Parry-Romberg Syndrome

A global survey of 205 people using the Internet

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Parry-Romberg Syndrome: a global survey of 205 people using the Internet.

Thank you to everyone who participated!

Thank you to everyone who has taken part in the Parry-Romberg Survey over the last 12 months. This is an updated version of the report that some of you will have received about 6 months ago. I am pleased to report that I had 214 responses to the web survey from all over the world. This is many times the number of responses I was hoping for. I am particularly grateful to Theresa Hildebrand and Marilyn Neal at the Romberg Connection, who helped promote this study through their website and personal support. I am also indebted to Adele Liu who had carried out an earlier survey and gave me a number of useful ideas as well as personal support. Thank you as well to Ann Houliston in Edinburgh who helped me a lot with some aspects of the study.

I hope the information from the survey is of some interest to everyone. As you'll know this is a condition which we know very little about. This the first time that a large number of patients have taken part in a research study to get an idea how common the various symptoms are in Parry-Romberg, who it affects and how useful people find the various treatments that they have had.

Introduction

As you all probably know Parry-Romberg syndrome is a condition in which there is shrinkage of the tissues and sometimes bone of one half of the face. Occasionally it can affect both sides. It overlaps with another condition, scleroderma 'en coup de sabre', to such a great extent that some people have wondered whether they are more or less the same disorder.

Various problems have been reported as being associated with Parry-Romberg syndrome including eye problems, migraine, epilepsy and problems with the arms and legs but the condition is so rare that it has been very hard to get an idea how common these things are and what the 'average person' with this condition can expect.

There are similar uncertainties about treatments. It is difficult for surgeons to build up experience with lots of patients with this condition so the survey attempted to get some idea of how successful surgery has been for people.

Finally, I hoped that the survey, by bringing together a large number of people with the condition might bring out some surprising aspect of the condition that hadn't come to light before which might advance our knowledge about it a little bit.

Carrying out the survey has certainly been a pleasure and an educational experience for me. I can't really do justice to the amount of detailed information many of you sent me - but I will do my best.
Who, where and how common?

137 people responded to the initial survey (118 female and 19 male). 129 people replied to the second half of the survey.

The average age of the people who replied to the survey was 33 years (youngest 4, oldest 64). 92% of you are right handed.

Below you can see a map of where people who replied to the survey live. Each dot on the map represents one person. As you can see most people came from the United States and other English speaking countries, but I did have replies from Argentina, India, Iran, Japan, Korea, Singapore, Tunisia and Venezuela. I had replies from people of all ethnic origins (although most were white).

Assuming that Parry-Romberg is not becoming rarer or more common and using the data from the commonest age band (31-40) - Parry-Romberg affects at least 1 in 700,000 people.
This must mean it is a lot commoner than that because even in the 31-40 band there will be people who haven't looked for the survey, did not want to fill it in or do not have access to the internet. I would guess its something like 1 in 250,000.

**Diagnosis**

The average age of which Parry-Romberg started was 10 years old although this included a range of ages of onset from birth to 50.

![Parry Romberg - Age of Onset](image)

On average there was a delay of 4 years between symptoms beginning and diagnosis, but again some people were diagnosed almost immediately and some people had to wait an awful lot longer.

![Parry Romberg - Delay to Diagnosis](image)

The table below shows the different labels that have been used by your doctors to describe this condition. The numbers add up to over 100% since some of you have more than one label.

**Diagnostic labels**

- 45% Parry-Romberg disease/syndrome
- 40% Romberg syndrome
- 16% (Progressive) Facial Hemiatrophy
- 16% Scleroderma 'en coup de sabre' (5% isolated)
- 6% Linear Scleroderma (1% isolated)
Location and Severity of Atrophy (or coup de sabre)

I asked you to rate how severe you thought your disease was and these were the results?

All of you had some form of atrophy affecting the face.
In 50% of people it affected the right side of the face and 48% of people the left.
2% of you have Romberg's affecting both sides of your face.

I asked you whether you:

1. Whether you had a line going down your forehead like Picture 1 to the corner of your eye? *

2. Whether it affected your cheek?

3. Whether you had a line or indentation on your chin?

See the next page for the results...
Here are the results from that bit!

<table>
<thead>
<tr>
<th>Location Affected</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Face</strong></td>
<td>100% (48% left, 50% right, 2% both sides)</td>
</tr>
<tr>
<td><strong>Forehead - as a line</strong></td>
<td>51% (10% in the corner of the eye, 20% in the middle of the eyebrow, 19% within one centimetre of the middle)</td>
</tr>
<tr>
<td><strong>Eye / Vision problems</strong></td>
<td>49%</td>
</tr>
<tr>
<td><strong>Cheek</strong></td>
<td>75%</td>
</tr>
<tr>
<td>- lips</td>
<td>55%</td>
</tr>
<tr>
<td>- teeth / gums</td>
<td>50%</td>
</tr>
<tr>
<td>- tongue</td>
<td>25% all on the same side</td>
</tr>
<tr>
<td><strong>Chin</strong></td>
<td>43%</td>
</tr>
<tr>
<td><strong>Overlap of ‘coup de sabre’ with lower facial hemiatrophy</strong></td>
<td>61% of patients with coup de sabre have lower facial involvement</td>
</tr>
<tr>
<td><strong>Arm</strong></td>
<td>9% (74% same side as face, 26% opposite side)</td>
</tr>
<tr>
<td><strong>Trunk</strong></td>
<td>14% (81% same side as face, 19% opposite side)</td>
</tr>
<tr>
<td><strong>Leg</strong></td>
<td>10% (75% same side as face, 25% opposite side)</td>
</tr>
<tr>
<td><strong>Any of Arm, Trunk or Leg</strong></td>
<td>19%</td>
</tr>
</tbody>
</table>

Involvement of the arm, trunk or legs is unusual, affecting around 1 in 5 of people. When this does occur it is usually on the same side as the face but sometimes on the opposite side. Some people reported just small patches of atrophy on their body. These would normally be called morphoea. I did not collect very good data on hair loss or pigmentation but many of you reported patches of hair loss (alopecia) and areas where the skin colour had become dark or paler than the surrounding skin.

**How much does Parry-Romberg overlap with coup de sabre?**

One of the questions I was keen to try and answer with the study was to what degree Parry-Romberg's Syndrome overlaps with the conditions Scleroderma "en coup de sabre"?

As you can see from the table 61% of those people who told me that they had been diagnosed with "coup de sabre" or linear scleroderma *also have involvement of the lower face*. It does seem, therefore, that the two conditions are so overlapping that they almost certainly represent the same underlying problem.
Associated Problems

Epilepsy

11% of people reported a history of epilepsy to me.

As a neurologist I was particularly interested in how common epilepsy is in this condition.

Since epilepsy affects between 2 and 5% of the population some of these cases may just be coincidental to Parry-Romberg Syndrome. However, nine people reported seizures where one side of their body jerks and this is almost certainly related to Parry-Romberg. Two of these people had seizures affecting the same side of their body as their face and in four of them it affected the other side. Epilepsy started an average of 9 years after the onset of the facial symptoms but in many cases it was at the same time and I am not sure that this result is reliable. Generally speaking the epilepsy seems to have been easy to control or has spontaneously resolved.

Migraine

I asked you whether you had a migraine type headache and 52% of you said that you did.

This seems to be a big problem with Parry-Romberg and although it has been reported it is not made very much off in the books.

In 55% of you the migraine tends to be on one side of your head (90% of the time this is the the same side of your head as the Parry-Romberg).

Twelve of you had tingling or weakness down one side as part of your migraine. This does sometimes happen in migraine whatever the cause. In 50% of these people the tingling and numbness was down the same side of the face and in 15% it was on the opposite side.

Many of you reported that the migraines you had were quite difficult to control.

Some of you descriptions sounded as if you’d had headaches on and off for so long that you had developed a chronic tension type headache, which often happens with bad migraine.

Occasionally migraine in patients with Parry-Romberg can be associated with changes in pupil size, which can be persistent.

Facial Pain

46% of you reported facial pain, which like migraine seems to be very common in Parry-Romberg.

When you mentioned the side of the pain to me it was always on the same side as the atrophy.

You described a number of different kinds of pain ranging from mild, hardly noticeable, occasional sensations to more frequent, burning or electric shock type pain. Often this had become a more dull persistent ache on your face. Again, this is not something that seems to have had a lot of attention paid to it in the medical literature.

Visual Symptoms

46% of you reported some problem with the eye on the affected side.

Common symptoms included globe retraction (the eye sinking back into the face a bit), the eye apparently drooping down a bit or difficulty closing eyelids leaving the eye dry and vulnerable to infection. In addition 17% of you have had actual inflammation of the eye either as a result of the eye being dry or because it's become spontaneously inflamed as it sometime does in this condition.
Associated Problems (continued)

Unusually Cold Hands

31% of you said that you have unusually cold hands.

I asked this question because both of the patients I saw with Parry-Romberg had this symptom and it did not seem to be particularly recognised. Many of you seem to have this problem quite badly but perhaps it has not been linked to your Parry-Romberg. I think we should interpret this result with a bit of caution as if you asked a hundred people without Parry-Romberg whether they had cold hands many of them would probably say they did. All the same it does seem to be a common problem and I suspect that it may be related in many cases to Parry-Romberg.

Jaw Symptoms

35% of you reported problems with your jaw.

Common difficulties were with trouble opening or closing your jaw or experiencing a sensation of "lock jaw" where you couldn't move your jaw at all. Some of you simply reported jaw pain or clicking sensations in the jaw. One or two of you have had surgery on your jaw as well.

Depression and Anxiety

Many people told me after I first launched the survey how having Parry-Romberg had led them to feeling quite down or tired so I decided to study this more carefully with the second survey.

Questionnaires are not an ideal way of working out how people feel about their problems and they certainly do not give an understanding of the complexity of what it is like to have a condition like Parry-Romberg.

However, according to the questionnaire, 46% of you had significant anxiety and 28% scored as having highly significant anxiety. Levels of depression were much lower with only 10% of you scoring significantly for depression and only 5% of you scoring highly significantly for depression.

I hope this part of the survey will highlight to your doctors and others that while not everyone is affected in this way, for a large number of people Parry-Romberg does have an emotional impact and that it may be important to recognise and treat this in its own right.

Other Medical Conditions

Below is a table of other medical conditions I asked you about. Many are autoimmune conditions. Perhaps the most interesting thing about this table is the quite high incidence of thyroid problems (this is much higher than in the general population). Depression also seems a little higher than in the general population. For the other conditions, the numbers are so small that I don't think we can really draw any conclusions.

<table>
<thead>
<tr>
<th>Medical Condition</th>
<th>No. patients</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Lupus (SLE)</td>
<td>4</td>
<td>2%</td>
</tr>
<tr>
<td>Systemic Scleroderma</td>
<td>9</td>
<td>5%</td>
</tr>
<tr>
<td>Rheumatoid Arthritis</td>
<td>7</td>
<td>4%</td>
</tr>
<tr>
<td>Raynaud's syndrome</td>
<td>6</td>
<td>3%</td>
</tr>
<tr>
<td>Multiple Sclerosis</td>
<td>0</td>
<td>0%</td>
</tr>
<tr>
<td>Inflammatory Bowel Disease</td>
<td>9</td>
<td>5%</td>
</tr>
<tr>
<td>Thyroid problems</td>
<td>19</td>
<td>10%</td>
</tr>
<tr>
<td>Ankylosing Spondylitis</td>
<td>3</td>
<td>2%</td>
</tr>
<tr>
<td>Eye inflammation</td>
<td>28</td>
<td>14%</td>
</tr>
<tr>
<td>Asthma</td>
<td>28</td>
<td>14%</td>
</tr>
<tr>
<td>Chronic Fatigue Syndrome</td>
<td>16</td>
<td>8%</td>
</tr>
<tr>
<td>Depression</td>
<td>69</td>
<td>35%</td>
</tr>
<tr>
<td>Vitiligo</td>
<td>33</td>
<td>17%</td>
</tr>
</tbody>
</table>
Blood Groups

I asked you about blood groups. For those of you that know your blood group, 44% of you have an 'A' blood group, 35% a 'O' blood group and 17% a 'B' blood group, 2% had 'AB'. This is more or less the same as what you would expect and therefore there doesn't seem to be any particular relations between blood groups and Parry-Romberg Syndrome. Rhesus status was also the same as the general population.

What Causes Parry-Romberg Syndrome?

I would not pretend that this survey can give us an answer to this question but I was interested to look at the few questions to see if we could any further.

Is it hereditary or genetic? (Family history of Parry-Romberg or facial hemiatrophy)

Seven of you out of 205 (3%) said there was someone in your family who had a problem with asymmetry of their face. I have to say that none of them have been definitely diagnosed with Parry-Romberg Syndrome, and it's important to recognise that there are other causes for an apparently asymmetrical face. Many people have slightly asymmetrical faces anyway so a little bit of asymmetry is not necessarily abnormal. Also other conditions such as Bell's Palsy may produce one side of the face that is weak, which may look similar to hemiatrophy. Because this is a question that many of you are interested in I have put in the table below the responses from the people who said that they did have someone in their family with a similar problem (with identifying details removed).

| 1. | "My grandmother. She has never been diagnosed but here problem is the same as mine. She has two small lips on one side and an indentation or line in the chin. Mine is more noticeable whereas with hers you have to really look at it to see it." |
| 2. | "My daughter is displaying symptoms of hemifacial atrophy and the asymmetry has been confirmed by our doctor. The doctor said that she has orbital asymmetry. As I said, more atrophy is obvious in a reflection. The doctor said that sometimes children do have asymmetry and that it can balance out later, possibly in her teens. He is not totally sure that it is Parry-Romberg's." |
| 3. | "Maybe one aunt, but she died young and she never had been diagnosed. Every body just always used to ask her who hit her on the face when she was a child because of the asymmetry on her face." |
| 4. | "But my grandmother who I take after has a smaller left side as well. My father's face is slightly smaller in the mirror. My grandmother's face was noticeable when she got older. She had never suffered Bell's Palsy or a stroke." |
| 5. | "My mother and grandfather both were born with mild facial asymmetry that did not progress through their lifetimes. They also both had the patches of white hair and my mother had vitiligo of the skin which got worse (concentrated on her face and hands) during her life. My mother also had eyes (the actual eyeballs) that did not match. She told me that once in the 1960's some doctor told her she had some kind of rare syndrome, but she never remembered what it was called and it never caused any problems and nothing changed in her condition over her lifetime, with the exception of the vitiligo." (I think this lady might be describing a condition known as hemifacial microsomia which is known to be more genetic and somewhat different to Parry-Romberg - JS) |
| 6. | 'My husband remembers an uncle that had 'something wrong with his face', it may have been muscular dystrophy. His older brother, age 19, also has some deformity in his face. He may need something to help bring his twisted jaws into some type of alignment. He cannot close his lips at all, his teeth are always showing.' |
| 7. | I'm unsure whether my 11 year old daughter may have it |

I think we can conclude from these results that there remains a possibility that in a very small minority of cases there is a risk of someone in the family also developing a similar problem but this seems to be at the very most only one in thirty and is probably less than this since the vast majority of you did not report anyone in their family with a similar problem.

How often is there a family history of autoimmune problems or epilepsy?

I asked whether there was anyone in the family with any medical condition that you thought was relevant. 31 of you mentioned medical problems but no pattern emerged of any condition that was more common than I would have expected.

Only 8% of you have someone in the family with a history of epilepsy. Often this was a quite distant relative and I don't think there are any connections here.
Parry-Romberg Syndrome - An global survey of 137 people using the Internet

Triggers

I asked you whether you thought anything had triggered the onset of your condition and allowed you to tell me about anything that you thought might be relevant...

Injuries

38% of you reported some sort of head injury as a child. The trouble with this question is that this may be about the same as the general population so may not mean anything.

However, 25 of you (12%) reported injuries that you felt might have been relevant. These included some striking stories of having stitches or an infection and the atrophy starting up afterwards in the same place. This either means that injuries are relevant to triggering it off in some cases or perhaps the Parry-Romberg was going to start anyway and it chose the 'point of least resistance' in the face. Here are some examples of the kind of injuries people reported.

• "There was a dog scratch right on the place it started"
• "Prior to the start of the disease my daughter collided with her brother causing a major bump and bruise on her forehead. Two weeks later she ran into the corner of a wall which also caused more bruising. It never went away..."
• "I got a volleyball spiked right in my face at close range"
• "I was hit in the forehead (right side) by a ball and the symptoms appeared as the bruising went away. The bruise lasted well over 6 months"
• "I fell off the dryer when I was 9 hitting my head very hard"
• "Just a month or so before I noticed the darkening on my face and neck, I had been hit extremely hard several times on the left side of my face. I always thought it was just coincidence."

Things that might Speed Parry-Romberg up

28% of you said that your Parry-Romberg had appeared to speed up at some point after it began.

Pregnancy or Childbirth?

Of those of you that answered yes to this question, 68% were woman who said that it got worse either during pregnancy or after childbirth. Some of you were a little vague about this but seven of you reported that it definitely got worse after birth and seven of you said that it got worse during pregnancy. Since this is only 25 of you, I guess this also means that a lot of you have been pregnant without noticing any change in your Parry-Romberg.

I left an open ended section for you record any other causes of your Parry-Romberg getting worse a third of you said that stress made it worse 8% thought that surgery or a bang on the head had speeded it up. 3% of you thought an infection or puberty had made it speed up.

Things that make Parry-Romberg Slow down or Stop

48% of you said that the condition had either slowed down or stopped completely (I suspect that this value was probably even higher but I probably asked the question in the wrong way).

It was difficult to get an idea for how long Parry-Romberg normally lasts. For many of you it seemed to start and stop several times throughout your life and many of you had developed the problem quite quickly and then it stopped quickly as well.

Investigations

There are no investigations that specifically need to be done in order to diagnose this condition, as it is a clinical condition diagnosed by the appearance of the face and soft tissues. Some of you have had a CT scan of your brain (62%), MRI Brain (44%) or EEG tracings (19%).
Treatments

Surgery

Surgery was by far the most common form of treatment that people had used with 63% of you having some form of surgical treatment or procedure at some stage. Of course the outcome of surgery depends a lot on where and when you had it done and how bad your atrophy was at the time. However, I collected very basic data from you about how successful the operations had been and this is presented below.

Table 2. Surgical Treatments and ratings of success in 128 people with Parry-Romberg Syndrome

<table>
<thead>
<tr>
<th>Treatment</th>
<th>% Successful</th>
<th>Somewhat Successful</th>
<th>Unsuccessful</th>
</tr>
</thead>
<tbody>
<tr>
<td>Any surgical treatment</td>
<td>62%</td>
<td>-</td>
<td>-</td>
</tr>
<tr>
<td>Fat injections</td>
<td>39%</td>
<td>19%</td>
<td>53%</td>
</tr>
<tr>
<td>Flap / pedicle procedure</td>
<td>19%</td>
<td>24%</td>
<td>55%</td>
</tr>
<tr>
<td>Bone implant</td>
<td>11%</td>
<td>45%</td>
<td>41%</td>
</tr>
<tr>
<td>Silicone injections</td>
<td>8%</td>
<td>13%</td>
<td>38%</td>
</tr>
</tbody>
</table>

Other Operations

<table>
<thead>
<tr>
<th>Procedure</th>
<th>Number of people</th>
<th>Outcome</th>
</tr>
</thead>
<tbody>
<tr>
<td>AlloDerm injections</td>
<td>2</td>
<td>Both successful</td>
</tr>
<tr>
<td>Tissue Expander</td>
<td>2</td>
<td>Both unsuccessful</td>
</tr>
<tr>
<td>Laser surgery</td>
<td>1</td>
<td>Somewhat successful</td>
</tr>
<tr>
<td>Stylane injections</td>
<td>1</td>
<td>Successful</td>
</tr>
<tr>
<td>Goretex injection</td>
<td>1</td>
<td>Somewhat successful</td>
</tr>
<tr>
<td>Excision of Atrophy / coup de sabre</td>
<td>2</td>
<td>Successful and Somewhat Successful</td>
</tr>
</tbody>
</table>

There were a wide variety of experiences of surgery.

As you can see from the table, surgery can be successful. Some people were very happy with the procedure that they’d had - the main reason the fat injections were unsuccessful was that the fat was simple absorbed often quite quickly after the injection.

Occasionally as people had got older the fat injections had meant that that side of their face was now more swollen than the other side and they had had to have operations to remove fat.

It seems the doctors have tried a number of other different types of injections and procedures. Looking at these results there certainly doesn’t seem any reason to favour one type of procedure over another. I am not a plastic surgeon, therefore I think it is probably best to take the advice of the surgeon that you’ve seen. Perhaps the only other comment from this section is that two patients told me that they had tissue expander inserted into their face and on both occasions this had been very unsuccessful with infections.

Medical Treatments

A few of you have tried drug treatment to try to control the atrophy. Steroids (Cortisone or Prednisolone) (8%), Methotrexate (4%) Azathioprine (Imuran) (1.5%), Penicillamine (2.2%), Intravenous Immunoglobulin (0.7%)

There are reports of this working in isolated cases but this has not been systematically evaluated. One person who had taken Hydroxychloroquine (Plaquenil) seemed to think it had helped slow down the process and someone else had had a success with Methotrexate. All of these drugs are potentially hazardous as they suppress the immune system and should only be prescribed by a specialist. Since Parry-Romberg is a condition which can sometimes stop by itself, then it is important to remember that some of these successes may relate to coincidence. I would not use the results of this survey to guide you on drug treatment.
Conclusions

- Parry-Romberg disease is a rare disease but perhaps not as rare as some others.
- My guess from this survey is that it must affect at least two in a million people and probably more.
- It may affect woman more than men but alternatively this may just be because women are more likely to complete Internet surveys!
- It does not effect one side of the face more than the other and their condition overlaps so strongly with the condition "Coup de Sabre" they are probably the same condition.
- Migraine or facial pain is common, affecting up to half of people with Parry-Romberg Syndrome; therefore these symptoms are important for doctors to look for and treat.
- Epilepsy is rare affecting only 10% of people, but can be caused by the syndrome.
- Having unusually cold hands may be part of Parry-Romberg Syndrome.
- Anxiety is common and depression to a lesser extent in patients with the condition.
- There is no definite evidence for Parry-Romberg being a genetic disorder although we have found a few examples of people who have relatives with facial asymmetry. (3%).
- There are some striking examples where a bang on the head or stitches to the head has coincided with the onset of Parry-Romberg and it is hard to ignore this as a possible factor.
- There were also some striking reports of the condition getting worse either during pregnancy or after delivery. While this may have simply been coincidence this is something that merits further investigation.
- In addition, stress can make any sort of symptom feel worse but if Parry-Romberg is partly due to the immune system then it is possible that stress may have a direct effect on the disease process. None of this means that the people with Parry-Romberg should not get pregnant or should not lead stressful life's if they want to.
- A number of types of treatment have been tried for Parry-Romberg and there are examples of where these have been successful but the evidence is not overwhelming. There is a range of surgical procedures and again none of them stand out as being definitely the best.
- There may be a link between Parry-Romberg disease and thyroid problems and this is potentially a new finding.
Final Thoughts

How does this all related to what we already know about Parry-Romberg Syndrome? I hope the results of this survey are not a disappointment to you, Parry-Romberg has been a mystery to doctors ever since it was first described and we still really have very little idea about what causes it. Some interesting French work recently has shown that animals can develop similar appearance to people with Parry-Romberg if a part of their nerve supply to the face (the sympathetic nerve supply) to the face is interrupted. In all likelihood Parry-Romberg represents a chance combination of a number of factors working together which is why it is so unusual. The picture below illustrates this.

![Diagram of factors associated with Parry-Romberg Syndrome]

**Figure 1. One idea about why people may get Parry-Romberg Syndrome**

My main aim in carrying out this survey was to give someone who is newly diagnosed with Parry-Romberg Syndrome would have some idea how common the various symptoms were in their conditions. I hope this survey has helped a bit in doing that.

I would like to thank everyone again who took part in the survey. It has been a real pleasure for me to correspond with you all and learn more about this condition. I will be keeping the web site going for the time being but due to other commitments will not be able to work on Parry-Romberg at least for a little while. I have been carrying out this study in my spare time and I now have to focus on other projects!

I wish you all the best from Scotland.

Yours sincerely

Dr Jon Stone

Edinburgh, Scotland, UK . February 2002

*An Historical Postscript....*

This picture is from Egypt AD 70-120. In a recent study scientists in London showed, using the skull of the person concerned, that they probably had Parry-Romberg syndrome. Definitely the oldest case on record!

Neurology in ancient faces.