



Physical & Mental Health

Content was combined and adapted from original articles with help from numerous professionals within the FXCRC, including Brenda Finucane, Dr. Rebecca Shaffer, Jayne Dixon-Weber, and members of the NFXF team.

The effects of Fragile X syndrome are most noticeable in the cognitive behavioral domain (cognitive deficits, behavioral issues, and autism spectrum disorder). Multiple associated physical problems, mostly related to loose connective tissue, can also occur. These include hypotonia, hyperflexibility, flat feet, heart murmur, seizures, early puberty, and recurrent ear infections.

There is currently no definitive specific treatment for these physical problems. Management typically involves the combined efforts of a multidisciplinary team including ideally, psychology, developmental and behavioral pediatrics, neurology, speech and occupational therapy, neurology, psychiatry and genetics. Early diagnosis and early intervention is key to managing physical and medical concerns in FXS.

Individuals with FXS may have co-occurring diagnoses that impact physical and mental well-being, like ADHD or anxiety, for example. These can be symptoms of FXS, and they can also be their own diagnosis; therefore, it can be challenging to

understand the root cause. Learn more about this from Brenda Finucane, a longtime genetic counselor and Fragile X champion: [Making Sense of Multiple Diagnoses](#).

Because of the complexity of FXS, a multidisciplinary team is always encouraged to support your loved one with FXS. Curious about what that looks like? Learn more by watching our [Multidisciplinary Treatment of Fragile X Syndrome](#) webinar with Drs. Craig Erickson, Laura Hess, Kerrie Chitwood, and Rebecca Shaffer.

Common Medical Issues: Birth to Adulthood

Infants & Toddlers

Most infants with [Fragile x syndrome](#) are healthy and don't require extensive medical intervention. There are some common medical conditions that can occur more often in babies with Fragile X syndrome.

Birth Defects

Babies with FXS have an increased chance of some congenital abnormalities such as cleft lip, cleft palate, clubfoot, congenital hip dislocation, and hernias. This may be related to loose connective tissue (more about this below).

Because these are all common birth defects among children without FXS, babies with these conditions are not routinely tested for FXS unless they present other symptoms, such as developmental delay, or a [family history of Fragile x syndrome](#).

Low Muscle Tone

Many babies with FXS have hypotonia, which is low muscle tone. This is sometimes referred to as being "floppy," which for a baby can cause difficulty in holding up their head, also known as "head lag."

Because low muscle tone affects gross motor skills, such as sitting up or rolling over, hypotonia can contribute to the developmental delays often seen in babies and toddlers with FXS.

Digestive Disorders

Although many babies with FXS do well in the newborn period, others have difficulty with feeding, vomiting, or gastroesophageal reflux disease (known as GERD), a digestive disease in which stomach acid or bile irritates the lining of the esophagus (the tube connecting the mouth to the stomach).

This may be related to connective tissue abnormalities, low muscle tone (hypotonia), dysfunction of the gastroesophageal sphincter, or a hypersensitive gag reflex.

Some parents report that vomiting in babies with GERD can be resolved by positioning them upright during feeding and after meals. Occasionally, medication is needed to decrease reflux.

Babies will sometimes be diagnosed with what is called “failure to thrive.” This is typically related to difficulties in sucking, GERD, or an aversion to food textures. If feeding difficulties are a problem, particularly sucking or intolerance of certain food textures, consultation with an occupational or speech and language therapist, or therapies such as oral desensitization, oral stimulation, and oral motor coordination, can be helpful.

Ear & Sinus Infections

Beginning in the first year of life, frequent otitis media (middle ear infections) are a problem for many boys (and some girls) with FXS. In fact, the speech and language delays seen in children with FXS are often mistakenly attributed to chronic ear infections, sometimes delaying the [Fragile X syndrome diagnosis](#).

Ear infections require treatment to avoid hearing loss that could further compromise language development. If a conductive hearing loss persists after acute antibiotic treatment, ear tubes (most commonly referred to as PE — pressure equalizing — tubes, but also known as tympanostomy or ventilating tubes) are often recommended.

Recurrent sinusitis (also referred to as sinus infection) is also a common problem in FXS, which may be related to the facial structure or the connective tissue problems that lead to recurrent ear infections.

Children and Adolescents

Urinary Tract Infections

Some children (more often boys), with FXS, may have an increased susceptibility to urinary tract infections and reflux (the backing up of urine into the urethra and bladder). The pediatrician may suspect an infection if the child has a fever of unknown origin with no other explanation, and thus a urine sample will be obtained.

Seizures

Approximately 20% of males and a smaller percentage of females with FXS have epilepsy. If your physician suspects your child is having [seizures](#), often an EEG (electroencephalogram, which detects electrical activity in the brain) is performed. If seizures are confirmed, seizure medication is usually prescribed.

[The largest and most definitive study yet published on seizures in FXS](#) was completed using the Fragile X Online Registry with Accessible Research Database (FORWARD). A short summary of the study:

- **Frequency:** In this study, the overall chance of having at least one seizure was 12% overall in Fragile X syndrome, 13.7% in males, and 6.2% in females.
- **Age of Onset and Resolution:** In the group with seizures, the average age of the first seizure was 6.4 years of age with the great majority — 86.7% of males and 81.8% of females — having the first seizure before age 10. The age of the last seizure followed a similar age dependence to the age of the first seizure, with 70.9% of seizures in males and 63.6% of seizures in females resolving by age 10.
- **Types:** Partial (focal) seizures were reported in 25% and generalized seizures in 31% of those with seizures, with febrile seizures in 8% and the remainder of seizures being of unknown type. Males and females did not show a different distribution of seizure types.
- **Association of Seizures with Other Fragile X Syndrome Characteristics:** As compared to individuals with FXS without seizures in FORWARD, those with seizures were more likely to have more severe intellectual disability, current sleep apnea, delayed acquisition of expressive language, autism spectrum disorder (ASD), hyperactivity, irritability, and stereotyped movements.
- **Treatment and Medications:** Treatment and management of seizures in

Fragile X syndrome is similar to seizure treatment in other conditions associated with seizures. There is no Fragile X syndrome-specific medication or approach to treating seizures. A person with seizures is usually treated with medications, known as anticonvulsants, after two (or sometimes more if there is a long time between seizures) seizures.

Seizures are reported to be easily controlled in most cases and have been thought to resolve during childhood in the majority of individuals with FXS.

From our consensus-based treatment recommendations, a deeper look at seizures related to Fragile X syndrome:

Related Treatment Recommendations

Seizures in Fragile X Syndrome

Based on study data, 12% of people with Fragile X syndrome have seizures, and treatment and management are similar to seizure treatment in other conditions associated with seizures. They are easily controlled in Fragile X syndrome and most patients grow out of their seizures before their 20s.

[Go to the treatment recommendation](#)

Physical Problems in Fragile X Syndrome

Multiple associated physical problems can occur with FXS, mostly related to loose connective tissue.

[Go to the treatment recommendation](#)

Eye Disorders

Children with Fragile X syndrome have an increased susceptibility for vision and eye disorders, which are common in childhood.

These include:

- **Strabismus:** Also called “lazy eye,” this condition causes the eye to appear “crossed” or drifting to the middle. This is often due to low muscle tone in the

eye, a symptom of the hypotonia present in many babies or children with Fragile X syndrome.

- **Ptosis:** Drooping of the eyelid, again often related to low muscle tone.
- **Nystagmus:** Shaking of the iris in a back and forth motion.
- **Myopia/Hyperopia:** Subtle near- or farsightedness.

It is recommended that all children with Fragile X syndrome be referred to an ophthalmologist if the pediatrician suspects any of these common eye or vision conditions.

Connective Tissue Issues

Connective tissue is the tissue that holds together the body such as skin, muscles, tendons, cartilage, ligaments, bones, and blood vessels. Many individuals with Fragile X syndrome have differences in their connective tissue. Some of these connective tissue problems include:

- **Scoliosis:** Curvature of the spine, usually not severe.
- **Flat feet:** Also called pes planus.
- **Hernias:** Inguinal (groin) hernias.
- **Cardiac murmur:** Often a functional or innocent murmur, it is sometimes indicative of mitral valve prolapse (or MVP, which is improper closure of the valve between the heart's upper and lower left chambers.). Occasionally an evaluation by a cardiologist is warranted to confirm MVP, and if confirmed, the cardiologist may recommend antibiotics prior to dental procedures or surgery.
- **Hyperflexible joints:** Particularly of the wrists, fingers, and elbows.
- **Soft skin**
- **Macroorchidism:** Testicular volume normally increases during the early stages of puberty, but in boys with Fragile X syndrome this increase is usually quite dramatic, leading to macroorchidism (enlarged testicles). These changes are typical for Fragile X syndrome, and do not require intervention. Some boys with Fragile X syndrome develop large testicles prior to the onset of puberty.

Adults

Sometimes what are seen as behavioral or emotional challenges in individuals with FXS may have an underlying medical cause; this is one of the reasons why having a primary care physician is so helpful. It can be helpful to seek an opinion from a doctor to exclude a medical cause of any changes in behavior, especially if it

persists.

You can find more information and a list of medical problems associated with Fragile X in the NFXF treatment recommendation, [Transition to Adult Services for Individuals with Fragile X Syndrome](#):

Related Treatment Recommendation

Transition to Adult Services for Individuals with Fragile X Syndrome

There are many new issues to address as an individual with FXS becomes an adult. In general, it is best to search for information on adult issues and services in the state where the adult resides. Services can vary widely from state to state and even vary within a state, so it is up to the parents or providers to find what is available and to set up the daily schedule for or with the person with FXS.

[Go to the treatment recommendation](#)

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Mental Health Throughout the Lifespan

Mental health contributes to our overall wellbeing. Mental health can impact our thoughts, feelings and actions. This then impacts how we interact with the world and others, make decisions, and handle stressors. Emotional, psychological, and social wellbeing are all parts of mental health.

Challenges with mental health are most prominent in adolescents and adults with FXS. It is important to seek out professional help and support for any mental health concerns. Learning strategies to support emotion regulation can be incredibly helpful.

Learning more about behaviors in FXS may also help. Visit our FXS topic page [Managing Behaviors](#) plus two webinars presented below.

Mental Health Webinars

Multidisciplinary Treatment of FXS

Emotional Regulation in Fragile X Syndrome

[See all Mental Health webinars →](#)

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Find a Fragile X Clinic

Fragile X clinics provide medical services -including medication evaluations and consultations- supervised by a physician and supported by the latest medical, educational, and research knowledge available. Multidisciplinary services, such as genetic counseling and occupational, speech, language, and behavioral therapies, are also available either at the clinic or by referral.

- [Find a U.S. Fragile X Clinic](#)
- [Find Clinics Specializing in the Treatment of FXTAS](#)
- [Find an International Clinic](#)

Preparing for an In-Person Clinic Visit

Preparing for an in-person visit to a Fragile X clinic can set up you and your loved one with FXS for success. The clinic can help you prepare too, so be sure to ask about any materials they have on hand about what to expect. Don't forget to bring comfort items and activities for you and your loved one; headphones can be especially helpful in waiting rooms! Don't be afraid to ask for accommodations if you need them. And don't be afraid to ask any and all questions you have — the clinics have heard it all and are there to help.



Your In-Person Visit to a Fragile X Clinic

The Fragile X Clinical & Research Consortium (FXCRC)

The Fragile X Clinical and Research Consortium (FXCRC) was created by the National Fragile X Foundation in response to the growing needs of families whose members have one of the Fragile X-associated conditions. The **FXCRC is composed of over 30 clinics**,¹ each of which has expertise in Fragile X.

Though all clinics serve individuals with FXS, many also serve those or have referrals for those with FXTAS, FXPOI, and other Fragile X premutation-associated health needs. Though each clinic operates independently, they often collaborate with one another, sharing resources, participating in research projects, and attending meetings of the FXCRC. New clinics are being added regularly across the U.S., and **around the world!**²

Where to Begin

We encourage families to visit the clinic closest to them, but, in some cases, visiting a different clinic might be advisable based on the expertise of a specific healthcare provider.

Once you **locate a clinic**, contact the clinic coordinator to discuss your concerns about your child or family member and what you hope to get out of the visit. Ask about the following:

- **What has to be done before the visit?** Paperwork. See "Information Needed" on the next page.
- **What does a typical visit entail?** Some clinics conduct evaluations over two days, which requires an overnight visit. Others may require only several hours.
- **In addition to seeing the clinic director, what evaluations are offered and by whom?** Examples might include speech, occupational, physical, and behavioral therapies, and genetic counseling.
- **What other specialists are available?** For example: psychologists, audiologists, neurologists, cardiologists, and ophthalmologists.
- **Are referrals made to local providers in your area?**
- **After the evaluation, will the clinic director be available to review and discuss the results?** Ask if each specialist will be available, or if the clinic director reviews the findings of each specialist with you.
- **When will a written evaluation be available?** Ask if there will be separate reports from each clinic specialist or one report, and if you can request the report be sent to your providers, such as teachers and therapists.
- **Who will be available to interact with your local providers?** You'll want to make sure treatment recommendations are clarified, discussed, and implemented.
- **Do they have a visual schedule of the visit you can access?**

¹ <https://fragilex.org/resources/clinics/>

² <https://fragilex.org/resources/int-clinics/>

Your In-Person Visit to a Fragile X Clinic

3 pages,

Updated 04/16/2025

A quick guide to visiting a Fragile X clinic, including how to begin the process, questions to ask, what you'll need to share with the clinic, and what to expect.

[Download the PDF](#)

SERIES DE INFORMACIÓN SOBRE EL X FRÁGIL: 

SU VISITA A UNA CLÍNICA DE X FRÁGIL: UNA VISITA EN PERSONA

Visión general

El cromosoma X frágil es un grupo de trastornos asociados con cambios en el gen X frágil (llamado FMR1) y situado en el cromosoma X. El gen FMR1 puede sufrir cambios, cuando se hereda, lo que afecta un patrón del ADN llamado repeticiones de CGG. Normalmente, el gen FMR1 tiene hasta 34 repeticiones de CGG, aunque el intervalo entre 40-54 repeticiones se denomina zona intermedia o gris. Un portador de premutación tiene de 55 a 200 repeticiones de CGG y alguien con una mutación completa tiene más de 200 repeticiones de CGG.

Cuando se presenta una premutación o una mutación completa, puede resultar en un Trastorno asociado con el cromosoma X frágil (Fragile X-associated Disorder, FXD). Los trastornos incluyen:

- Síndrome del cromosoma X frágil (DXF): Un trastorno hereditario que afecta el desarrollo intelectual, conductual y social. Ocurre tanto en hombres como en mujeres que tienen una mutación completa del gen FMR1.
- Síndrome de temblor/ataxia asociado al cromosoma X frágil (FXTAS): Una afección neurológica que inicia en la edad adulta (mayores de 50 años), más común y más grave en algunos hombres portadores de premutación. Puede causar temblores, problemas de memoria y de equilibrio.
- Insuficiencia ovárica primaria asociada al cromosoma X frágil (FXPO): Una afección que afecta la función ovárica que puede provocar infertilidad y menopausia precoz en algunas mujeres portadoras de premutación.
- Otros problemas pueden estar presentes en los portadores de premutación y los investigadores continúan estudiando las posibles asociaciones con otros y otros problemas de salud en personas con premutaciones del cromosoma X frágil.

El Consorcio Clínico y de Investigación del Cromosoma X Frágil (FXCRC)

El Consorcio Clínico y de Investigación del Cromosoma X Frágil (FXCRC, por sus siglas en inglés) fue creado por la Fundación Nacional del Cromosoma X Frágil (NFXF, por sus siglas en inglés) en respuesta a las crecientes necesidades de las familias cuyos miembros tienen una de los Trastornos asociados con el Cromosoma X Frágil (FXD, por sus siglas en inglés). El FXCRC está compuesto por más de 30 clínicas, cada una de las cuales tiene experiencia en el cromosoma X frágil.

Aunque todas las clínicas atienden a personas con síndrome del temblor/ataxia asociado al cromosoma X frágil (FXTAS), insuficiencia ovárica primaria asociada al cromosoma X frágil (FXPO) y otras afecciones portadoras de premutación.

Aunque cada clínica opera de forma independiente, a menudo colaboran entre sí, compartiendo recursos, participando en proyectos de investigación y asistiendo a reuniones del consorcio. Se están agregando nuevas clínicas regularmente en los Estados Unidos y en todo el mundo. Para obtener un mapa actualizado regularmente e información clínica, consulte: <https://fragilex.org/clinica/>

Por dónde empezar

Comience por llamar al coordinador de la clínica para discutir sus inquietudes sobre su hijo o familiar y lo que espera obtener de la visita. Pregunte sobre lo siguiente:

- ¿Qué hay que hacer antes de la visita? (Documentación, etc.). Consulte la información necesaria a continuación.
- ¿Qué implica una visita típica? Algunas clínicas realizan evaluaciones durante dos días, lo que requiere pasar la noche en la clínica. Otras visitas pueden requerir solo varias horas.
- Además de ver al director de la clínica, ¿qué evaluaciones se ofrecen y por quién? Los ejemplos pueden incluir al habla, terapia ocupacional, física y conductual, y asesoramiento genético.
- ¿Qué otros especialistas están disponibles? Por ejemplo: psicólogos, audiólogos, neurólogos, cardiólogos, oftalmólogos, etc.
- ¿Se hacen remisiones a proveedores locales en su área?
- Después de la evaluación, ¿está disponible el director de la clínica para revisar y discutir los resultados? ¿Está disponible cada especialista, o el director de la clínica revisa los hallazgos de cada especialista con usted?
- ¿Cuándo estará disponible una evaluación por escrito? ¿Consistirá de informes separados de cada especialista de la clínica o de un solo informe? (Puede solicitar que el informe se envíe a sus proveedores, maestros, terapeutas, etc.)
- ¿Quién estará disponible para interactuar con sus proveedores locales para asegurarse de que las recomendaciones de tratamiento se entiendan, discutan e implementen?
- ¿Tienen un horario visual de la visita a la que puede acceder?

Pregunte también sobre

- ¿Qué materiales escritos o en línea están disponibles? (Una lista de recursos estatales y locales)? ¿Folletos o artículos sobre FXD para que los lleve de regreso al personal de la escuela, a los médicos y a otros proveedores?
- ¿Hay la posibilidad de participar en una investigación?
- ¿Qué tipo de seguro acepta la clínica? Además, consulte con su seguro para ver cuáles serán sus responsabilidades, requisitos financieros y de otro tipo. (Si no tiene seguro, pregunte si hay planes de pago alternativos y / o oportunidades para participar en ensayos de investigación).
- ¿Tienen recomendaciones de alojamiento para satisfacer sus necesidades con respecto al precio, la proximidad a la clínica, las opciones de comida, etc.? ¿Alguno de ellos ofrece descuentos para quienes visitan el hospital?

Su Visita A Una Clínica De X Frágil: Una Visita En Persona

2 pages

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Preparing for a Telehealth Visit

Many NFXF Fragile X Clinical & Research Consortium clinics offer telehealth visits in addition to in-person. Note there are specific parameters to these visits, and they vary from clinic to clinic and state to state. We have gathered information to help you make the most of your online telehealth visit to a Fragile X clinic.

Learn more about [Telehealth Visits: Suggestions for Parents on How to Prepare](#)

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Medications

Medications are at times helpful to facilitate the individual's ability to attain optimal life skills and allow for better integration into educational, adult, and social environments.

Our treatment recommendation and consensus of the Fragile X Clinical & Research Consortium provide the most current guidance.

Medications for Individuals with Fragile X Syndrome

Medications are, at times, helpful to facilitate the individual's ability to attain optimal life skills and allow for better integration into educational, adult, and social environments.

[Go to the treatment recommendation](#)

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Non-Medication Treatments & Therapeutic Interventions

Non-medication treatments and therapeutic interventions may be used in addition to medications or on their own. When a medication is recommended to help address symptoms or specific behaviors, it may be recommended in combination with other therapeutic interventions. Examples of these interventions are behavioral, cognitive, developmental, vocational, and educational therapies.

Complementary and Alternative Therapies

Integrative Health Recommendations for the Treatment of Fragile X Syndrome

Integrative health emphasizes a broader communication between healthcare providers and patients to determine the best combination of traditional and non-traditional therapies.

[Go to the treatment recommendation](#)

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Additional Resources

Health Watch Table – Fragile X Syndrome

| CONSIDERATIONS | RECOMMENDATIONS |
|---|--|
| 1. HEENT (HEAD, EYES, EARS, NOSE, THROAT) | |
| Children: Vision, strabismus, refractive errors are common Hearing: recurrent otitis media is common Nose: sinusitis is common Adults: strabismus and refractive errors are common | <ul style="list-style-type: none"> ☐ Undertake newborn vision and hearing screening and an auditory brainstem response (ABR). ☐ Refer for a comprehensive ophthalmologic examination by 4 years of age. ☐ Visualize tympanic membranes at each visit. ☐ Undertake hearing and vision screening at each visit with particular attention to myopia and hearing loss. |
| 2. DENTAL | |
| Children and Adults: High arched palate and dental malocclusion are common | ☐ Refer to a dentist for an annual exam. |
| 3. CARDIOVASCULAR | |
| Children: Mitral Valve Prolapse (MVP) is less common in children (~10%), but may develop during adolescence Adults: MVP is common (~30%). Aortic root dilation usually is not progressive Hypertension is common and exacerbated by anxiety | <ul style="list-style-type: none"> ☐ Appropriate for running or other at each visit. If present, do an ECG and echocardiogram; refer to cardiologist, if indicated. ☐ Undertake an annual clinical exam. Based on findings, obtain an ECG and echocardiogram. Refer to cardiologist, as appropriate. ☐ Measure BP at each visit and at least annually. ☐ Treat hypertension when present. |
| 4. RESPIRATORY | |
| Children & Adults: Obstructive sleep apnea (OSA) may be due to enlarged adenoids, hypotonia or connective tissue dysplasia | <ul style="list-style-type: none"> ☐ Ascertain a sleep history and assess for evidence of OSA. ☐ Obtain a sleep study as appropriate. |
| 5. GASTROINTESTINAL | |
| Children: In infants, feeding problems are common with recurrent emesis associated with Gastroesophageal Reflux Disease (GERD) in ~30% of infants | ☐ Refer for assessment of GERD. Thickened feeds and upright positioning may be sufficient to manage GERD. |
| 6. GENITOURINARY | |
| Children and Adults: Inguinal hernias are relatively common in males Macroorchidism generally develops in late childhood and early adolescence and persists Urinary reflux may persist into adulthood | <ul style="list-style-type: none"> ☐ Assess for inguinal hernia annually beginning at age 1 year. ☐ Macroorchidism can be measured with an orchidometer; reassure parents and patients that it does not require treatment. ☐ Evaluate recurring urinary tract infections (UTI) with cystourethrogram and renal ultrasound. Refer to a nephrologist or urologist as needed. ☐ Consider and assess for a renal etiology, such as scarring, as the basis for persistent hypertension. |
| 7. SEXUAL FUNCTION | |
| Adults: Males and females are fertile | ☐ Consider discussion of recurrence risk and reproductive options as a basis for referral to a geneticist. Make such a referral even if fragile X is only suspected so that molecular testing can be undertaken if the person concerned and relevant family members. |

1

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Health Watch Table for Fragile X Syndrome

4 pages,

Updated 10/01/2012

This is also found in our [Physical Problems in Fragile X Syndrome](#) treatment recommendation, a consensus of the Fragile X Clinical & Research Consortium.

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Single Dose Medication Study Results in Fragile X Syndrome



Craig A. Erickson, MD

Professor of Psychiatry

Director of Research, Division of Child & Adolescent Psychiatry

Director, Fragile X Research & Treatment Center

Cincinnati Children's Hospital Medical Center

University of Cincinnati College of Medicine-Affiliated



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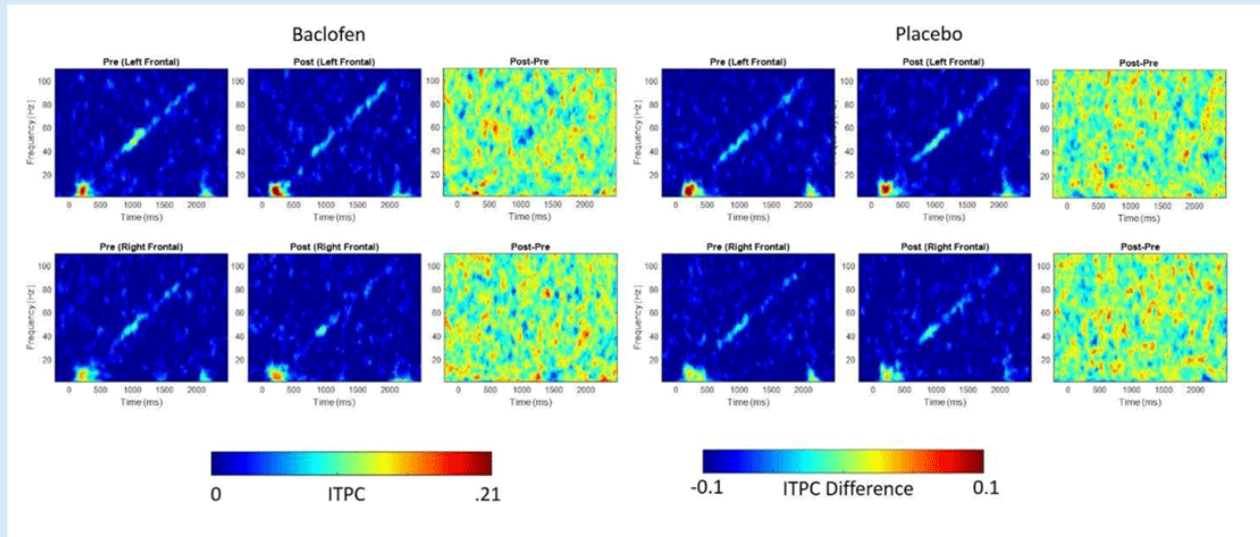
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