- Different methods of recording associations



Activity propositions are regularly suggested as way forward

Not the subject of this presentation but ideal framework for raising the correct questions

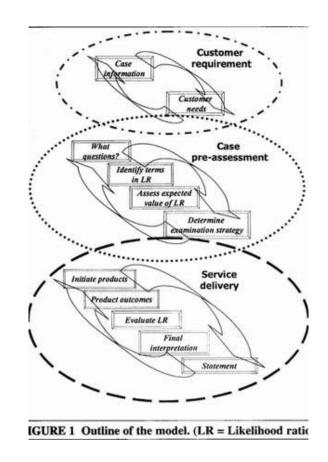
Tools to address these issues

Case Assessment and Interpretation

Model formulated to deal with trace evidence

- "A model for case assessment and interpretation" R COOK,IW EVETT*, G JACKSON,PJ JONES, JA LAMBERT; Science & Justice 1998: 38: 151-156
- Questions to be addressed arise from investigation and are the drivers behind the formulation of the propositions for the LR
- Later paper describes the effect of different levels of the hierarchy
- "A hierarchy of Propositions: deciding which level to address in casework" R COOK, IW EVETT*, G JACKSON, PJ JONES and JA LAMBERT; Science & Justice 1998; 38: 231-239

Illustration of the CAI model from original paper



Iterative nature is a precursor of the need to reassess WOE of association in light of case circumstances

Hierarchy of Propositions – questions being addressed

Adding value **Activity** propositions particularly relevant when transfer and persistence are an issue as in trace/touch

Offense

Court issues guilt/innocence

Generally not the concern of the forensic scientist Who, Why, What, How, When, Why, Intent ---

Activity

Forensic scientist needs to consider what's expected in light of different actions; answered such as when or how in addition to who

Source

Whether or not two materials share the same source – who Matching DNA when the cell type is known

Sub-Source

DNA when the cell type is not known – generally the case with mixtures and touch

Sub-sub-Source

Consider one genotype in a mixture

Some negative responses in the Court in Australia to DNA evidence



2007: The Queen v Hillier [2007] HCA 13

The conviction was quashed on the basis that the DNA evidence was considered in isolation from the other evidence. The weight accorded to DNA evidence must be considered in the context of all of the evidence presented by the prosecution



2007: Murdoch v The Queen [2007] NTCCA 1

The second issue was contamination. It was decided that the possibility of contamination could be excluded beyond reasonable doubt.

2014: Fitzgerald v The Queen [2014] HCA 28

During a burglary, one victim was murdered and serious brain injuries were inflicted on another. Fitzgerald was convicted of murder after DNA evidence linked him to a didgeridoo at the crime scene. There was no other evidence linking him to the crime. The conviction was appealed on the basis that the verdict was unreasonable as there were other possible ways the DNA could have been transferred. The DNA evidence was not sufficient to prove guilt beyond reasonable doubt, because there was no information about the circumstances in which the DNA was transferred. The DNA could have been deposited as a result of secondary transfer such as shaking hands with someone who did participate in the burglary.

Recent developments in DNA evidence Marcus Smith and Monique Mann ISSN 1836-2206 © Australian Institute of Criminology 2015 2009: R v Jama (Unreported, Supreme Court of Victoria, Court of Appeal, 2009)

A woman was found unconscious in a toilet cubicle. While she did not have any

The case highlights the potential for miscarriages of justice to occur when too much reliance is placed on DNA evidence, or it is the sole evidence in a case.

2010: Forbes v The Queen [2010] HCA Trans 120

Forbes was convicted of sexual assault. DNA found on the victim's trousers and bra was the sole evidence linking him to the crime. Two expert witnesses testified that there was 'extremely strong' and 'strong' evidence that the DNA profile on the victim's clothing belonged to Forbes. This evidence was challenged on appeal. It was not possible to prove the guilt of Forbes beyond reasonable doubt. Statistical estimates of the frequency that Forbes' DNA profile occurs in the population were calculated on the basis of a sample of 620 people (Gans 2011).





Possible to take the view that continue to report sub-source and leave someone else to figure it out because anything else is too dangerous

Can not separate knowledge of transfer, contamination risks and error rate when considering the need for robust appropriate findings

Context correct questions being asked Robust Scientific **Findings** Valid – Relevance applied transfer validity – and transfer contaminat history ion

Previous slide is not to suggest court determines scientific validity But if results are useful they must be fit for purpose. Another way of considering is that they need to answer the appropriate questions and if community doesn't take action risk undermining results anyway







FORENSIC GENETICS

What can DNA tell you about a crime?

Published in 2017

DNA IS EVERYWHERE

DNA is transferred by body fluids, shedding skin cells, talking, coughing and sneezing.

There is even DNA present in house dust:

So DNA from individuals who have nothing to do with a crime might be present at a crime scene. DNA will last almost indefinitely untouched in a dry, cool environment, although activities such as cleaning degrade it. DNA deposited before the crime so unrelated to it is called background DNA.





The DNA that is present in a room changes rapidly as people and objects interact within it, which is why it's so important for forensic experts to investigate a crime scene as soon as possible so that important DNA does not get moved and new DNA is not introduced.



17. WHERE CAN WE DETECT DNA?

- 1. DNA should not be used as the sole evidence in a criminal case_{3,4}
- 2. There is a considerable danger if the importance of the DNA evidence is inappropriately afforded greater weight than other evidence.

More recent studies suggest that background needs to be considered more

when evaluating DNA findings

Awareness of absence in instances when transfer expected Consider wider sampling of scenes

van den Berge, M., van de Merwe, L. and Sijen, T. (2017) 'DNA transfer and cell type inference to assist activity level reporting: Post-activity background samples as a control in dragging scenario', *Forensic Science International: Genetics Supplement Series*, 6(Supplement C), pp. e591-e592.

Jackson, G. (2013) 'The impact of commercialization on the evaluation of DNA evidence', *Front Genet*, 4.

Lehmann, V. J., Mitchell, R. J., Ballantyne, K. N. and Oorschot, R. V. (2015) 'Following the transfer of DNA: How does the presence of background DNA affect the transfer and detection of a target source of DNA?', *Forensic Sci Int Genet*, 19, pp. 68-75.



Increasing acceptance that approach to DNA needs to be driven by questions raised by the case rather than focus on single result

Impact of information on transfer and persistence

Holistic value from transfer studies information even if incomplete

- Transfer studies can be used to frame the samples to be taken at a scene
- Well known that some areas will be less useful than others
- Efforts to reduce mixtures almost instinctive avoid door handles and aim for areas connected with crime but not regularly handled
- Information in the studies highlight that we need even more caution

Therefore need mechanisms to impart the information to scene examiners

In laboratory and at scene an awareness of risk of contamination

When reporting or receiving reports

Red flag in situations where only one genotype is recovered from a scene where multiples might be expected

Worth considering areas likely to give rise to culprit profile Areas grabbed during a crime – work on ankles and armpits Work of colleagues in NFI – crowd science

How Science Should

Speak to Court –
recommended free
course from Coursera

Some issues being considered

- That the CAI model with Hierarchy of Propositions be used to evaluate DNA except in situations where there is no uncertainty re relevance of source of sample.
- If not possible the report states clearly that the rarity of the profile is not linked to its relevance in the case
- Consideration be given to obtaining background samples ie additional samples from items, in instances where contamination is a possibility ie all sub source sampling.
- In any situation other than stranger rape, DNA findings are evaluated at activity level and possibilities of sperm transferring unto clothing shared in households be considered.
- Anti-contamination measures be put in place in police facilities as well as laboratories
- Elimination databases for police and laboratory personnel are used as well as other relevant people in fixed situations other householders for example.

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Take-away messages

- Relevance of sub-source level associations can never be taken for granted in light of information about transfer and contamination
- Therefore need to be aware of what affects transfer
- Information about transfer affects scene, laboratory and reporting/court
- Think of alternatives when examining the findings
- Consider the implication of absence when unexpected
- Be particularly careful when dealing with a single sample in a case
- The findings in a case should form a coherent narrative
- In critical situations ad-hoc tests to replicate proposed scenario is worth considering
- In present state of knowledge it is difficult to extrapolate literature studies to a specific laboratory











Additional references for shedder status and knife experiments

- Manoli, P., Antoniou, A., Bashiardes, E., Xenophontos, S., Photiades, M., Stribley, V., Mylona, M., Demetriou, C. and Cariolou, M. A. (2016) 'Sex-specific age association with primary DNA transfer', *Int J Legal Med*, 130(1), pp. 103-12.
- Poetsch, M., Bajanowski, T. and Kamphausen, T. (2013) 'Influence of an individual's age on the amount and interpretability of DNA left on touched items', *Int J Legal Med*, 127(6), pp. 1093-6.
- Szkuta, B., Ballantyne, K. N. and van Oorschot, R. A. (2017) 'Transfer and persistence of DNA on the hands and the influence of activities performed',
 Forensic Sci Int Genet, 28, pp. 10-20.
- Oldoni, F., Castella, V. and Hall, D. (2016) 'Shedding light on the relative DNA contribution of two persons handling the same object', *Forensic Sci Int Genet*, 24, pp. 148-157.
- Oleiwi, A. A., Morris, M. R., Schmerer, W. M. and Sutton, R. (2015) 'The relative DNA-shedding propensity of the palm and finger surfaces', Sci Justice, 55(5), pp. 329-34.
- Lowe, A., Murray, C., Whitaker, J., Tully, G. and Gill, P. (2002) 'The propensity of individuals to deposit DNA and secondary transfer of low level DNA from individuals to inert surfaces', *Forensic Sci Int*, 129(1), pp. 25-34.
- Farmen, R. K., Jaghø, R., Cortez, P. and Frøyland, E. S. (2008) 'Assessment of individual shedder status and implication for secondary DNA transfer', Forensic Science International: Genetics Supplement Series, 1(1), pp. 415-417.
- Cale, C. M., Earll, M. E., Latham, K. E. and Bush, G. L. (2016) 'Could Secondary DNA Transfer Falsely Place Someone at the Scene of a Crime?', *J Forensic Sci*, 61(1), pp. 196-203.
- Samie, L., Hicks, T., Castella, V. and Taroni, F. (2016) 'Stabbing simulations and DNA transfer', Forensic Sci Int Genet, 22, pp. 73-80
- Meakin, G. E., Butcher, E. V., van Oorschot, R. A. H. and Morgan, R. M. (2015) 'The deposition and persistence of indirectly-transferred DNA on regularly-used knives', *Forensic Science International: Genetics Supplement Series*, 5(Supplement C), pp. e498-e500.
- In 2001, Peter Gill published a paper on the topic introducing the idea of sub-source Croatian Medical Journal42(3):229-232,2001

Thank you very much for your attention



