**[LMN Template for Chromosome Analysis/ Karyotype]**

\*applicable for suspected diagnoses of ring 14/17/20, 8p inv dup del, and others

Patient: \*\*\*

Insurance Company: \*\*\*

Subscriber Name: \*\*\*

Policy #:\*\*\*

Reference Number: \*\*\*

Dear Claims Specialist,

I am writing this letter of medical necessity on behalf of my patient [Patient Name] to request coverage for high resolution chromosome analysis (karyotype) offered through [Lab Name], a high complexity CLIA certified laboratory located in [Lab Location].

**Information on Patient’s Condition:**

[Patient Name] is a [age][sex] with [symptoms, family history information]. [Patient Name]’s clinical symptoms and results of the routine diagnostic tests suggest a structural chromosome abnormality [insert specific condition suspected if applicable]. Next-generation sequencing, whole exome sequencing, and chromosomal microarray are unable to reliably detect structural chromosome abnormalities including balanced translocations, ring chromosomes, inversions, and other complicated rearrangements. The only way to confirm a specific diagnosis is to perform genetic testing on this patient. Results from this genetic test may have a direct impact on this patient’s treatment and management.

**This [patient/family] has had pre-test counseling regarding the potential outcomes of this test, and a certified genetic counselor will be involved in discussion of any abnormal results.** [Remove if inapplicable]

**Information on Chromosome Analysis:**

Chromosome abnormalities are a common cause of genetic disease, the leading known cause of intellectual disability, and occur in approximately 1/150 live births. There are two major classes of chromosome abnormalities: numerical abnormalities which include aneuploidy (loss or gain of whole chromosomes) and polyploidy (gain of 1 or more entire haploid sets of chromosomes); and structural abnormalities (translocations, inversions, deletions, duplications). Chromosomes are stained by G-banding and arranged in a karyotype for analysis of the number and structure of the individual chromosomes. Chromosome analysis is an excellent complement to whole genome or targeted array comparative genomic hybridization (aCGH) since it can detect balanced chromosome abnormalities not detectable by aCGH, and can sometimes clarify the mechanisms of chromosome rearrangements detected by the array.

While NGS, WES/WGS, and/or chromosomal microarray/aCGH can identify a genetic diagnosis for many individuals with epilepsy and other neurological disorders, certain patients with potentially recognizable phenotypes may be missed by these testing methodologies. Notably, most patients with a diagnosis of ring 20 syndrome, ring 14 syndrome, ring 17 syndrome, and certain chromosome 8p disorders, among others [add your suspected diagnosis if applicable] cannot be diagnosed by NGS methodologies and must receive a karyotype in order to accurately make these diagnoses. Epilepsy due to structural chromosome abnormalities is likely underdiagnosed due to underutilization of karyotype in current genetic testing paradigms. Karyotyping is recommended for patients with intractable epilepsy for whom genetic testing with NGS methodologies has been nondiagnostic.

**Clinical Utility of Chromosome Analysis:**

* **A specific diagnosis may influence the treatment for this patient.** For example, certain medications may be more effective based on the diagnosis and some may be contraindicated.
* **Proper diagnosis will help the physician in managing the patient more efficiently, while limiting doctor visits and insurance claims**
* **A specific genetic diagnosis will allow for family members to be tested for the specific mutation for a drastically lower cost**

**We request your review of the following testing for preauthorization:**

Test Requested:  \*\*\*

Test Code: \*\*\*

Lab Name: \*\*\*

Lab Phone: \*\*\*

CPT codes: \*\*\*

**ICD10 Codes:** \*\*\*

The blood draw would be completed at [Lab/Hospital] for the family’s convenience. Billing would be coordinated by [Billing Institution/Provider]. Ordering provider would be [Ordering Provider].

Thank you for your review and consideration. I hope you will support this request for genetic testing coverage for [Patient Name]. If you have questions, or if I can be of further assistance, please do not hesitate to contact me at [Clinician Contact Info].

Sincerely,

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**References:**

Thomas RH, Berkovic SF. The hidden genetics of epilepsy-a clinically important new paradigm. Nat Rev Neurol. 2014;10(5):283–92.

Sheidley BR, Malinowski J, Bergner AL, Bier L, Gloss DS, Mu W, et al. Genetic testing for the epilepsies: a systematic review. Epilepsia. 2022;63:375–87.

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James WD, Roth R, Fitzgerald M. Ring 20 syndrome: A call to action. *Epilepsia*. 2024;65(4):1147-1148. doi:10.1111/epi.17941

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