David Longpre, Study about the frequency of symptoms directly or indirectly related to Klippel-Feil syndrome. 2011

SUMMARY

Klippel-Feil syndrome is a rare and little known syndrome for which there is little research. We have documented the main present symptoms in 100 people who have been diagnosed with Klippel-Feil syndrome.

OBJECTIVE

To document the frequency of the symptoms linked directly or indirectly in Klippel-Feil syndrome.

REVIEW OF THE LITERATURE

Klippel-Feil syndrome is a rare (some researchers suggest a probability of 1 in 42,000.) and little known condition. There is not a lot of research about it.

A typical case of this syndrome, dating back 4500 years to 4000 years BC, has been identified by archaeologist Patrick Moniatis. (http://www.klippel-feil.info/histoire.html)

In 1912 Klippel and Feil first described this syndrome in cervical vertebral fusion, limited range of neck movement, short neck, low posterior hairline.
Various researchers have studied the major medical challenges associated with this syndrome and the frequency of various symptoms experienced by patients, but the main limitation of studies to date is the small sample sizes, varying from one person to several tens of people.

Over time, simultaneous presence of the symptoms described by Maurice Klippel and Andre Feil was not necessarily required to diagnose the Klippel-Feil syndrome. More symptoms associated directly or indirectly to the syndrome have been described by researchers. It would take too long to make an exhaustive description, but we should mention particularly hearing problems, cardio-respiratory, neurological, renal and urological problems.

METHODOLOGY

We invited 100 people diagnosed with Klippel-Feil, or their parents when patients were not major, to complete a questionnaire listing the main symptoms known to be present in the person's Syndrome

Klippel-Feil. The name of symptoms was performed using both the scientifically words and a respondent accessible vocabulary.

Among the main strengths of this method, there is the sample representativity. In our knowledge, this is the first study considering a hundred patients. Patients came from several places in the world, including France, Belgium, Switzerland, Italy, Canada, Spain, English Canada, the United States, Mexico, England, Australia and some African countries. Questionnaires they had to answer were written either in French or English.

The completeness of the symptoms mentioned in the questionnaire is also as a strength of the methodology used, although questions about the anomalies lung and dental could shed additional light.

Finally, the authenticity of the respondents also appears as a force, each of which provide contact information.

In return, by reading the results of this research, we must keep in mind certain methodological limitations. First, the results reflect only the patient's situation at some point. This is important because may be the presence of certain symptoms may be underestimated. For example, as is often the case among some patients with progressive type of the syndrome, a patient of twelve years old could have not yet developed renal disease or hearing, or do not know about these symptoms. Second, data were provided to the best knowledge of the patients or their parents, and have not been validated by doctors, although patients have had complete questionnaires in the light of symptoms they discussed with their doctor. Finally, this study does not compare the data with the symptoms presence in a general population. It's a safe bet that some infrequent symptoms could also be present in a similar proportion among people not Klippel-Feil syndrome diagnosed.

RESULTS

33% of these patients were male, 67% were female.
19% were born prematurely.

Symptoms present, the most frequent to least frequent:

- Cervical vertebral fusion (one or more): 92%
- Short neck: 82%
- Cervical limited range of motion: 82%
- Low posterior hairline: 65%
- Chronic pain: 64%
- Muscle spasms: 62%
- Elevation of one or both scapulae (Sprengel's deformity): 58%

• Scoliosis: 57%
• Deafness: 49%
• neck bent to one side: 46%
• Sleep trouble: 44%
• Important abnormal and repetitive fatigue: 41%
• Paresthesia (tingling or numbness) in the arms or hands: 39%
• Facial asymmetry: 37%
• Limited jaw opening: 36%
• Vision problems: 35% (including 4% Duane syndrome)
• Gastrointestinal problems: 35%
• webbed neck: 33%
• kidney abnormality or deformity: 32%
• Chronic sinusitis or chronic nasal congestion: 31%
• Psychological irritability : 30%
• Hemivertebrae (one or more): 28%
• Misaligned jaws: 27%
• Thoracic vertebral fusion: 24%
• cardiac or vascular trouble: 22%
• cervical ribs: 22%
• Missing or fused ribs: 21%
• Mirror movements: 20%
• Eyes convergence problem (strabismus): 20%
• Drooping eyelids (ptosis): 16%
• Spina bifida: 15%
• Hypothyroidism or Hyperthyroidism: 14%
• Sphincter trouble: 14% (difficulty opening, sphincter incontinence or lack of)
• Hyperflexion thumb: 13%
• Abnormality of the fingers (missing or too many fingers): 12%
• Lumbar vertebral fusion (one or more): 11%
• Urethra or ureter (malformation or stenosis): 10%
• Lordosis: 9%
• Cleft palate or lip: 9%
• Abnormalities of the reproductive system / female genital (absence or malformation of the vagina, biscornu uterus, malformation or hypoplasia of the vagina, absence of ovaries, uterus or the presence of inverted two complete reproductive systems): 8%
• male reproductive system abnormalities or abnormalities of the scrotum: 8%
.Arnold-Chiari malformation • 8%
• Intellectual Disability: 7%
• Delay School: 6%

Symptoms mentioned by the patient without it being asked in the questionnaire:
• dental malformations: 4
• Meningocele or syringomyelia: 4
• Lack of "sensitivity" to a body part: 3
• Static or dynamic dizziness: 3

• Pulmonary hypoplasia: 3
• Multiple discal protuberances: 3
• Limited range of arm movements: 3
• Craniostenosis: 2
• Paralyzed vocal cord: 1
• Cerebral palsy: 1
• Facial palsy: 1
• Frequent Hiccups
• Impaired concentration, depression, anxiety or other psychological illness or cognitive
• Fused liver and stomach

CONCLUSION

The symptoms described by the first researchers are frequently represented in a population of 100 persons diagnosed with Klippel-Feil. (fusion of one or more cervical vertebrae 92%, 82% short neck, limited neck range of motion 82%, low posterior hairline 65%).

The presence of instability between the fused segments of the spine as well as limited cervical range of motion seems common cause of chronic pain (64%), muscle tension or spasm (62%). (Sprengel's deformity) is also frequently encountered (58%) as well as scoliosis (57%)
Deafness (49%), neck bent to one side (46%), sleep disturbances (44%), severe and abnormal repetitive fatigue (41%) as well as paresthesia of the arms or hands (39%) seem to affect a relatively large percentage of the sample being studied.

Various other symptoms, although less frequent in this population studied, were identified several times by different authors as sometimes present along with Klippel-Feil syndrome. In particular, facial asymmetry (37%), a limited opening of the jaw (36%), vision problems (35% including 4% Duane syndrome), gastrointestinal problems (35%), webbed neck (33 %), hypoplasia or malformation of the kidneys (32%), hemivertebra (28%), thoracic cervical fusion (24%), cervical ribs (22%), the absence or fusion of ribs (21%), mirror movements (20%), ptosis of the eye (16%), spina bifida (15%), sphincter disturbances (14%), abnormal fingers (12%), urethra or ureter stenosis or hypolasia (10%), cleft palate (9%), abnormalities of the reproductive system (8%) and the Arnold-Chiari malformation (8%).

This research documents the higher presence of certain symptoms in relation to previous research. In particular, the facial asymmetry, described in the past as present in 20% of people diagnosed with Klippel-Feil, affects 37% of our study population. Sprengel's deformity affects 58% of our study population compared to 35% in previous studies. Heart and vascular problems affect 22% of our population vs 10% in the previous data. Genitourinary abnormalities and kidney

damage, including malformations of the urethra or the ureter, are comparable with previous data (50% vs. 40-65%). Deafness is also comparable (49% vs. 30-50%), as well as mirror movements (20% vs 20%).

David Longpre, moderator
For Regroupement francophone international Klippel-Feil
http://www.klippel-feil.info
In collaboration with O-Circle-O Klippel-Feil Syndrome