

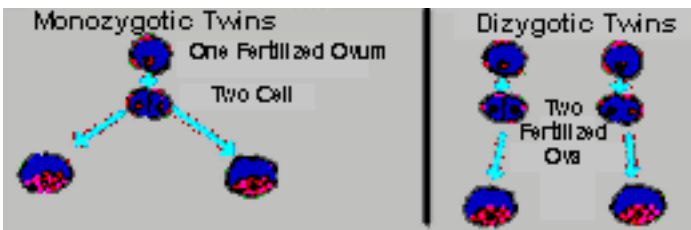


TWINS EYE STUDY

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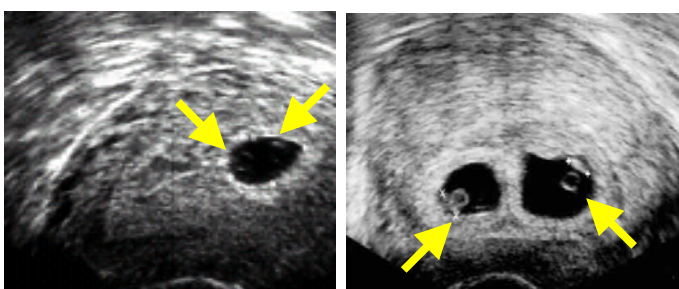
Identical or non-identical twins?

Determining whether twins are identical (monozygotic; MZ) or non-identical (dizygotic; DZ) is the first and most fundamental step in twin research. MZ means that the twins came from a single egg fertilised by a single sperm that subsequently split in half. DZ means that the twins came from two eggs fertilized by two sperms and thus have different genes.



Can we tell DZ from MZ before birth?

Often, but not always, MZ twins share one sack and one placenta and this may be noted on ultrasound. In contrast DZ twins usually but not always have separate sacs and placentas, which may be evident on ultrasound. The same may be noted at birth. Sometimes parents have been told that the twins are MZ or DZ when they were born but later it becomes evident that they are the other.



Ultrasound image on left shows MZ twins with one gestational sac with 2 yolk sacs visible inside it. The image on right resulted from implantation of 2 genetically different embryos (DZ twins) Yolk sacs and the early foetus are completely separate.

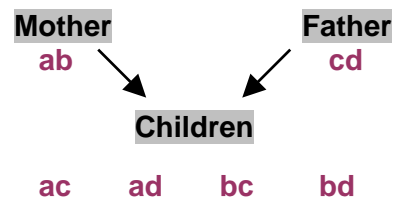
Usually it is fairly obvious when twins are identical as they are said to be “like 2 peas in a pod” or are often confused by people who know them well. Non-identical twins may be obviously different, particularly if one is male and the other female. (We have heard some great answers when mothers are asked if their twin boy and girl are identical!)

How accurate is zygosity testing?

Questionnaires to determine whether twins are DZ or MZ are very, but not 100%, accurate. If in doubt a DNA test can be performed. Some twins who do not look alike turn out to be identical. They may be different in size, often since birth with one twin not catching up. This shows us that the environment does have a role in some measurements.

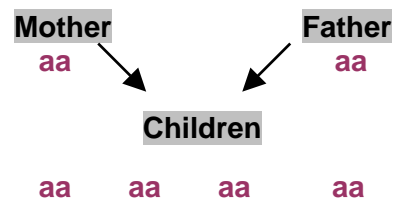
DNA testing is something that many people are familiar with due to TV shows such as CSI. MZ twins will have all the same genes, while DZ twins will share 50% of their genes (just the same as any two siblings).

We each have two copies of almost every gene, one from each parent. One copy of each gene is provided by each parent, thus in DZ twins or any other siblings the possibilities include:



Children can inherit one of four combinations. Thus for non-identical twins there is a 1/4 chance of them being exactly the same for any one gene marker.

Sometimes parents have exactly the same gene markers:



Thus all children will have the same markers and therefore this is not an informative marker.

If twins have a different marker they are clearly DZ. However, it is possible for DZ twins to have the same two genes in a particular location on their DNA, and therefore the same markers. There would be a 1/4 chance of this happening for each marker we use. If we test two markers it will be a 1/4 x 1/4 or 1/16 chance that we could mistake DZ twins for MZ.

Early DNA markers were blood groups. Some twins have had this done but as only a few markers were used we could not be as certain of the results. Many twins were told that there was 97% certainty that they were 100% identical (not 97% identical, which is sometimes misquoted).

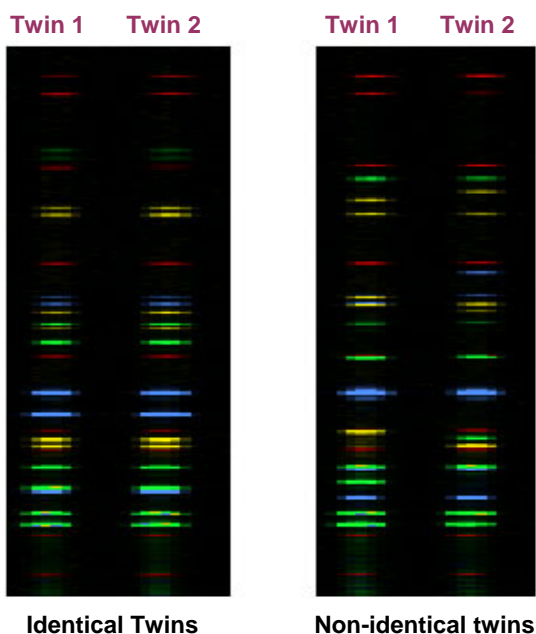
Today we normally test 10 DNA markers giving:
 $\frac{1}{4} \times \frac{1}{4} \times \frac{1}{4} \times \frac{1}{4} \times \frac{1}{4} \times \frac{1}{4} \times \frac{1}{4} \times \frac{1}{4} \times \frac{1}{4} \times \frac{1}{4}$
 =1/1,048,576 chance we could miscall DZ twins MZ. This one in a million chance of a mistake is very unlikely. Nonetheless, we could make a mistake, for example, mixing up the blood samples. Sometimes the marker sizes are not measured correctly and MZ twins may be mislabelled as DZ. So even if the DNA testing says MZ or DZ, but it doesn't seem right, we recheck.



Figure showing fingerprints from identical twins, although very similar, they are not identical.



Identical twins have very similar iris colour and detail. Right and left eyes of twin 1 are above, right and left eye of twin 2 are shown below.



Images showing DNA markers for two twin pairs, the two twins on the left have gene markers at identical locations to each other. Although similar, the markers for the two twins shown on the right do not line up completely. Therefore these are non-identical twins.

We have been testing at least 10 markers on all twins (except the male/female sets) to check how accurate our interpretation is. This has been the reason for the delay in getting results back. In addition sometimes the DNA sample just doesn't work and we need to recollect it.

Although the DNA for MZ twins is 100% the same, the twins are not 100% identical. Most features will be very similar, but slightly different. For example, the twins will have similar but different fingerprints and iris patterns. This is one of the reasons that these features are being introduced, by the US immigration authorities, for identification purposes.

We have attached a copy of your DNA testing data. If you think we are wrong please get in touch by phone, fax, email or in person and we will recheck. As a general rule, twins who think they are identical will be correct and twins who think they are not identical will be correct. Those who are not sure most often turn out to be identical but may look different (again showing that the environment does have some influence).

Comparing 10 markers allows us to tell MZ from DZ. Comparing 1000s of markers in the DZ twins is the next step in identifying the genes that are responsible for the variations in all the measurements we have taken. We will keep you informed of these results with our newsletters, but feel free to contact us at any time.

The twins who have participated in this research have made an enormous contribution to helping prevent blindness. Thank you. Please email (D.Mackey@utas.edu.au), phone (03 99298713 / 03 62228553), fax (03 99298711) or visit us if you have any questions about any of the DNA testing or the research.